

### **User Guide**

# **AWS HealthOmics**



## **AWS HealthOmics: User Guide**

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## What is AWS HealthOmics?

AWS HealthOmics is an AWS service that helps users such as bioinformaticians, researchers, and scientists to store, query, analyze, and generate insights from genomics and other biological data. It simplifies and accelerates the process of storing and analyzing genomic information for research and clinical organizations, and makes scientific discovery and insight generation faster.

HealthOmics has three primary components. HealthOmics Storage helps you store and share petabytes of genomics data efficiently and at low cost per gigabase. HealthOmics Analytics simplifies how you prepare genomics data for multiomics and multimodal analyses. HealthOmics Workflows automatically provisions and scales the underlying infrastructure for your bioinformatics computation.

### **Topics**

- Important notice
- Features of AWS HealthOmics

## Important notice

AWS HealthOmics is not a substitute for professional medical advice, diagnosis, or treatment, and is not intended to cure, treat, mitigate, prevent, or diagnose any disease or health condition. You are responsible for instituting human review as part of any use of AWS HealthOmics, including in association with any third-party product intended to inform clinical decision-making.

AWS HealthOmics is intended only for the transferring, storing, formatting, or displaying of data, and for the provision of infrastructure and configuration support for managing workflows. AWS HealthOmics is not intended to directly perform variant calling or genomic analysis and interpretation. AWS HealthOmics is not intended to interpret or analyze clinical laboratory tests or other device data, results, and findings, and is not a substitute for third-party tools intended for use in genomic analyses.

## **Features of AWS HealthOmics**

HealthOmics offers the following features.

 HealthOmics Storage — helps you store and share petabytes of raw genomics data efficiently and at low cost per gigabase.

Important notice 1

• HealthOmics Analytics — simplifies how you prepare genomics data for multiomics and multimodal analyses.

• HealthOmics Workflows — automatically provisions and scales the underlying infrastructure for your bioinformatics workflows.

Each component can be used independently or as part of an integrated end-to-end solution.

HealthOmics offers you the following benefits.

- Securely store and combine genomic data HealthOmics integrates with other AWS services such as AWS Lake Formation and Amazon Athena. You can securely store your genomics data and then query or combine it with medical history data for better diagnoses and personalized treatment plans.
- Protect patient privacy HealthOmics is HIPAA eligible. It also integrates with IAM and Amazon CloudWatch so that you can control and log data access, as well as how the data has been used in analyses.
- Built to scale Support large population data analyses with simplified billing and new collaboration tools.
- Maximize efficiency Use automated workflows and integrated tools to streamline data processing and analysis.

You can use HealthOmics for the following biomedical applications:

- Population sequencing Query thousands of genomes at once to understand how genomic variation maps to phenotypes across a population.
- Clinical genomics Build reproducible genomics workflows from sequencer output to reportable data. You can also optimize for high volume throughput and set the compute requirements for high-priority clinical samples to reduce turnaround time.
- Clinical trials Integrate genome analysis into clinical trials to better understand the efficacy of new drug candidates. Simplify and accelerate clinical trials with long-term cost savings and data provenance to meet regulations from governing bodies.
- Enhance research and innovation Streamline and control storage, access, and analysis of anonymized genomics data with built-in row and column-based access control.

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### **Related services**

The following services work with AWS HealthOmics.

- Amazon ECR
- AWS Lake Formation
- Amazon S3
- Amazon Athena

### **Accessing AWS HealthOmics**

AWS HealthOmics features can be accessed by console, CLI, or API.

- 1. AWS Management Console– Provides a web interface that you can use to access HealthOmics.
- 2. AWS Command Line Interface (AWS CLI) Provides commands for a broad set of AWS services, including AWS HealthOmics, and is supported on Windows, macOS, and Linux. For more information about installing the AWS CLI, see AWS Command Line Interface.
- 3. AWS SDKs AWS provides SDKs (Software Development Kits) that consist of libraries and sample code for various programming languages and platforms (Java, Python, Ruby, .NET, iOS, Android, etc.). The SDKs provide a convenient way to use Amazon Omics programmatically. For more information, see the AWS SDK Developer Center.

Related services 3

## **Setting up AWS HealthOmics**

## Sign up for an AWS account

If you do not have an AWS account, complete the following steps to create one.

### To sign up for an AWS account

- 1. Open https://portal.aws.amazon.com/billing/signup.
- 2. Follow the online instructions.

Part of the sign-up procedure involves receiving a phone call and entering a verification code on the phone keypad.

When you sign up for an AWS account, an AWS account root user is created. The root user has access to all AWS services and resources in the account. As a security best practice, assign administrative access to an administrative user, and use only the root user to perform tasks that require root user access.

AWS sends you a confirmation email after the sign-up process is complete. At any time, you can view your current account activity and manage your account by going to <a href="https://aws.amazon.com/">https://aws.amazon.com/</a> and choosing **My Account**.

### Create an administrative user

After you sign up for an AWS account, secure your AWS account root user, enable AWS IAM Identity Center, and create an administrative user so that you don't use the root user for everyday tasks.

#### Secure your AWS account root user

 Sign in to the <u>AWS Management Console</u> as the account owner by choosing **Root user** and entering your AWS account email address. On the next page, enter your password.

For help signing in by using root user, see <u>Signing in as the root user</u> in the *AWS Sign-In User Guide*.

2. Turn on multi-factor authentication (MFA) for your root user.

Sign up for an AWS account

For instructions, see <u>Enable a virtual MFA device for your AWS account root user (console)</u> in the *IAM User Guide*.

#### Create an administrative user

1. Enable IAM Identity Center.

For instructions, see <u>Enabling AWS IAM Identity Center</u> in the *AWS IAM Identity Center User Guide*.

2. In IAM Identity Center, grant administrative access to an administrative user.

For a tutorial about using the IAM Identity Center directory as your identity source, see Configure user access with the default IAM Identity Center directory in the AWS IAM Identity Center User Guide.

### Sign in as the administrative user

 To sign in with your IAM Identity Center user, use the sign-in URL that was sent to your email address when you created the IAM Identity Center user.

For help signing in using an IAM Identity Center user, see <u>Signing in to the AWS access portal</u> in the *AWS Sign-In User Guide*.

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# **AWS HealthOmics permissions**

You can use AWS Identity and Access Management (IAM) to manage access to the AWS HealthOmics API and resources. For users and applications in your account that use AWS HealthOmics, you manage permissions in a permissions policy that you can apply to IAM users, groups, or roles.

To manage permissions for users and applications in your accounts, <u>use the policies that AWS HealthOmics provides</u>, or write your own. The AWS HealthOmics console uses multiple services to get information about your function's configuration and triggers. You can use the provided policies as-is, or as a starting point for more restrictive policies.

AWS HealthOmics uses IAM <u>service roles</u> to access other services on your behalf. For example, you would create or choose a service role when you run a workflow that reads data from Amazon S3. For some features, you also need to <u>configure permissions on resources in other services</u>. Review these requirements before you start working with AWS HealthOmics

For more information about IAM, see What is IAM? in the IAM User Guide.

### **Topics**

- Identity-based IAM policies for AWS HealthOmics
- Service roles for AWS HealthOmics
- Resource permissions
- Permissions for data access using Amazon S3 URIs

## **Identity-based IAM policies for AWS HealthOmics**

To grant users in your account access to AWS HealthOmics, you use identity-based policies in AWS Identity and Access Management (IAM). Identity-based policies can apply directly to IAM users, or to IAM groups and roles that are associated with a user. You can also grant users in another account permission to assume a role in your account and access your AWS HealthOmics resources.

The following IAM policy allows a user to access all AWS HealthOmics API actions, and to pass service roles to AWS HealthOmics.

### **Example User policy**

{

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```
"Version": "2012-10-17",
  "Statement": [
      "Effect": "Allow",
      "Action": [
        "omics:*"
      ],
      "Resource": "*"
    },
      "Effect": "Allow",
      "Action": [
        "iam:PassRole"
      ],
      "Resource": "*",
      "Condition": {
        "StringEquals": {
          "iam:PassedToService": "omics.amazonaws.com"
        }
      }
  ]
}
```

When you use AWS HealthOmics, you also interact with other AWS services. To access these services, use the managed policies provided by each service. To limit access to a subset of resources, you can use the managed policies as a starting point to create your own more restrictive policies.

- <u>AmazonS3FullAccess</u> Access to Amazon S3 buckets and objects used by jobs.
- <u>AmazonEC2ContainerRegistryFullAccess</u> Access to Amazon ECR registries and repositories for workflow container images.
- <u>AWSLakeFormationDataAdmin</u> Access to Lake Formation databases and tables created by analytics stores.
- <u>ResourceGroupsandTagEditorFullAccess</u> Tag AWS HealthOmics resources with AWS HealthOmics tagging API operations.

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The preceding policies do not allow a user to create IAM roles. For a user with these permissions to run a job, an administrator must create the service role that grants AWS HealthOmics permission to access data sources. For more information, see Service roles for AWS HealthOmics.

## Service roles for AWS HealthOmics

You can use service roles to grant AWS HealthOmics permission to access data and upload logs while processing a workflow or importing data to a Omics Storage or Omics Analytics data store. A service role is an AWS Identity and Access Management (IAM) role that an AWS service can use to access resources from other services in your account. You pass a service role to AWS HealthOmics when you start a job.

Service roles must have the following trust policy.

The trust policy allows AWS HealthOmics to assume the role.

#### **Sections**

- Sample IAM policies
- Sample Cloudwatch templates

## Sample IAM policies

The GitHub repository for this guide provides sample IAM policies that you can use as reference for developing service roles. You can use a single role that grants permission for both importing data and sending alerts by combining the applicable policies.

Service roles 8

### **Example Service role**

```
{
    "Version": "2012-10-17",
    "Statement": [
        {
            "Effect": "Allow",
            "Action": [
                "s3:GetObject",
                "s3:PutObject"
            ],
            "Resource": [
                "arn:aws:s3:::my-bucket/*"
            ]
        },
            "Effect": "Allow",
            "Action": [
                "omics:*"
            ],
            "Resource": [
                "arn:aws:omics:us-west-2:123456789012:referenceStore/*"
            ]
        },
        {
            "Effect": "Allow",
            "Action": [
                "s3:ListBucket"
            ],
            "Resource": [
                "arn:aws:s3:::my-bucket"
            ]
        },
            "Effect": "Allow",
            "Action": [
                "logs:DescribeLogStreams",
                "logs:CreateLogStream",
                "logs:PutLogEvents"
            ],
            "Resource": [
                "arn:aws:logs:us-west-2:123456789012:log-group:/aws/omics/
WorkflowLog:log-stream:*"
```

Sample IAM policies

```
},
{
    "Effect": "Allow",
    "Action": [
        "logs:CreateLogGroup"
],
    "Resource": [
        "arn:aws:logs:us-west-2:123456789012:log-group:/aws/omics/
WorkflowLog:*"
    ]
}
]
}
```

## **Sample Cloudwatch templates**

The following sample template creates a service role that gives AWS HealthOmics permission to access S3 buckets that have names prefixed with omics-, and to upload workflow logs.

#### Example Reference store, Amazon S3 and CloudWatch Logs permissions

```
Parameters:
  bucketName:
    Description: Bucket name
    Type: String
Resources:
  serviceRole:
    Type: AWS::IAM::Role
    Properties:
      Policies:
        - PolicyName: read-reference
          PolicyDocument:
            Version: 2012-10-17
            Statement:
            - Effect: Allow
              Action:
                - omics:*
              Resource: !Sub arn:${AWS::Partition}:omics:${AWS::Region}:
${AWS::AccountId}:referenceStore/*
        - PolicyName: read-s3
          PolicyDocument:
            Version: 2012-10-17
```

```
Statement:
            - Effect: Allow
              Action:
                - s3:ListBucket
              Resource: !Sub arn:${AWS::Partition}:s3:::${bucketName}
            - Effect: Allow
              Action:
                - s3:GetObject
                - s3:PutObject
              Resource: !Sub arn:${AWS::Partition}:s3:::${bucketName}/*
        - PolicyName: upload-logs
          PolicyDocument:
            Version: 2012-10-17
            Statement:
            - Effect: Allow
              Action:
                - logs:DescribeLogStreams
                - logs:CreateLogStream
                - logs:PutLogEvents
              Resource: !Sub arn:${AWS::Partition}:logs:${AWS::Region}:
${AWS::AccountId}:loggroup:/aws/omics/WorkflowLog:log-stream:*
            - Effect: Allow
              Action:
                logs:CreateLogGroup
              Resource: !Sub arn:${AWS::Partition}:logs:${AWS::Region}:
${AWS::AccountId}:loggroup:/aws/omics/WorkflowLog:*
      AssumeRolePolicyDocument: |
          "Version": "2012-10-17",
          "Statement": [
            {
              "Action": Γ
                "sts:AssumeRole"
              ],
              "Effect": "Allow",
              "Principal": {
                "Service": [
                  "omics.amazonaws.com"
                ]
              }
            }
          ]
        }
```

## **Resource permissions**

AWS HealthOmics creates and accesses resources in other services on your behalf when you run a job or create a store. In some cases, you need to configure permissions in other services to access resources or to allow AWS HealthOmics to access them.

#### **Sections**

- Lake Formation
- Amazon ECR

### **Lake Formation**

Before you use analytics features in AWS HealthOmics, configure default database settings in Lake Formation.

#### To configure resource permissions in Lake Formation

- 1. Open the Data catalog settings page in the Lake Formation console.
- Uncheck the IAM access control requirements for databases and tables under **Default** permissions for newly created databases and tables.
- 3. Choose **Save**.

HealthOmics Analytics will auto accept data as long as your service policy has the correct RAM permissions, such as the following example.

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```
"ram:GetResourceShareInvitations"
],
    "Resource": "*"
}
]
```

### **Amazon ECR**

When you run a workflow, you provide access to one or more containers for AWS HealthOmics by using Amazon Elastic Container Registry (Amazon ECR). To access container images on your behalf, AWS HealthOmics needs access to your private registry. This policy needs to be added to each container registry that will be used with HealthOmics workflows. Private and cross-account containers are supported as long as they are in the same region. HealthOmics can't currently access public containers.

### To grant AWS HealthOmics permission to access Amazon ECR

- Open the <u>private repositories</u> page in the Amazon ECR console and select the repository you are granting access to.
- 2. From the side bar navigation, select **Permissions**.
- Choose Edit JSON.
- 4. Choose Add Statement.
- 5. Add the following policy statement for **Conditions** and then select **Save Policy**.

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```
]
```

The resource-based policy on the registry grants HealthOmics permission to acquire a container image in the repository.

To use cross-account containers in the same region, you would add a permission like the following example.

```
{
    "Version": "2012-10-17",
    "Statement": [
        {
            "Sid": "OmicsAccessPrincipal",
            "Effect": "Allow",
            "Principal": {
                 "Service": "omics.amazonaws.com"
            },
            "Action": [
                 "ecr:GetDownloadUrlForLayer",
                 "ecr:BatchGetImage",
                 "ecr:BatchCheckLayerAvailability"
            ]
        },
            "Sid": "OmicsAccessCrossAccount",
            "Effect": "Allow",
            "Principal":{
                 "AWS": "arn:aws:iam::{{AWS-account-ID}}:root"
        },
            "Action":[
            "ecr:GetDownloadUrlForLayer",
            "ecr:BatchGetImage",
            "ecr:BatchCheckLayerAvailability"
           ]
        }
   ]
}
    ]
}
```

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## Permissions for data access using Amazon S3 URIs

When a sequence store is created, the creator's root account will automatically have S3:GetObject, S3GetObjectTagging, and S3:ListBucket access. Additionally, if the AWS KMS key on the sequence store is owned by HealthOmics, the root account is added with kms:Decrypt access.

For a user in the account to access the data, you create a policy and attach it to the user or role to allow access to the files using Amazon S3 API operations. To use HealthOmics API operations, you must add AWS HealthOmics permissions to your IAM policy. A policy allowing Amazon S3 API access can be applied at the sequence store level or at a read set level. At the read set level, permission can be restricted either through the prefix or using resource tag filters for sample or subject ID patterns.

The following example gives a user access to a sequence store. You can fine-tune the access with additional conditions or resource-based filters.

```
{
    "Version": "2012-10-17",
    "Statement": [
            "Sid": "S3DirectAccess",
            "Effect": "Allow",
            "Action": [
                "s3:GetObject",
                "s3:ListBucket",
                "s3:GetObjectTagging"
            ],
            "Resource": "*",
            "Condition": {
                "StringEquals": {
                     "s3:DataAccessPointArn": "arn:aws:s3:us-
west-2:555555555555:accesspoint/592761533288-4891675750"
            }
        },
        {
            "Sid": "DefaultSequenceStoreKMS",
            "Effect": "Allow",
            "Action": "kms.Decrypt",
            "Resource": "arn:aws:kms:us-west-2:555555555555:key/fa3b30f5-835d-4a6d-
b3f9-d3898fe0e648"
        }
```

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}

]

To learn more about using IAM policies with HealthOmics, see Service roles for AWS HealthOmics.

Depending on who you are sharing your data with, there are three ways you can use Amazon S3 URIs to share your data. The options are as follows:

- For sharing with users and roles within your account Write a user access policy that includes access to the AWS KMS key and access to the access point. This makes the data accessible for use with the Amazon S3 API operations.
- For sharing with users outside of your account Create a role within the data owner's account that has an access policy that allows the user to assume that role. Adding the user with direct access is not supported.
- Presigned URLs You can also generate a shareable URL for a file in the sequence store.

To learn more about creating presigned URLs by using Amazon S3, see <u>Using presigned URL</u> in the Amazon S3 documentation.

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## **Getting started with AWS HealthOmics**

The following tutorials and lists are intended to help you learn the concepts of each component of AWS HealthOmics. You can start using the service by creating a data store or setting up a basic workflow.

#### **Topics**

- Key Concepts
- CLI Tutorials
- Getting Started (Console)

## **Key Concepts**

Definitions for key concepts and terms that are specific to the functions of AWS HealthOmics are provided in this section. These definitions are provided to help you understand the terminology of AWS HealthOmics and this guide.

### **Topics**

- Storage
- Analytics
- Workflows

### **Storage**

Data storage is separated into sequence stores, for your genomics sequences and related information, and a reference store, for all of your reference genomes. The following terms describe the implementations that are specific to AWS HealthOmics.

- Sequence store A data store for the storage of genomics files. You can have one or more sequence stores within AWS HealthOmics. Access permissions and AWS KMS encryption can be set on a sequence store to control who has access to the data.
- Read set Read sets are an abstraction of genomics reads, which are stored in FASTQ, BAM, or CRAM formats. Read sets can be imported into sequence stores and annotated with metadata. You can apply permissions to read sets using attribute based access control (ABAC).

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• Reference – A genome reference is used with reads to identify where in a genome a specific read, or group of reads, is mapped to. These are in FASTA format and stored in the reference store.

• Reference store – A data store for the storage of reference genomes. You can have a single reference store in each account and region.

### **Analytics**

You can transform and analyze your genomics data with HealthOmics Analytics. Create a variant store or annotation store to include additional information for your queries.

- Variant Store data store that stores variant data at a population scale. Variant stores support both genomic Variant Call Format (gVCF) and VCF inputs.
- Annotation Store A data store representing an annotation database, such as one from a TSV/ CSV, VCF, or General Feature Format (GFF3) file. Annotation Stores are mapped to the same coordinate system as variant stores during an import.

### **Workflows**

With HealthOmics Workflows, you can process and analyze your genomics data.

- Workflow The overall definition of an end to end process including parameters and references
  to tools. Workflow definitions can be expressed as WDL, Nextflow, or CWL. Each created
  workflow will have a unique identifier.
- Run/Workflow Run A single invocation of a workflow. An individual run uses your defined input data and produces an output. Each created run will have a unique identifier.
- *Task* The individual processes within a run. HealthOmics Workflows will use these defined compute specifications to run your task. Each task will have a unique identifier.
- Run Group A group of runs for which you can set the max vCPU, max duration, or max
  concurrent runs to help limit the compute resources used per run. You can specify and configure
  priorities for your workflow runs within a run group. For example, you can specify that a high
  priority run will be performed before one that is lower priority, creating a priority queue. It is
  optional to use a Run Group, and each Run Group will have a unique identifier.

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### **CLI Tutorials**

The following tutorials are intended to help you start using the service by creating a data store or setting up a basic workflow. As prerequisites, your data must be in an Amazon S3 bucket in same region (for example, us-west-2) and the most recent version of the AWS CLI installed.

### Sequence store tutorial

### Creating a Sequence Store using the AWS Command Line Interface

The following example demonstrates using the CreateSequenceStore operation with the AWS CLI. To run the example, you must install the AWS CLI.

The example is formatted for Unix, Linux, and macOS. For Windows, replace the backslash (\) Unix continuation character at the end of each line with a caret (^).

HealthOmics Storage provides storage for genomic files in FASTQ, BAM, and CRAM formats. These files are stored in Read Sets, which are an AWS resource. To store Read Sets, you need to first create a Sequence store, as shown in the following example.

```
aws omics create-sequence-store --name "MySequenceStore"
```

You will receive the following response in JSON, which include the ID number for your newly created Sequence store.

```
{
    "id": "3936421177",
    "arn": "arn:aws:omics:us-west-2:(account):sequenceStore/3936421177",
    "name": "MySequenceStore",
    "creationTime": "2022-07-13T20:09:26.038Z"
}
```

### Variant store tutorial

### Creating a Variant Store using the AWS Command Line Interface

The following example demonstrates using the CreateVariantStore operation with the AWS CLI. To run the example, you must install the latest version of the AWS CLI.

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The example is formatted for Unix, Linux, and macOS. For Windows, replace the backslash (\) Unix continuation character at the end of each line with a caret (^).

To create a variant store, we will need a referenceName and name parameter. The variant store is ready to ingest data when its status is shown as READY.

```
aws omics create-variant-store --name (storeName) --reference (referenceArn)
```

To confirm the creation of your variant store, you will receive the following response.

```
{
    "id": "b533f097bade",
    "reference": {
        "referenceArn": "arn:aws:omics:us-
west-2:451654099157:referenceStore/5638433913/reference/5871590330"
    },
    "status": "CREATING",
    "name": "variantstore",
    "creationTime": "2022-11-08T01:29:36.594566+00:00"
}
```

### **Workflow tutorial**

### Creating a workflow using the AWS Command Line Interface

You will need both input and output Amazon S3 buckets, as well as an IAM role with access to those buckets. The following is an example IAM policy that grants permission to access the contents of an Amazon S3 bucket, the ECR containers, and Cloud Watch logs.

```
"arn:aws:s3:::[[s3path]]/*"
            ]
        },
        }
            "Effect": "Allow",
            "Action": [
                "s3:ListBucket"
            ],
            "Resource": [
                "arn:aws:s3:::[[s3path]]"
            ]
        },
        {
            "Effect": "Allow",
            "Action": [
                "s3:PutObject"
            ],
            "Resource": [
                "arn:aws:s3:::[[output_s3path]]/*"
            ]
        },
            "Effect": "Allow",
            "Action": [
                "logs:DescribeLogStreams",
                "logs:CreateLogStream",
                "logs:PutLogEvents"
            ],
            "Resource": [
                "arn:aws:logs:{{region}}:{{accountId}}:log-group:/aws/omics/
WorkflowLog:log-stream:*"
            1
        },
        }
            "Effect": "Allow",
            "Action": [
                 "logs:CreateLogGroup"
            ],
            "Resource": [
                "arn:aws:logs:{{region}}:{{accountId}}:log-group:/aws/omics/
WorkflowLog: *"
            ]
        },
```

The role will need to authorize the service to assume it before it can be used in a workflow run. This can be done by adding "trust relationships" similar to the following statement.

```
{
    "Effect": "Allow",
    "Principal": {
        "Service": "omics.amazonaws.com"
            },
            "Action": "sts:AssumeRole"
}
```

Workflow definitions must be written in the supported languages, either WDL or Nextflow. The following is a basic example that demonstrates a workflow that reads the contents of an INPUT file and writes them into a RESULT file.

```
version 1.0

# Simple demo workflow, copy input file into output file

workflow TestFlow {
   input {
     File input_txt_file
   }

#copies input file data to output.
```

```
call TxtFileCopyTask{
        input:
            input_txt_file = input_txt_file,
    }
    output {
        File output_txt_file = TxtFileCopyTask.output_txt_file
    }
}
#Task Definitions
task TxtFileCopyTask {
    input {
        File input_txt_file
    }
    command {
        cat ~{input_txt_file} > outfile.txt
    }
    output {
        File output_txt_file = "outfile.txt"
    }
}
```

The workflow definition files need to be zipped before calling the HealthOmics CreateWorkflow API operation.

```
zip definition.zip main.wdl
```

Define your parameters with a parameter-template file like the following JSON file.

```
"image": {
    "description": "Optional ECR image",
    "optional": true
},
"file": {
    "description": "Required input file"
}
```

}

Once you've defined your workflow and the parameters, you can create a workflow using the API as shown.

```
aws omics create-workflow --name Sample --description BasicExample --definition-zip fileb://definition.zip --parameter-template file://params_sample_description.json
```

Once you've created your workflow, you should receive the following response to confirm that the workflow has been created.

```
{
    "arn": "arn:aws:omics:us-west-2:....",
    "id": "12345",
    "status": "CREATING",
    "tags": {
        "resourceArn": "arn:aws:omics:us-west-2:...."
    }
}
```

## **Getting Started (Console)**

The AWS HealthOmics console features three main areas: Storage, Analytics, and Workflows. The following examples will help you start using each component.

### **Topics**

- HealthOmics Storage
- HealthOmics Analytics
- HealthOmics Workflows

## **HealthOmics Storage**

With HealthOmics Storage, you can create a sequence or reference store, import a reference genome, and import genomics files. After you have created your stores and imported your genomic data, you can access and analyze your sequence data.

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#### Create a reference store

#### To create a reference store

- Open the AWS HealthOmics console https://console.aws.amazon.com/omics/.
- 2. On the AWS HealthOmics home page, choose **Get started with HealthOmics** and then choose **Reference genomes** from the Genomics data storage options.
- 3. You can either choose a previously imported reference genome or import a new one. If you haven't imported a reference genome, select **Import reference genome** in the top right.
- 4. On the **Create reference genome import job** page, choose either the **Quick create** or **Manual create** option to create a reference store, and then provide the following information.
  - Reference genome name A unique name for this store.
  - **Description** (optional) A description of this reference store.
  - IAM Role Select a role with access to your reference genome.
  - Reference from Amazon S3 Select your reference sequence file in an Amazon S3 bucket.
  - Tags (optional) Provide up to 50 tags for this reference store.

### Create a sequence store

#### To create a sequence store

- 1. Open the AWS HealthOmics console <a href="https://console.aws.amazon.com/omics/">https://console.aws.amazon.com/omics/</a>.
- 2. On the AWS HealthOmics home page, choose

in the upper left corner of the screen to open the navigation pane. Select **Sequence stores**.

- 3. On the **Create sequence store** page, provide the following information
  - **Sequence store name** A unique name for this store.
  - **Description** (optional) A description of this sequence store.
  - **Data Encryption** Select whether you want data encryption to be owned and managed by AWS or to use a customer managed CMK.
  - **Tags** (optional) Provide up to 50 tags for this sequence store.

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### Import genomics files

#### To import a genomics file

Open the AWS HealthOmics console <a href="https://console.aws.amazon.com/omics/">https://console.aws.amazon.com/omics/</a>.

2. On the AWS HealthOmics home page, choose

in the upper left corner of the screen to open the navigation pane. Select **Sequence stores**.

- On the Sequence stores page, choose the sequence store that you want to import your files into.
- 4. On the individual sequence store page, choose **Import genomic files**.
- 5. On the **Specify import details** page, provide the following information
  - IAM role The IAM role that can access the genomic files on S3.
  - Reference genome The reference genome for this genomics data.
- 6. On the **Specify import manifest** page, specify the following information **Manifest file**. The manifest file is a JSON or YAML file that describes essential information of your genomics data. For information about the manifest file, see Sequence store imports.
- 7. Click Create import job.

### **HealthOmics Analytics**

With HealthOmics Analytics, you can create variant stores and annotation stores. After you have created the stores and imported your data, you can explore and analyze your stores using analytics engines, such as Amazon Athena.

#### Create a variant store

- 1. Open the AWS HealthOmics console https://console.aws.amazon.com/omics/.
- 2. On the AWS HealthOmics home page, choose

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in the upper left corner of the screen to open the navigation pane. Select Variant stores.

- 3. On the **Create variant store** page, provide the following information
  - Variant store name A unique name for this store.

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- **Description** (optional) A description of this variant store.
- Reference genome The reference genome for this variant store.
- **Data Encryption** Choose whether you want data encryption to be owned and managed by AWS or by yourself.
- Tags (optional) Provide up to 50 tags for this variant store.
- 4. Choose **Create variant store**.

#### Create an annotation store

#### To create an annotation store

- 1. Open the AWS HealthOmics console https://console.aws.amazon.com/omics/.
- 2. On the AWS HealthOmics home page, choose

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in the upper left corner of the screen to open the navigation pane. Select Annotation stores.

- 3. On the **Annotation stores** page, choose **Create annotation store**.
- 4. On the Create annotation store page, provide the following information
  - Annotation store name A unique name for this store.
  - **Description** (optional) A description of this reference genome.
  - **Data format and schema details** Select data file format and upload the schema definition for this store.
  - Reference genome The reference genome for this annotation.
  - **Data Encryption** Choose whether you want data encryption to be owned and managed by AWS or by yourself.
  - Tags (optional) Provide up to 50 tags for this annotation store.
- Choose Create annotation store.

## **HealthOmics Workflows**

With HealthOmics Workflows, you can create workflows, runs, and run groups.

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### Create a workflow

#### To create a workflow

Open the AWS HealthOmics console https://console.aws.amazon.com/omics/.

2. On the AWS HealthOmics home page, choose

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in the upper left corner of the screen to open the navigation pane. Select Workflows.

- 3. On the **Create workflow** page, provide the following information
  - Workflow main definion path The file path that directs to the workflow definition.
  - Workflow name A distinctive name for this workflow.
  - **Description** (optional) A description of this workflow.
  - Run storage capacity(optional) The default amount of storage needed for this workflow. The default is 1.2 TB. This storage is deleted after the run completes.
  - **Workflow definition** The Amazon S3 path to the workflow definition zip. Choose whether it is written in Nextflow or WDL from the drop down box.
  - Tags (optional) Provide up to 50 tags for this workflow.
- Choose Next.
- 5. On the **Add workflow parameters** page, provide the workflow parameters. You can either upload a JSON file that specifies the parameters or manually enter your workflow parameters.
- 6. Choose **Create workflow**.

#### Start a run

#### To start a workflow run

- 1. Open the AWS HealthOmics console <a href="https://console.aws.amazon.com/omics/">https://console.aws.amazon.com/omics/</a>.
- 2. On the AWS HealthOmics home page, choose

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in the upper left corner of the screen to open the navigation pane. Select Runs.

- 3. On the **Create run** page, provide the following information
  - Workflow ID The workflow ID associated with this run.
  - Run name A distinctive name for this run.

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• IAM role - The IAM role that can access the data locations referenced in your parameter values. It should also contain a Cloud Watch policy for the service to publish logs to your Cloud Watch account.

- **Run priority** The priority of this run. Higher numbers specify a higher priority, and the highest priority tasks are run first.
- Run storage capacity The amount of temporary storage needed for the run. By default, the run storage capacity that was set for the workflow will be selected. You can select a different run storage capacity for your run.
- **Select S3 output destination** The S3 location where the run outputs will be saved.
- 4. Choose Next.
- 5. On the **Add parameter values** page, provide the workflow parameters. You can either upload a JSON file that specifies the parameters or manually enter your workflow parameters.
- 6. Choose Next.
- 7. On the **Add run groups and tags** page, provide the run group details. Optionally, you can optionally provide up to 50 tags for this run.
- 8. Choose Create run.

### Create a run group

#### To create a run group

- 1. Open the AWS HealthOmics console https://console.aws.amazon.com/omics/.
- 2. On the AWS HealthOmics home page, choose

in the upper left corner of the screen to open the navigation pane. Select **Run groups**.

- 3. On the **Run groups** page, choose **Create run group**.
- 4. On the Create run group details page, provide the following information
  - Run group name A unique name for this run group.
  - Max vCPU for concurrent runs The maximum number of vCPUs running in parallel across the run group.
  - Max run time (hrs) per run The maximum amount of time that a run can be active.
  - **Tags** (optional) Provide up to 50 tags for this sequence store.
- 5. Choose **Create run group**.

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# **Security in AWS HealthOmics**

Cloud security at AWS is the highest priority. As an AWS customer, you benefit from data centers and network architectures that are built to meet the requirements of the most security-sensitive organizations.

Security is a shared responsibility between AWS and you. The <u>shared responsibility model</u> describes this as security *of* the cloud and security *in* the cloud:

- Security of the cloud AWS is responsible for protecting the infrastructure that runs AWS services in the AWS Cloud. AWS also provides you with services that you can use securely. Third-party auditors regularly test and verify the effectiveness of our security as part of the <u>AWS</u>
   <u>Compliance Programs</u>. To learn about the compliance programs that apply to AWS HealthOmics, see AWS Services in Scope by Compliance Program.
- **Security in the cloud** Your responsibility is determined by the AWS service that you use. You are also responsible for other factors including the sensitivity of your data, your company's requirements, and applicable laws and regulations.

This documentation helps you understand how to apply the shared responsibility model when using AWS HealthOmics. The following topics show you how to configure AWS HealthOmics to meet your security and compliance objectives. You also learn how to use other AWS services that help you to monitor and secure your AWS HealthOmics resources.

### **Topics**

- Data protection in AWS HealthOmics
- Identity and access management for AWS HealthOmics
- Compliance validation for AWS HealthOmics
- Resilience in AWS HealthOmics

## **Data protection in AWS HealthOmics**

### **Encryption at rest**

AWS HealthOmics provides encryption by default to protect sensitive customer data at rest by using a service owned AWS Key Management Service (AWS KMS) key. Customer-managed KMS

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keys are also supported. To learn more about Customer-managed KMS Key, see <u>Amazon Key</u> Management Service.

All HealthOmics data stores (Storage and Analytics) support the use of Customer-managed KMS keys. The encryption configuration cannot be changed after a data store has been created. If a data store is using an AWS owned KMS Key, it will be denoted as AWS\_OWNED\_KMS\_KEY and you will not see the specific key used for encryption at rest.

For HealthOmics Workflows, customer-managed keys are not supported by the temporary file system; however, all data is encrypted at rest automatically using XTS-AES-256 block cipher encryption algorithm to encrypt the file system. The IAM user and role used to start a workflow run must also have access to the AWS KMS keys used for workflow input and output buckets. Workflows does not use grants, and AWS KMS encryption is limited to input and output Amazon S3 buckets. The IAM role used both for workflow APIs must also have access to the AWS KMS keys used as well as the input and output Amazon S3 buckets. You can use either IAM roles and permissions to control access or AWS KMS policies. To learn more, see <a href="Authentication and access control for AWS KMS">Authentication and access control for AWS KMS</a>.

Additionally, when using AWS Lake Formation with HealthOmics Analytics, any decrypt permissions associated with the Lake Formation are also given to the input and output Amazon S3 buckets. More information about how AWS Lake Formation manages permissions can be found in the <u>AWS</u> Lake Formation documentation.

HealthOmics Analytics grants Lake Formation kms:Decrypt permissions to read the encrypted data in an S3 bucket. As long as you have permissions to query the data through Lake Formation, you will be able to read the encrypted data. Access to the data is controlled through data access control in Lake Formation, not through a KMS key policy. To learn more, see the <a href="AWS Integrated">AWS Integrated</a> AWS service requests in the Lake Formation documentation.

#### **AWS owned KMS key**

AWS HealthOmics uses these keys by default to automatically encrypt potentially sensitive information such as personally identifiable or Protected Health Information (PHI) data at rest. AWS owned KMS keys aren't stored in your account. They're part of a collection of KMS keys that AWS owns and manages for use in multiple AWS accounts.

AWS services can use AWS owned KMS keys to protect your data. You can't view, manage, use AWS owned KMS keys, or audit their use. However, you don't need to do any work or change any programs to protect the keys that encrypt your data.

You're not charged a monthly fee or a usage fee if you use AWS owned KMS keys, and they don't count against AWS KMS quotas for your account. For more information, see AWS owned keys.

#### **Customer managed KMS keys**

AWS HealthOmics supports the use of a symmetric customer managed KMS key that you create, own, and manage to add a second layer of encryption over the existing AWS owned encryption. Because you have full control of this layer of encryption, you can perform such tasks as:

- Establishing and maintaining key policies, IAM policies, and grants
- Rotating key cryptographic material
- Enabling and disabling key policies
- Adding tags
- Creating key aliases
- Scheduling keys for deletion

You can also use CloudTrail to track the requests that AWS HealthOmics sends to AWS KMS on your behalf. Additional AWS KMS charges apply. For more information, see customer owned keys.

#### Create a customer managed key

You can create a symmetric customer managed key by using the AWS Management Console, or the AWS KMS APIs.

Follow the steps for <u>Creating symmetric customer managed key</u> in the AWS Key Management Service Developer Guide.

Key policies control access to your customer managed key. Every customer managed key must have exactly one key policy, which contains statements that determine who can use the key and how they can use it. When you create your customer managed key, you can specify a key policy. For more information, see <a href="Managing access to customer managed keys">Management Management Manageme

To use your customer managed key with your AWS HealthOmics resources, <a href="kms:CreateGrant">kms:CreateGrant</a> operations must be permitted in the key policy. This adds a grant to a customer managed key that controls access to a specified KMS key. This key gives a user access to the <a href="kms:grant operations">kms:grant operations</a> that AWS HealthOmics requires. See Using grants for more information.

To use your customer managed KMS key with your AWS HealthOmics resources, the following API operations must be permitted in the key policy:

- kms:CreateGrant adds grants to a specific customer managed KMS key, which allows access to grant operations in HealthOmics Analytics and HealthOmics Storage. HealthOmics Workflows does not use grants.
- kms:DescribeKey provides the customer managed key details needed to validate the key. This is required for all operations.
- kms:GenerateDataKey provides access to encrypt resources at rest for all write operations.
- kms:Decrypt provides access to read or search operations for encrypted resources.

The following is a policy statement example that allows a role to create and interact with a data store in AWS HealthOmics which is encrypted by that key:

```
{
    "Statement": [
{
    "Effect": "Allow",
    "Principal": {
        "Service": "omics.amazonaws.com"
    },
    "Action": [
        "kms:Decrypt",
         "kms:DescribeKey",
         "kms:Encrypt",
         "kms:GenerateDataKey*",
         "kms:ReEncrypt*"
    ],
    "Resource": "*"
        }
    ]
}
```

The following policy would create permissions for a data store to decrypt data from an Amazon S3 bucket.

```
{
    "Version": "2012-10-17",
    "Statement": [
```

```
{
            "Effect": "Allow",
            "Action": [
                "omics:GetReference",
                "omics:GetReferenceMetadata"
            ],
            "Resource": [
                "arn:AWS:omics:{{region}}:{{accountId}}:referenceStore/*"
            ]
        },
            "Effect": "Allow",
            "Action": [
                 "s3:GetObject"
            ],
            "Resource": [
                "arn:AWS:s3:::[[s3path]]/*"
            ]
        },
        {
            "Effect": "Allow",
            "Action": [
                "kms:Decrypt"
            ],
            "Resource": [
                 "arn:AWS:kms:{{region}}:{{account_id}}:key/{{key_id}}"
            ]
             "Condition": {
                "StringEquals": {
                     "kms:ViaService": [
                       "s3.{{region}}.amazonAWS.com"
                }
            }
        }
    ]
}
```

# Required IAM permissions for using a customer managed KMS key

When creating a resource such as a data store with AWS KMS encryption using a customer managed KMS key, there are required permissions for both the key policy and the IAM policy for the IAM user or role.

You can use the <u>kms:ViaService condition key</u> to limit use of the KMS key to only requests that originate from AWS HealthOmics.

For more information about key policies, see <u>Enabling IAM policies</u> in the AWS Key Management Service Developer Guide.

The IAM user or role creating your repositories must have the kms:CreateGrant, kms:GenerateDataKey, and kms:DescribeKey permissions plus the necessary AWS HealthOmics permissions.

#### How AWS HealthOmics uses grants in AWS KMS

HealthOmics Analytics requires a <u>grant</u> to use your customer managed KMS key. Grants are not required or used for either HealthOmics Workflows or HealthOmics Storage. When you create a data store encrypted with a customer managed KMS key, AWS HealthOmics creates a grant on your behalf by sending a <u>CreateGrant</u> request to AWS KMS. Grants in AWS KMS are used to give AWS HealthOmics access to a KMS key in a customer account.

It is not recommended to revoke or retire the grants that AWS HealthOmics creates on your behalf. If you revoke or retire the grant that gives AWS HealthOmics permission to use the AWS KMS keys in your account, AWS HealthOmics cannot access this data, encrypt new resources pushed to the data store, or decrypt them when they are pulled. When you revoke or retire a grant for AWS HealthOmics, the change occurs immediately. To revoke access rights, you should delete the data store rather than revoking the grant. When a data store is deleted, AWS HealthOmics retires the grants on your behalf.

#### Monitoring your encryption keys for AWS HealthOmics

You can use CloudTrail to track the requests that AWS HealthOmics sends to AWS KMS on your behalf when using a customer managed KMS key. The log entries in the CloudTrail log show AWS HealthOmics.amazonAWS.com in the userAgent field to clearly distinguish requests made by AWS HealthOmics.

The following examples are CloudTrail events for CreateGrant, GenerateDataKey, Decrypt, and DescribeKey to monitor AWS KMS operations called by AWS HealthOmics to access data encrypted by your customer managed key.

The following also shows how to use CreateGrant to allow AWS HealthOmics to access a customer provided KMS key, enabling AWS HealthOmics to use that KMS key to encrypt all customer data at rest.

You are not required to create your own grants. AWS HealthOmics creates a grant on your behalf by sending a CreateGrant request to AWS KMS. Grants in AWS KMS are used to give AWS HealthOmics access to a AWS KMS key in a customer account.

```
{
   "eventVersion": "1.08",
   "userIdentity": {
       "type": "AssumedRole",
       "principalId": "xx:test",
       "accountId": "xx",
       "accessKeyId": "xxx",
       "sessionContext": {
           "sessionIssuer": {
               "type": "Role",
               "principalId": "xxxx",
               "arn": "arn:AWS:iam::555555555555:role/user-admin",
               "accountId": "55555555555",
               "userName": "user-admin"
           },
           "webIdFederationData": {},
           "attributes": {
               "creationDate": "2022-11-11T01:36:17Z",
               "mfaAuthenticated": "false"
           }
       },
       "invokedBy": "apigateway.amazonAWS.com"
   },
   "eventTime": "2022-11-11T02:34:41Z",
   "eventSource": "kms.amazonAWS.com",
   "eventName": "CreateGrant",
   "AWSRegion": "us-west-2",
   "sourceIPAddress": "apigateway.amazonAWS.com",
   "userAgent": "apigateway.amazonAWS.com",
   "requestParameters": {
       "granteePrincipal": "AWS Internal",
       "keyId": "arn:AWS:kms:us-west-2:555555555555:key/a6e87d77-cc3e-4a98-a354-
e4c275d775ef",
       "operations": [
           "CreateGrant",
           "RetireGrant",
           "Decrypt",
           "GenerateDataKey"
```

```
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    },
    "responseElements": {
        "grantId": "4869b81e0e1db234342842af9f5531d692a76edaff03e94f4645d493f4620ed7",
        "keyId": "arn:AWS:kms:us-west-2:245126421963:key/xx-cc3e-4a98-a354-
e4c275d775ef"
    },
    "requestID": "d31d23d6-b6ce-41b3-bbca-6e0757f7c59a",
    "eventID": "3a746636-20ef-426b-861f-e77efc56e23c",
    "readOnly": false,
    "resources": [
        {
            "accountId": "245126421963",
            "type": "AWS::KMS::Key",
            "ARN": "arn:AWS:kms:us-west-2:245126421963:key/xx-cc3e-4a98-a354-
e4c275d775ef"
        }
    ],
    "eventType": "AWSApiCall",
    "managementEvent": true,
    "recipientAccountId": "245126421963",
    "eventCategory": "Management"
}
```

The following example shows how to use GenerateDataKey to ensure the user has the necessary permissions to encrypt data before storing it.

```
"eventVersion": "1.08",
"userIdentity": {
    "type": "AssumedRole",
    "principalId": "EXAMPLEUSER",
    "arn": "arn:AWS:sts::111122223333:assumed-role/Sampleuser01",
    "accountId": "111122223333",
    "accessKeyId": "EXAMPLEKEYID",
    "sessionContext": {
        "sessionIssuer": {
            "type": "Role",
            "principalId": "EXAMPLEROLE",
            "arn": "arn:AWS:iam::111122223333:role/Sampleuser01",
            "accountId": "111122223333",
            "userName": "Sampleuser01"
```

```
},
            "webIdFederationData": {},
            "attributes": {
                "creationDate": "2021-06-30T21:17:06Z",
                "mfaAuthenticated": "false"
            }
        },
        "invokedBy": "omics.amazonAWS.com"
    },
    "eventTime": "2021-06-30T21:17:37Z",
    "eventSource": "kms.amazonAWS.com",
    "eventName": "GenerateDataKey",
    "AWSRegion": "us-east-1",
    "sourceIPAddress": "omics.amazonAWS.com",
    "userAgent": "omics.amazonAWS.com",
    "requestParameters": {
        "keySpec": "AES_256",
        "keyId": "arn:AWS:kms:us-east-1:111122223333:key/EXAMPLE_KEY_ARN"
    },
    "responseElements": null,
    "requestID": "EXAMPLE_ID_01",
    "eventID": "EXAMPLE_ID_02",
    "readOnly": true,
    "resources": [
        {
            "accountId": "111122223333",
            "type": "AWS::KMS::Key",
            "ARN": "arn:AWS:kms:us-east-1:111122223333:key/EXAMPLE_KEY_ARN"
        }
    ],
    "eventType": "AWSApiCall",
    "managementEvent": true,
    "recipientAccountId": "111122223333",
    "eventCategory": "Management"
}
```

#### Learn more

The following resources provide more information about data at rest encryption.

For more information about <u>AWS Key Management Service basic concepts</u>, see the AWS KMS documentation.

For more information about Security best practices in the AWS KMS documentation.

#### **Encryption in transit**

AWS HealthOmics uses TLS 1.2+ to encrypt data in transit through the public endpoints and through backend services.

# Identity and access management for AWS HealthOmics

AWS Identity and Access Management (IAM) is an AWS service that helps an administrator securely control access to AWS resources. IAM administrators control who can be *authenticated* (signed in) and *authorized* (have permissions) to use AWS HealthOmics resources. IAM is an AWS service that you can use with no additional charge.

#### **Topics**

- Audience
- · Authenticating with identities
- Managing access using policies
- How AWS HealthOmics works with IAM
- Identity-based policy examples for AWS HealthOmics
- AWS managed policies for AWS HealthOmics
- Troubleshooting AWS HealthOmics identity and access

#### **Audience**

How you use AWS Identity and Access Management (IAM) differs, depending on the work that you do in AWS HealthOmics.

**Service user** – If you use the AWS HealthOmics service to do your job, then your administrator provides you with the credentials and permissions that you need. As you use more AWS HealthOmics features to do your work, you might need additional permissions. Understanding how access is managed can help you request the right permissions from your administrator. If you cannot access a feature in AWS HealthOmics, see <u>Troubleshooting AWS HealthOmics identity and access</u>.

**Service administrator** – If you're in charge of AWS HealthOmics resources at your company, you probably have full access to AWS HealthOmics. It's your job to determine which AWS HealthOmics

features and resources your service users should access. You must then submit requests to your IAM administrator to change the permissions of your service users. Review the information on this page to understand the basic concepts of IAM. To learn more about how your company can use IAM with AWS HealthOmics, see How AWS HealthOmics works with IAM.

**IAM administrator** – If you're an IAM administrator, you might want to learn details about how you can write policies to manage access to AWS HealthOmics. To view example AWS HealthOmics identity-based policies that you can use in IAM, see <u>Identity-based policy examples for AWS</u> HealthOmics.

### **Authenticating with identities**

Authentication is how you sign in to AWS using your identity credentials. You must be *authenticated* (signed in to AWS) as the AWS account root user, as an IAM user, or by assuming an IAM role.

You can sign in to AWS as a federated identity by using credentials provided through an identity source. AWS IAM Identity Center (IAM Identity Center) users, your company's single sign-on authentication, and your Google or Facebook credentials are examples of federated identities. When you sign in as a federated identity, your administrator previously set up identity federation using IAM roles. When you access AWS by using federation, you are indirectly assuming a role.

Depending on the type of user you are, you can sign in to the AWS Management Console or the AWS access portal. For more information about signing in to AWS, see <a href="How to sign in to your AWS">How to sign in to your AWS</a> account in the AWS Sign-In User Guide.

If you access AWS programmatically, AWS provides a software development kit (SDK) and a command line interface (CLI) to cryptographically sign your requests by using your credentials. If you don't use AWS tools, you must sign requests yourself. For more information about using the recommended method to sign requests yourself, see <u>Signing AWS API requests</u> in the *IAM User Guide*.

Regardless of the authentication method that you use, you might be required to provide additional security information. For example, AWS recommends that you use multi-factor authentication (MFA) to increase the security of your account. To learn more, see <a href="Multi-factor authentication">Multi-factor authentication</a> in the AWS IAM Identity Center User Guide and <a href="Using multi-factor authentication">Using multi-factor authentication</a> (MFA) in AWS in the IAM User Guide.

#### AWS account root user

When you create an AWS account, you begin with one sign-in identity that has complete access to all AWS services and resources in the account. This identity is called the AWS account *root user* and is accessed by signing in with the email address and password that you used to create the account. We strongly recommend that you don't use the root user for your everyday tasks. Safeguard your root user credentials and use them to perform the tasks that only the root user can perform. For the complete list of tasks that require you to sign in as the root user, see <u>Tasks that require root user credentials</u> in the *IAM User Guide*.

#### **Federated identity**

As a best practice, require human users, including users that require administrator access, to use federation with an identity provider to access AWS services by using temporary credentials.

A federated identity is a user from your enterprise user directory, a web identity provider, the AWS Directory Service, the Identity Center directory, or any user that accesses AWS services by using credentials provided through an identity source. When federated identities access AWS accounts, they assume roles, and the roles provide temporary credentials.

For centralized access management, we recommend that you use AWS IAM Identity Center. You can create users and groups in IAM Identity Center, or you can connect and synchronize to a set of users and groups in your own identity source for use across all your AWS accounts and applications. For information about IAM Identity Center, see <a href="What is IAM Identity Center">What is IAM Identity Center</a>? in the AWS IAM Identity Center User Guide.

#### IAM users and groups

An <u>IAM user</u> is an identity within your AWS account that has specific permissions for a single person or application. Where possible, we recommend relying on temporary credentials instead of creating IAM users who have long-term credentials such as passwords and access keys. However, if you have specific use cases that require long-term credentials with IAM users, we recommend that you rotate access keys. For more information, see <u>Rotate access keys regularly for use cases that require long-term credentials</u> in the <u>IAM User Guide</u>.

An <u>IAM group</u> is an identity that specifies a collection of IAM users. You can't sign in as a group. You can use groups to specify permissions for multiple users at a time. Groups make permissions easier to manage for large sets of users. For example, you could have a group named *IAMAdmins* and give that group permissions to administer IAM resources.

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Users are different from roles. A user is uniquely associated with one person or application, but a role is intended to be assumable by anyone who needs it. Users have permanent long-term credentials, but roles provide temporary credentials. To learn more, see <a href="When to create an IAM user">When to create an IAM user</a> (instead of a role) in the IAM User Guide.

#### IAM roles

An <u>IAM role</u> is an identity within your AWS account that has specific permissions. It is similar to an IAM user, but is not associated with a specific person. You can temporarily assume an IAM role in the AWS Management Console by <u>switching roles</u>. You can assume a role by calling an AWS CLI or AWS API operation or by using a custom URL. For more information about methods for using roles, see <u>Using IAM roles</u> in the <u>IAM User Guide</u>.

IAM roles with temporary credentials are useful in the following situations:

- Federated user access To assign permissions to a federated identity, you create a role and define permissions for the role. When a federated identity authenticates, the identity is associated with the role and is granted the permissions that are defined by the role. For information about roles for federation, see <a href="Creating a role for a third-party Identity Provider">Creating a role for a third-party Identity Provider</a> in the IAM User Guide. If you use IAM Identity Center, you configure a permission set. To control what your identities can access after they authenticate, IAM Identity Center correlates the permission set to a role in IAM. For information about permissions sets, see <a href="Permission sets">Permission sets</a> in the AWS IAM Identity Center User Guide.
- **Temporary IAM user permissions** An IAM user or role can assume an IAM role to temporarily take on different permissions for a specific task.
- Cross-account access You can use an IAM role to allow someone (a trusted principal) in a
  different account to access resources in your account. Roles are the primary way to grant crossaccount access. However, with some AWS services, you can attach a policy directly to a resource
  (instead of using a role as a proxy). To learn the difference between roles and resource-based
  policies for cross-account access, see <a href="How IAM roles differ from resource-based policies">How IAM roles differ from resource-based policies</a> in the
  IAM User Guide.
- Cross-service access Some AWS services use features in other AWS services. For example, when you make a call in a service, it's common for that service to run applications in Amazon EC2 or store objects in Amazon S3. A service might do this using the calling principal's permissions, using a service role, or using a service-linked role.
  - Forward access sessions (FAS) When you use an IAM user or role to perform actions in AWS, you are considered a principal. When you use some services, you might perform an

action that then initiates another action in a different service. FAS uses the permissions of the principal calling an AWS service, combined with the requesting AWS service to make requests to downstream services. FAS requests are only made when a service receives a request that requires interactions with other AWS services or resources to complete. In this case, you must have permissions to perform both actions. For policy details when making FAS requests, see Forward access sessions.

- Service role A service role is an <u>IAM role</u> that a service assumes to perform actions on your behalf. An IAM administrator can create, modify, and delete a service role from within IAM. For more information, see <u>Creating a role to delegate permissions to an AWS service</u> in the *IAM User Guide*.
- Service-linked role A service-linked role is a type of service role that is linked to an AWS service. The service can assume the role to perform an action on your behalf. Service-linked roles appear in your AWS account and are owned by the service. An IAM administrator can view, but not edit the permissions for service-linked roles.
- Applications running on Amazon EC2 You can use an IAM role to manage temporary credentials for applications that are running on an EC2 instance and making AWS CLI or AWS API requests. This is preferable to storing access keys within the EC2 instance. To assign an AWS role to an EC2 instance and make it available to all of its applications, you create an instance profile that is attached to the instance. An instance profile contains the role and enables programs that are running on the EC2 instance to get temporary credentials. For more information, see <a href="Using an IAM role to grant permissions to applications running on Amazon EC2 instances">Using an IAM role to grant permissions to applications running on Amazon EC2 instances</a> in the IAM User Guide.

To learn whether to use IAM roles or IAM users, see When to create an IAM role (instead of a user) in the IAM User Guide.

#### Managing access using policies

You control access in AWS by creating policies and attaching them to AWS identities or resources. A policy is an object in AWS that, when associated with an identity or resource, defines their permissions. AWS evaluates these policies when a principal (user, root user, or role session) makes a request. Permissions in the policies determine whether the request is allowed or denied. Most policies are stored in AWS as JSON documents. For more information about the structure and contents of JSON policy documents, see <a href="Overview of JSON policies">Overview of JSON policies</a> in the IAM User Guide.

Administrators can use AWS JSON policies to specify who has access to what. That is, which **principal** can perform **actions** on what **resources**, and under what **conditions**.

By default, users and roles have no permissions. To grant users permission to perform actions on the resources that they need, an IAM administrator can create IAM policies. The administrator can then add the IAM policies to roles, and users can assume the roles.

IAM policies define permissions for an action regardless of the method that you use to perform the operation. For example, suppose that you have a policy that allows the iam: GetRole action. A user with that policy can get role information from the AWS Management Console, the AWS CLI, or the AWS API.

#### **Identity-based policies**

Identity-based policies are JSON permissions policy documents that you can attach to an identity, such as an IAM user, group of users, or role. These policies control what actions users and roles can perform, on which resources, and under what conditions. To learn how to create an identity-based policy, see <a href="Creating IAM policies">Creating IAM policies</a> in the IAM User Guide.

Identity-based policies can be further categorized as *inline policies* or *managed policies*. Inline policies are embedded directly into a single user, group, or role. Managed policies are standalone policies that you can attach to multiple users, groups, and roles in your AWS account. Managed policies include AWS managed policies and customer managed policies. To learn how to choose between a managed policy or an inline policy, see <u>Choosing between managed policies and inline policies</u> in the *IAM User Guide*.

#### **Resource-based policies**

Resource-based policies are JSON policy documents that you attach to a resource. Examples of resource-based policies are IAM *role trust policies* and Amazon S3 *bucket policies*. In services that support resource-based policies, service administrators can use them to control access to a specific resource. For the resource where the policy is attached, the policy defines what actions a specified principal can perform on that resource and under what conditions. You must <u>specify a principal</u> in a resource-based policy. Principals can include accounts, users, roles, federated users, or AWS services.

Resource-based policies are inline policies that are located in that service. You can't use AWS managed policies from IAM in a resource-based policy.

#### Access control lists (ACLs)

Access control lists (ACLs) control which principals (account members, users, or roles) have permissions to access a resource. ACLs are similar to resource-based policies, although they do not use the JSON policy document format.

Amazon S3, AWS WAF, and Amazon VPC are examples of services that support ACLs. To learn more about ACLs, see <u>Access control list (ACL) overview</u> in the *Amazon Simple Storage Service Developer Guide*.

#### Other policy types

AWS supports additional, less-common policy types. These policy types can set the maximum permissions granted to you by the more common policy types.

- Permissions boundaries A permissions boundary is an advanced feature in which you set the maximum permissions that an identity-based policy can grant to an IAM entity (IAM user or role). You can set a permissions boundary for an entity. The resulting permissions are the intersection of an entity's identity-based policies and its permissions boundaries. Resource-based policies that specify the user or role in the Principal field are not limited by the permissions boundary. An explicit deny in any of these policies overrides the allow. For more information about permissions boundaries, see Permissions boundaries for IAM entities in the IAM User Guide.
- Service control policies (SCPs) SCPs are JSON policies that specify the maximum permissions for an organization or organizational unit (OU) in AWS Organizations. AWS Organizations is a service for grouping and centrally managing multiple AWS accounts that your business owns. If you enable all features in an organization, then you can apply service control policies (SCPs) to any or all of your accounts. The SCP limits permissions for entities in member accounts, including each AWS account root user. For more information about Organizations and SCPs, see <a href="How SCPs work">How SCPs</a> work in the AWS Organizations User Guide.
- Session policies Session policies are advanced policies that you pass as a parameter when you programmatically create a temporary session for a role or federated user. The resulting session's permissions are the intersection of the user or role's identity-based policies and the session policies. Permissions can also come from a resource-based policy. An explicit deny in any of these policies overrides the allow. For more information, see Session policies in the IAM User Guide.

#### Multiple policy types

When multiple types of policies apply to a request, the resulting permissions are more complicated to understand. To learn how AWS determines whether to allow a request when multiple policy types are involved, see Policy evaluation logic in the *IAM User Guide*.

#### How AWS HealthOmics works with IAM

Before you use IAM to manage access to AWS HealthOmics, learn what IAM features are available to use with AWS HealthOmics.

#### IAM features you can use with AWS HealthOmics

IAM feature	AWS HealthOmics support
Identity-based policies	Yes
Resource-based policies	No
Policy actions	Yes
Policy resources	Yes
Policy condition keys	No
ACLs	No
ABAC (tags in policies)	Yes
Temporary credentials	Yes
Principal permissions	Yes
Service roles	Yes
Service-linked roles	No

To get a high-level view of how AWS HealthOmics and other AWS services work with most IAM features, see AWS services that work with IAM in the IAM User Guide.

#### Cross-service confused deputy prevention

The confused deputy problem is a security issue where an entity that doesn't have permission to perform an action can coerce a more-privileged entity to perform the action. In AWS, cross-service impersonation can result in the confused deputy problem. Cross-service impersonation can occur when one service (the *calling service*) calls another service (the *called service*). The calling service can be manipulated to use its permissions to act on another customer's resources in a way it should not otherwise have permission to access. To prevent this, AWS provides tools that help you protect your data for all services with service principals that have been given access to resources in your account.

We recommend using the <u>aws:SourceArn</u> and <u>aws:SourceAccount</u> global condition context keys in resource policies to limit the permissions that AWS HealthOmics gives another service to the resource.

To prevent the confused deputy problem in roles assumed by HealthOmics, set the value of aws:SourceArn to arn:aws:omics:region:accountNumber:\* in the role's trust policy. The wildcard (\*) applies the condition for all HealthOmics resources.

The following trust relationship policy grants HealthOmics access to your resources and uses the aws:SourceArn and aws:SourceAccount global condition context keys to prevent the confused deputy problem. Use this policy when you create a role for HealthOmics.

```
"Version": "2012-10-17",
"Statement": [
  {
    "Sid": "",
    "Effect": "Allow",
    "Principal": {
      "Service": [
        "omics.amazonaws.com"
      ]
    },
    "Action": "sts:AssumeRole",
    "Condition": {
      "StringEquals": {
        "aws:SourceAccount": "accountNumber"
      },
      "StringLike": {
```

```
"aws:SourceArn": "arn:aws:omics:region:accountNumber:*"
}
}
}
```

# **Identity-based policies for AWS HealthOmics**

Supports identity-based policies Yes

Identity-based policies are JSON permissions policy documents that you can attach to an identity, such as an IAM user, group of users, or role. These policies control what actions users and roles can perform, on which resources, and under what conditions. To learn how to create an identity-based policy, see <a href="Creating IAM policies">Creating IAM policies</a> in the IAM User Guide.

With IAM identity-based policies, you can specify allowed or denied actions and resources as well as the conditions under which actions are allowed or denied. You can't specify the principal in an identity-based policy because it applies to the user or role to which it is attached. To learn about all of the elements that you can use in a JSON policy, see <a href="IAM JSON policy elements reference">IAM JSON policy elements reference</a> in the IAM User Guide.

#### **Identity-based policy examples for AWS HealthOmics**

To view examples of AWS HealthOmics identity-based policies, see <u>Identity-based policy examples</u> for AWS HealthOmics.

#### **Resource-based policies within AWS HealthOmics**

Supports resource-based policies No

Resource-based policies are JSON policy documents that you attach to a resource. Examples of resource-based policies are IAM *role trust policies* and Amazon S3 *bucket policies*. In services that support resource-based policies, service administrators can use them to control access to a specific resource. For the resource where the policy is attached, the policy defines what actions a specified principal can perform on that resource and under what conditions. You must specify a principal

in a resource-based policy. Principals can include accounts, users, roles, federated users, or AWS services.

To enable cross-account access, you can specify an entire account or IAM entities in another account as the principal in a resource-based policy. Adding a cross-account principal to a resource-based policy is only half of establishing the trust relationship. When the principal and the resource are in different AWS accounts, an IAM administrator in the trusted account must also grant the principal entity (user or role) permission to access the resource. They grant permission by attaching an identity-based policy to the entity. However, if a resource-based policy grants access to a principal in the same account, no additional identity-based policy is required. For more information, see How IAM roles differ from resource-based policies in the IAM User Guide.

#### **Policy actions for AWS HealthOmics**

Supports policy actions

Yes

Administrators can use AWS JSON policies to specify who has access to what. That is, which **principal** can perform **actions** on what **resources**, and under what **conditions**.

The Action element of a JSON policy describes the actions that you can use to allow or deny access in a policy. Policy actions usually have the same name as the associated AWS API operation. There are some exceptions, such as *permission-only actions* that don't have a matching API operation. There are also some operations that require multiple actions in a policy. These additional actions are called *dependent actions*.

Include actions in a policy to grant permissions to perform the associated operation.

To see a list of AWS HealthOmics actions, see <u>Actions Defined by AWS HealthOmics</u> in the *Service Authorization Reference*.

Policy actions in AWS HealthOmics use the following prefix before the action:

healthomics

To specify multiple actions in a single statement, separate them with commas.

"Action": [

```
"healthomics:action1",
"healthomics:action2"
]
```

To view examples of AWS HealthOmics identity-based policies, see <u>Identity-based policy examples</u> for AWS HealthOmics.

#### **Policy resources for AWS HealthOmics**

Supports policy resources Yes

Administrators can use AWS JSON policies to specify who has access to what. That is, which **principal** can perform **actions** on what **resources**, and under what **conditions**.

The Resource JSON policy element specifies the object or objects to which the action applies. Statements must include either a Resource or a NotResource element. As a best practice, specify a resource using its <a href="Managen Resource Name (ARN)"><u>Amazon Resource Name (ARN)</u></a>. You can do this for actions that support a specific resource type, known as *resource-level permissions*.

For actions that don't support resource-level permissions, such as listing operations, use a wildcard (\*) to indicate that the statement applies to all resources.

```
"Resource": "*"
```

To see a list of AWS HealthOmics resource types and their ARNs, see <u>Resources Defined by AWS HealthOmics</u> in the <u>Service Authorization Reference</u>. To learn with which actions you can specify the ARN of each resource, see Actions Defined by AWS HealthOmics.

To view examples of AWS HealthOmics identity-based policies, see <u>Identity-based policy examples</u> for AWS HealthOmics.

# **Policy condition keys for AWS HealthOmics**

Policy condition keys are not supported in HealthOmics.

#### Access control lists (ACLs) in AWS HealthOmics

Supports ACLs	No
---------------	----

Access control lists (ACLs) control which principals (account members, users, or roles) have permissions to access a resource. ACLs are similar to resource-based policies, although they do not use the JSON policy document format.

#### Attribute-based access control (ABAC) with AWS HealthOmics

	policies)
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Attribute-based access control (ABAC) is an authorization strategy that defines permissions based on attributes. In AWS, these attributes are called *tags*. You can attach tags to IAM entities (users or roles) and to many AWS resources. Tagging entities and resources is the first step of ABAC. Then you design ABAC policies to allow operations when the principal's tag matches the tag on the resource that they are trying to access.

ABAC is helpful in environments that are growing rapidly and helps with situations where policy management becomes cumbersome.

To control access based on tags, you provide tag information in the <u>condition element</u> of a policy using the aws:ResourceTag/*key-name*, aws:RequestTag/*key-name*, or aws:TagKeys condition keys.

If a service supports all three condition keys for every resource type, then the value is **Yes** for the service. If a service supports all three condition keys for only some resource types, then the value is **Partial**.

For more information about ABAC, see <u>What is ABAC?</u> in the *IAM User Guide*. To view a tutorial with steps for setting up ABAC, see <u>Use attribute-based access control (ABAC)</u> in the *IAM User Guide*.

For more information about tagging AWS HealthOmics resources, see <u>Tagging resources in AWS</u> HealthOmics.

The following example shows how you can write an IAM policy denying access to a resource without a specific tag.

```
{
    "Version": "2012-10-17",
    "Statement": [
        {
             "Effect": "Deny",
             "Action": [
                 "omics:*"
             ],
             "Resource": [
                 11 * 11
             ],
             "Condition": {
                 "Null": {
                   "aws:RequestTag/MyCustomTag": "true"
             }
        }
    ]
}
```

You can also limit access to a runs within a run group, as shown.

```
{
    "Version": "2012-10-17",
    "Statement": [
        {
            "Effect": "Allow",
            "Action": [
                "omics:StartRun"
            ],
            "Resource": [
                "arn:aws:omics:us-west-2:123456789012:run/*",
                "arn:aws:omics:us-west-2:123456789012:workflow/1234567",
                "arn:aws:omics:us-west-2:123456789012:runGroup/2345678"
            ],
            "Condition": {
                "StringLike": {
                    "omics:Workflow": "arn:aws:omics:us-east-1:123456789012:workflow/*"
                },
                "StringLike": {
```

```
"omics:RunGroup": "arn:aws:omics:us-east-1:123456789012:runGroup/*"
                }
            }
        },
            "Effect": "Allow",
            "Action": [
                 "omics:GetRun",
                 "omics:ListRunTasks",
                "omics:GetRunTask",
                 "omics:CancelRun",
                 "omics:DeleteRun"
            ],
            "Resource": [
                 "arn:aws:omics:us-west-2:123456789012:run/*"
            ]
        },
    ]
}
```

#### **Using Temporary credentials with AWS HealthOmics**

Supports temporary credentials Yes

Some AWS services don't work when you sign in using temporary credentials. For additional information, including which AWS services work with temporary credentials, see <u>AWS services that</u> work with IAM in the *IAM User Guide*.

You are using temporary credentials if you sign in to the AWS Management Console using any method except a user name and password. For example, when you access AWS using your company's single sign-on (SSO) link, that process automatically creates temporary credentials. You also automatically create temporary credentials when you sign in to the console as a user and then switch roles. For more information about switching roles, see <a href="Switching to a role">Switching to a role (console)</a> in the IAM User Guide.

You can manually create temporary credentials using the AWS CLI or AWS API. You can then use those temporary credentials to access AWS. AWS recommends that you dynamically generate temporary credentials instead of using long-term access keys. For more information, see <a href="Temporary security credentials in IAM">Temporary security credentials in IAM</a>.

#### **Cross-service principal permissions for AWS HealthOmics**

Supports forward access sessions (FAS) Yes

When you use an IAM user or role to perform actions in AWS, you are considered a principal. When you use some services, you might perform an action that then initiates another action in a different service. FAS uses the permissions of the principal calling an AWS service, combined with the requesting AWS service to make requests to downstream services. FAS requests are only made when a service receives a request that requires interactions with other AWS services or resources to complete. In this case, you must have permissions to perform both actions. For policy details when making FAS requests, see Forward access sessions.

#### Service roles for AWS HealthOmics

Supports service roles	Yes
------------------------	-----

A service role is an IAM role that a service assumes to perform actions on your behalf. An IAM administrator can create, modify, and delete a service role from within IAM. For more information, see Creating a role to delegate permissions to an AWS service in the IAM User Guide.



#### Marning

Changing the permissions for a service role might break AWS HealthOmics functionality. Edit service roles only when AWS HealthOmics provides guidance to do so.

#### Service-linked roles for AWS HealthOmics

|--|

A service-linked role is a type of service role that is linked to an AWS service. The service can assume the role to perform an action on your behalf. Service-linked roles appear in your AWS account and are owned by the service. An IAM administrator can view, but not edit the permissions for service-linked roles.

For details about creating or managing service-linked roles, see <u>AWS services that work with IAM</u>. Find a service in the table that includes a Yes in the **Service-linked role** column. Choose the **Yes** link to view the service-linked role documentation for that service.

#### **Identity-based policy examples for AWS HealthOmics**

By default, users and roles don't have permission to create or modify AWS HealthOmics resources. They also can't perform tasks by using the AWS Management Console, AWS Command Line Interface (AWS CLI), or AWS API. To grant users permission to perform actions on the resources that they need, an IAM administrator can create IAM policies. The administrator can then add the IAM policies to roles, and users can assume the roles.

To learn how to create an IAM identity-based policy by using these example JSON policy documents, see Creating IAM policies in the IAM User Guide.

For details about actions and resource types defined by AWS HealthOmics, including the format of the ARNs for each of the resource types, see <u>Actions, Resources, and Condition Keys for AWS</u> HealthOmics in the *Service Authorization Reference*.

#### **Topics**

- Policy best practices
- Using the AWS HealthOmics console
- · Allow users to view their own permissions

#### **Policy best practices**

Identity-based policies determine whether someone can create, access, or delete AWS HealthOmics resources in your account. These actions can incur costs for your AWS account. When you create or edit identity-based policies, follow these guidelines and recommendations:

- Get started with AWS managed policies and move toward least-privilege permissions To
  get started granting permissions to your users and workloads, use the AWS managed policies
  that grant permissions for many common use cases. They are available in your AWS account. We
  recommend that you reduce permissions further by defining AWS customer managed policies
  that are specific to your use cases. For more information, see <u>AWS managed policies</u> or <u>AWS</u>
  managed policies for job functions in the IAM User Guide.
- Apply least-privilege permissions When you set permissions with IAM policies, grant only the permissions required to perform a task. You do this by defining the actions that can be taken on

specific resources under specific conditions, also known as *least-privilege permissions*. For more information about using IAM to apply permissions, see <u>Policies and permissions in IAM</u> in the *IAM User Guide*.

- Use conditions in IAM policies to further restrict access You can add a condition to your policies to limit access to actions and resources. For example, you can write a policy condition to specify that all requests must be sent using SSL. You can also use conditions to grant access to service actions if they are used through a specific AWS service, such as AWS CloudFormation. For more information, see IAM JSON policy elements: Condition in the IAM User Guide.
- Use IAM Access Analyzer to validate your IAM policies to ensure secure and functional
  permissions IAM Access Analyzer validates new and existing policies so that the policies
  adhere to the IAM policy language (JSON) and IAM best practices. IAM Access Analyzer provides
  more than 100 policy checks and actionable recommendations to help you author secure and
  functional policies. For more information, see <a href="IAM Access Analyzer policy validation">IAM User Guide</a>.
- Require multi-factor authentication (MFA) If you have a scenario that requires IAM users or a root user in your AWS account, turn on MFA for additional security. To require MFA when API operations are called, add MFA conditions to your policies. For more information, see Configuring MFA-protected API access in the IAM User Guide.

For more information about best practices in IAM, see <u>Security best practices in IAM</u> in the *IAM User Guide*.

#### **Using the AWS HealthOmics console**

To access the AWS HealthOmics console, you must have a minimum set of permissions. These permissions must allow you to list and view details about the AWS HealthOmics resources in your AWS account. If you create an identity-based policy that is more restrictive than the minimum required permissions, the console won't function as intended for entities (users or roles) with that policy.

You don't need to allow minimum console permissions for users that are making calls only to the AWS CLI or the AWS API. Instead, allow access to only the actions that match the API operation that they're trying to perform.

#### Allow users to view their own permissions

This example shows how you might create a policy that allows IAM users to view the inline and managed policies that are attached to their user identity. This policy includes permissions to complete this action on the console or programmatically using the AWS CLI or AWS API.

```
{
    "Version": "2012-10-17",
    "Statement": [
        {
            "Sid": "ViewOwnUserInfo",
            "Effect": "Allow",
            "Action": [
                "iam:GetUserPolicy",
                "iam:ListGroupsForUser",
                "iam:ListAttachedUserPolicies",
                "iam:ListUserPolicies",
                "iam:GetUser"
            ],
            "Resource": ["arn:aws:iam::*:user/${aws:username}"]
        },
        {
            "Sid": "NavigateInConsole",
            "Effect": "Allow",
            "Action": [
                "iam:GetGroupPolicy",
                "iam:GetPolicyVersion",
                "iam:GetPolicy",
                "iam:ListAttachedGroupPolicies",
                "iam:ListGroupPolicies",
                "iam:ListPolicyVersions",
                "iam:ListPolicies",
                "iam:ListUsers"
            ],
            "Resource": "*"
        }
    ]
}
```

# **AWS managed policies for AWS HealthOmics**

An AWS managed policy is a standalone policy that is created and administered by AWS. AWS managed policies are designed to provide permissions for many common use cases so that you can start assigning permissions to users, groups, and roles.

Keep in mind that AWS managed policies might not grant least-privilege permissions for your specific use cases because they're available for all AWS customers to use. We recommend that you reduce permissions further by defining <a href="customer managed policies">customer managed policies</a> that are specific to your use cases.

You cannot change the permissions defined in AWS managed policies. If AWS updates the permissions defined in an AWS managed policy, the update affects all principal identities (users, groups, and roles) that the policy is attached to. AWS is most likely to update an AWS managed policy when a new AWS service is launched or new API operations become available for existing services.

For more information, see AWS managed policies in the IAM User Guide.

# AWS managed policy: AmazonOmicsFullAccess

You can attach the AmazonOmicsFullAccess policy to your IAM identities to give them full access to AWS HealthOmics.

This policy grants full access permissions to all AWS HealthOmics actions. When you create an annotation or variant store, Omics will also give you access to those stores through a Resource Share Invitation in the Resource Access Manager (RAM) console. For more information on Resource Share invitations through Lake Formation, see the <u>Lake Formation documentation</u>. For an Omics admin policy, you will also need the following permissions to access your Amazon S3 bucket.

- PutObject
- GetObject
- ListBucket
- AbortMultipartUpload

#### • ListMultipartUploadParts

```
{
 "Version": "2012-10-17",
 "Statement": [
   "Effect": "Allow",
   "Action": [
   "omics:*"
  ],
  "Resource": "*"
  },
  {
   "Effect": "Allow",
   "Action": [
    "ram:AcceptResourceShareInvitation",
    "ram:GetResourceShareInvitations"
   ],
   "Resource": "*",
   "Condition": {
    "StringEquals": {
     "aws:CalledViaLast": "omics.amazonaws.com"
    }
  }
  },
   "Effect": "Allow",
   "Action": "iam:PassRole",
   "Resource": "*",
   "Condition": {
    "StringEquals": {
     "iam:PassedToService": "omics.amazonaws.com"
    }
   }
  }
]
}
```

#### AWS managed policy: AmazonOmicsReadOnlyAccess

You can attach the AWSOmicsReadOnlyAccess policy to your IAM identities when you wish to limit the permissions for that identity to read-only access.

```
{
  "Version": "2012-10-17",
  "Statement": [
    {
        "Effect": "Allow",
        "Action": [
        "omics:Get*"
        "omics:List*"
    ],
        "Resource": "*"
    }
}
```

#### AWS HealthOmics updates to AWS managed policies

View details about updates to AWS managed policies for AWS HealthOmics since this service began tracking these changes. For automatic alerts about changes to this page, subscribe to the RSS feed on the AWS HealthOmics Document history page.

Change	Description	Date
AmazonOmicsFullAccess - New policy added	AWS HealthOmics added a new policy to grant a user full access to all actions and resources. To learn more, see <a href="Mailto:AmazonOmicsFullAccess">AmazonOmicsFullAccess</a> .	February 23, 2023
AWS HealthOmics started tracking changes	AWS HealthOmics started tracking changes for its AWS managed policies.	November 29, 2022

Change	Description	Date
AmazonOmicsReadOnl yAccess - New policy added	AWS HealthOmics added a new policy that limits access to read only. To learn more, <a href="Mailto:AmazonOmicsReadOnlyAccess">AmazonOmicsReadOnlyAccess</a> .	November 29, 2022

#### **Troubleshooting AWS HealthOmics identity and access**

Use the following information to help you diagnose and fix common issues that you might encounter when working with AWS HealthOmics and IAM.

#### **Topics**

- I am not authorized to perform an action in AWS HealthOmics
- I am not authorized to perform iam:PassRole
- I want to allow people outside of my AWS account to access my AWS HealthOmics resources

#### I am not authorized to perform an action in AWS HealthOmics

If you receive an error that you're not authorized to perform an action, your policies must be updated to allow you to perform the action.

The following example error occurs when the mateojackson IAM user tries to use the console to view details about a fictional my-example-widget resource but doesn't have the fictional healthomics: GetWidget permissions.

```
User: arn:aws:iam::123456789012:user/mateojackson is not authorized to perform: healthomics:GetWidget on resource: my-example-widget
```

In this case, the policy for the mateojackson user must be updated to allow access to the *my-example-widget* resource by using the healthomics: *GetWidget* action.

If you need help, contact your AWS administrator. Your administrator is the person who provided you with your sign-in credentials.

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#### I am not authorized to perform iam:PassRole

If you receive an error that you're not authorized to perform the iam: PassRole action, your policies must be updated to allow you to pass a role to AWS HealthOmics.

Some AWS services allow you to pass an existing role to that service instead of creating a new service role or service-linked role. To do this, you must have permissions to pass the role to the service.

The following example error occurs when an IAM user named marymajor tries to use the console to perform an action in AWS HealthOmics. However, the action requires the service to have permissions that are granted by a service role. Mary does not have permissions to pass the role to the service.

```
User: arn:aws:iam::123456789012:user/marymajor is not authorized to perform: iam:PassRole
```

In this case, Mary's policies must be updated to allow her to perform the iam: PassRole action.

If you need help, contact your AWS administrator. Your administrator is the person who provided you with your sign-in credentials.

# I want to allow people outside of my AWS account to access my AWS HealthOmics resources

You can create a role that users in other accounts or people outside of your organization can use to access your resources. You can specify who is trusted to assume the role. For services that support resource-based policies or access control lists (ACLs), you can use those policies to grant people access to your resources.

To learn more, consult the following:

- To learn whether AWS HealthOmics supports these features, see <u>How AWS HealthOmics works</u> with IAM.
- To learn how to provide access to your resources across AWS accounts that you own, see Providing access to an IAM user in another AWS account that you own in the IAM User Guide.
- To learn how to provide access to your resources to third-party AWS accounts, see <a href="Providing access to AWS accounts owned by third parties in the IAM User Guide">IAM User Guide</a>.

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• To learn how to provide access through identity federation, see <u>Providing access to externally</u> authenticated users (identity federation) in the *IAM User Guide*.

• To learn the difference between using roles and resource-based policies for cross-account access, see How IAM roles differ from resource-based policies in the IAM User Guide.

# **Compliance validation for AWS HealthOmics**

Third-party auditors assess the security and compliance of AWS HealthOmics as part of multiple AWS compliance programs. This includes HIPAA, FedRAMP, and others.

For a list of AWS services in scope of specific compliance programs, see <u>AWS Services in Scope by</u> <u>Compliance Program</u>. For general information, see <u>AWS Compliance Programs</u>.

You can download third-party audit reports using AWS Artifact. For more information, see Downloading Reports in AWS Artifact.

Your compliance responsibility when using AWS HealthOmics is determined by the sensitivity of your data, your company's compliance objectives, and applicable laws and regulations. AWS provides the following resources to help with compliance:

- <u>Security and Compliance Quick Start Guides</u> These deployment guides discuss architectural
  considerations and provide steps for deploying security- and compliance-focused baseline
  environments on AWS.
- <u>Architecting for HIPAA Security and Compliance Whitepaper</u> This whitepaper describes how companies can use AWS to create HIPAA-compliant applications.
- <u>AWS Compliance Resources</u> This collection of workbooks and guides might apply to your industry and location.
- <u>Evaluating Resources with Rules</u> in the *AWS Config Developer Guide* AWS Config; assesses how well your resource configurations comply with internal practices, industry guidelines, and regulations.
- <u>AWS Security Hub</u> This AWS service provides a comprehensive view of your security state within AWS that helps you check your compliance with security industry standards and best practices.

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# **Resilience in AWS HealthOmics**

The AWS global infrastructure is built around AWS Regions and Availability Zones. AWS Regions provide multiple physically separated and isolated Availability Zones, which are connected with low-latency, high-throughput, and highly redundant networking. With Availability Zones, you can design and operate applications and databases that automatically fail over between zones without interruption. Availability Zones are more highly available, fault tolerant, and scalable than traditional single or multiple data center infrastructures.

For more information about AWS Regions and Availability Zones, see AWS Global Infrastructure.

In addition to the AWS global infrastructure, AWS HealthOmics offers several features to help support your data resiliency and backup needs.

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# **Regions and endpoints for AWS HealthOmics**

# **Regions and endpoints for AWS HealthOmics**

For a full list of regions and endpoints, see the AWS General Reference.

In addition to the AWS regions that are active by default, there are also *Opt-in Regions* which need to be activated. To learn more about how to activate or deactivate a Region, see <a href="Specify which AWS">Specify which AWS</a> Regions your account can use in the AWS Account Management guide.

# Logging AWS HealthOmics API calls using AWS CloudTrail

AWS HealthOmics is integrated with AWS CloudTrail, a service that provides a record of actions taken by a user, role, or an AWS service in AWS HealthOmics. CloudTrail captures all API calls for AWS HealthOmics as events. The calls captured include calls from the AWS HealthOmics console and code calls to the AWS HealthOmics API operations. If you create a trail, you can enable continuous delivery of CloudTrail events to an Amazon S3 bucket, including events for AWS HealthOmics. If you don't configure a trail, you can still view the most recent events in the CloudTrail console in **Event history**. Using the information collected by CloudTrail, you can determine the request that was made to AWS HealthOmics, the IP address from which the request was made, who made the request, when it was made, and additional details.

To learn more about CloudTrail, see the AWS CloudTrail User Guide.

#### AWS HealthOmics information in CloudTrail

CloudTrail is enabled on your AWS account when you create the account. When activity occurs in AWS HealthOmics, that activity is recorded in a CloudTrail event along with other AWS service events in **Event history**. You can view, search, and download recent events in your AWS account. For more information, see <u>Viewing events with CloudTrail Event history</u>.

For an ongoing record of events in your AWS account, including events for AWS HealthOmics, create a trail. A *trail* enables CloudTrail to deliver log files to an Amazon S3 bucket. By default, when you create a trail in the console, the trail applies to all AWS Regions. The trail logs events from all Regions in the AWS partition and delivers the log files to the Amazon S3 bucket that you specify. Additionally, you can configure other AWS services to further analyze and act upon the event data collected in CloudTrail logs. For more information, see the following:

- Overview for creating a trail
- CloudTrail supported services and integrations
- Configuring Amazon SNS notifications for CloudTrail
- Receiving CloudTrail log files from multiple regions and Receiving CloudTrail log files from multiple accounts

All AWS HealthOmics actions are logged by CloudTrail and are documented in the <u>AWS</u>

<u>HealthOmics API Reference</u>. For example, calls to the CreateReferenceeStore,

StartVariantImportJob and CreateWorkflow actions generate entries in the CloudTrail log files.

Every event or log entry contains information about who generated the request. The identity information helps you determine the following:

- Whether the request was made with IAM user credentials.
- Whether the request was made with temporary security credentials for a role or federated user.
- Whether the request was made by another AWS service.

For more information, see the CloudTrail userIdentity element.

### **Understanding AWS HealthOmics log file entries**

A trail is a configuration that enables delivery of events as log files to an Amazon S3 bucket that you specify. CloudTrail log files contain one or more log entries. An event represents a single request from any source and includes information about the requested action, the date and time of the action, request parameters, and so on. CloudTrail log files aren't an ordered stack trace of the public API calls, so they don't appear in any specific order.

The following example shows a CloudTrail log entry that demonstrates the CreateWorkflow action.

```
{
    "eventVersion": "1.08",
    "userIdentity": {
        "type": "AssumedRole",
        "principalId": "AROAIU53LOGOMTOPXXNPG:username",
        "arn": "arn:aws:sts::account:assumed-role/admin/username",
        "accountId": "account-id",
        "accessKeyId": "accessKeyId",
        "sessionContext": {
            "sessionIssuer": {
                "type": "Role",
                "principalId": "AROAIU53LOGOMTOPXXNPG",
                "arn": "arn:aws:iam::account:role/admin",
                "accountId": "account",
                "userName": "admin"
            },
```

```
"webIdFederationData": {},
            "attributes": {
                "creationDate": "2022-07-23T18:26:09Z",
                "mfaAuthenticated": "false"
            }
        }
    },
    "eventTime": "2022-07-23T18:46:42Z",
    "eventSource": "omics.amazonaws.com",
    "eventName": "CreateWorkflow",
    "awsRegion": "us-west-2",
    "sourceIPAddress": "205.251.233.176",
    "userAgent": "aws-cli/1.22.45 Python/3.9.13 Darwin/20.6.0 botocore/1.23.45",
    "requestParameters": {
        "name": "parameter_name",
        "definitionZip": "czM6Ly93b3JrZmxvd2RlZi1oZWxsby9kZWZpbml0aW9uLnppcA==",
        "requestId": "d788a73c-b81b-45fb-a8a6-d8bb4449ec8a"
    },
    "responseElements": {
        "id": "1002571",
        "arn": "arn:aws:omics:us-west-2:555555555555:instance/i-b188560f ",
        "status": "CREATING",
        "tags": {
            "resourceArn": "arn:aws:omics:us-west-2:083685709690:workflow/1002571"
        }
    },
    "requestID": "842d731d-f264-4b08-a2c9-2f7d45e1eaa3",
    "eventID": "76872ca2-f208-4193-807d-7dd7ea34e6b2",
    "readOnly": false,
    "eventType": "AwsApiCall",
    "managementEvent": true,
    "recipientAccountId": "083685709690",
    "eventCategory": "Management"
}
```

### **Monitoring AWS HealthOmics**

Monitoring is an important part of maintaining the reliability, availability, and performance of AWS HealthOmics and your other AWS solutions. AWS provides the following monitoring tools to watch HealthOmics, report when something is wrong, and take automatic actions when appropriate:

- Amazon CloudWatch monitors your AWS resources and and the applications you run on AWS
  in real time. You can collect and track metrics, create customized dashboards, and set alarms
  that notify you or take actions when a specified metric reaches a threshold that you specify.
  For example, you can have CloudWatch track CPU usage or other metrics of your Amazon EC2
  instances and automatically launch new instances when needed. For more information, see the
  Amazon CloudWatch User Guide.
- Amazon CloudWatch Logs enables you to monitor, store, and access your log files from Amazon EC2 instances, CloudTrail, and other sources. CloudWatch Logs can monitor information in the log files and notify you when certain thresholds are met. You can also archive your log data in highly durable storage. For more information, see the Amazon CloudWatch Logs User Guide.
- AWS CloudTrail captures API calls and related events made by or on behalf of your AWS account and delivers the log files to an Amazon S3 bucket that you specify. You can identify which users and accounts called AWS, the source IP address from which the calls were made, and when the calls occurred. For more information, see the AWS CloudTrail User Guide.

Amazon EventBridge is a serverless event bus service that makes it easy to connect your applications with data from a variety of sources. EventBridge delivers a stream of real-time data from your own applications, Software-as-a-Service (SaaS) applications, and AWS services and routes that data to targets such as Lambda. This enables you to monitor events that happen in services, and build event-driven architectures. For more information, see the <a href="mailto:Amazon EventBridge">Amazon EventBridge</a> User Guide.

## Monitoring HealthOmics with Amazon CloudWatch

You can monitor HealthOmics using CloudWatch, which collects raw data and processes it into readable, near real-time metrics. These statistics are kept for 15 months, so that you can access historical information and gain a better perspective on how your web application or service is performing. You can also set alarms that watch for certain thresholds, and send notifications or take actions when those thresholds are met. For more information, see the <a href="Managenglewith"><u>Amazon CloudWatch User Guide.</u></a>

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The HealthOmics service reports the following metrics in the AWS/Omics namespace.

API Call Count metrics are reported for the following HealthOmics APIs. Only the API Operation dimension is reported.

- Reference and reference store APIs —CreateReferenceStore, DeleteReferenceStore, StartReferenceImportJob
- Sequence store and read set APIs —CreateSequenceStore, DeleteSequenceStore, StartReadSetImportJob, StartReadSetActivationJob, StartReadSetExportJob
- Variant store APIs CreateVariantStore, DeleteVariantStore, StartVariantImportJob, CancelVariantImportJob
- Annotation store APIs CreateAnnotationStore, DeleteAnotationStore, StartAnnotationImportJob, CancelAnnotationImportJob
- Workflow, run, and run group APIs CreateWorkflow, DeleteWorkflow, StartRun, CancelRun, DeleteRun, CreateRunGroup, DeleteRunGroup

#### Viewing *HealthOmics* metrics

CloudWatch metrics for HealthOmics are viewable in the CloudWatch console.

#### To view metrics (CloudWatch console)

- 1. Sign in to the AWS Management Console and open the CloudWatch console.
- Choose Metrics, choose All Metrics, and then choose AWS/Usage.
- 3. Filter Service for HealthOmics.
- 4. Choose the dimension, choose a metric name, then choose **Add to graph**.
- 5. Choose a value for the date range. The metric count for the selected date range is displayed in the graph.

## Creating an alarm using CloudWatch

A CloudWatch alarm watches a single metric over a specified time period, and performs one or more actions: sending a notification to an Amazon Simple Notification Service (Amazon SNS) topic or Auto Scaling policy. The action or actions are based on the value of the metric relative to a given threshold over a number of time periods that you specify. CloudWatch can also send you an Amazon SNS message when the alarm changes state.

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CloudWatch alarms invoke actions only when the state changes and has persisted for the period you specify.

#### To view metrics (CloudWatch console)

- 1. Sign in to the AWS Management Console and open the CloudWatch console.
- 2. Choose **Alarms**, and then choose **Create Alarm**.
- 3. Choose AWS/Usage, and then choose an HealthOmics metric using the Service dimension.
- 4. For **Time Range**, choose a time range to monitor, and then choose **Next**.
- 5. Enter a **Name** and **Description**.
- 6. For Whenever, choose >=, and type a maximum value.
- 7. If you want CloudWatch to send an email when the alarm state is reached, in the Actions section, for Whenever this alarm, choose State is **ALARM**. For Send notification to, choose a mailing list or choose **New list** and create a new mailing list.
- 8. Preview the alarm in the Alarm Preview section. If you are satisfied with the alarm, choose **Create Alarm**.

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## **Using EventBridge with AWS HealthOmics**

AWS HealthOmics generates and sends events to Amazon EventBridge as a best effort delivery to your account's default bus. After EventBridge is enabled, all events are sent to EventBridge. You can use EventBridge rules to route events to additional targets. The following table lists the events that are sent to EventBridge.

Resource or event	Possible status description
Variant or annotation store	Creating, created, updating, updated, deleting, deleted, or creation failed
Reference or sequence store	Created or deleted
Read sets	Processing upload, upload failed, active, archived, activating, or deleted
Variant or annotation import job	Submitted, in progress, cancelled, completed, failed, or completed with failures
Reference import or export job, read set import or export	Submitted, in progress, completed, failed, or completed with failures
Read set activation job	Submitted, in progress, completed, failed, or completed with failures
Workflow status	Possible statuses are creation success, creation failure, deletion success, or deletion failure
Run group status	Active or deleted
Run or task	Pending, starting, running, stopping, completed, deleted, failed, or cancelled
Reference	Active or deleted

#### EventBridge event message structure and examples

AWS HealthOmics sends events to Amazon EventBridge whenever a resource is created, updated, deleted, or changes state. You can use EventBridge and these events to write rules that take actions, such as notifying you when a resource changes state. For more information, see <a href="What is Amazon EventBridge">What is Amazon EventBridge</a>?

HealthOmics provides best effort delivery of state changes to EventBridge. The event is an object with JSON structure that also contains metadata details. You can use the metadata as input to either recreate the event or learn more information. The following fields are included:

- version Currently 0 (zero) for all events.
- id A Version 4 UUID generated for every event.
- detail-type The type of event that's being sent.
- account The 12-digit AWS account ID of the bucket owner.
- source Identifies the service that generated the event.
- time The time the event occurred.
- region Identifies the AWS Region of the bucket.
- resources A JSON array that contains the Amazon Resource Name (ARN) of the bucket.
- detail A JSON object that contains information about the event.

For run events, the following fields are included:

- uuid The universally unique identifier for the run.
- runOutputUri The URI for where the run will write its output data.

The following is an example of an event that's created when a read set status changes.

```
{
  "version": "0"
  "id": "64ca0eda-9751-dc55-c41a-1bd50b4fc9b7",
  "detail-type": "Read Set Status Change",
  "source": "aws.omics",
  "account": "123456789012",
  "time": "2023-04-04T17:53:06Z",
```

```
"region": "us-west-2",
    "resources": ["arn:aws:omics:us-west-2:123456789012:sequenceStore/1234567890/
readSet/3456789012"],
    "detail": {
        "omicsVersion": "1.0.0",
        "arn": "arn:aws:omics:us-west-2:123456789012:sequenceStore/1234567890/
readSet/3456789012",
        "sequenceStoreId": "1234567890",
        "id": "3456789012",
        "status": "PROCESSING_UPLOAD"
    }
}
```

A similar event gets created for a variant store import job.

```
{
    "version": "0",
    "id": "6a7e8feb-b491-4cf7-a9f1-bf3703467718",
    "detail-type": "Variant Store Status Change",
    "source": "aws.omics",
    "account": "123456789012",
    "time": "2015-12-22T18:43:48Z",
    "region": "us-east-1",
    "resources": ["arn:aws:omics:us-east-1:123456789012:bcvariantstore2"],
    "detail": {
        "omicsVersion": "1.0.0",
        "arn": "arn:aws:omics:us-east-1:123456789012:bcvariantstore2",
        "status": "CREATED",
        "storeId": "6710c5f02610",
        "storeName": "bcvariantstore2",
    }
}
```

The following is an event that would be created for a change in import job status.

```
"version": "0",
"id": "6a7e8feb-b491-4cf7-a9f1-bf3703467718",
"detail-type": "Variant Import Job Status Change",
"source": "aws.omics",
"account": "123456789012",
"time": "2015-12-22T18:43:48Z",
"region": "us-east-1",
```

```
"resources": ["arn:aws:omics:us-
east-1:123456789012:vincent_load_test_variant_store/
b64ea9a3-459f-4b68-92c3-3ddb83209fe9"],
   "detail": {
        "omicsVersion": "1.0.0",
        "arn": "arn:aws:omics:us-east-1:123456789012:vincent_load_test_variant_store/
b64ea9a3-459f-4b68-92c3-3ddb83209fe9",
        "status": "COMPLETED",
        "jobId": "b64ea9a3-459f-4b68-92c3-3ddb83209fe9",
        "storeId": "a74869f91e20",
        "storeName": "vincent_load_test_variant_store"
    }
}
```

For run events, two additional fields are included in the detail field. These are the uuid and runOutputUri, as shown in the following example.

```
{
    "version":"0",
    "id": "c0e540f4-df38-b986-86c1-3e3730f971fe",
    "detail-type": "Run Status Change",
    "source": "aws.omics",
    "account": "123456789012",
    "time":"2022-10-20T22:07:35Z",
    "region": "us-west-2",
    "resources":[
        "arn:aws:omics:us-west-2:123456789012:run/2101313"
    ],
    "detail":{
        "omicsVersion":"1.0.0",
        "arn": "arn: aws: omics: us-west-2:123456789012: run/2101313",
        "status": "COMPLETED",
        "uuid": "153893cd-097a-40ec-aec7-838a97cd2b21",
        "runOutputUri": "s3://integ-test-0ee27e7e/run-output/2101313"
    }
}
```

## Tagging resources in AWS HealthOmics

#### Important notice

AWS HealthOmics protects customer data under the AWS Shared Responsibility Model policies. This means that all customer data is encrypted both in transition and at-rest. However, not all customer-inputed names for resources such as data stores or job-based operations are encrypted. They should never contain Personally Identifiable Information or Protected Health Information. For more information, see Security in AWS HealthOmics.

#### **Tagging HealthOmics resources**

You can assign metadata to your AWS resources using *tags*. Each tag is a label consisting of a user-defined key and value. Tags can help you manage, identify, organize, search for, and filter resources.

This topic describes commonly used tagging categories and strategies to help you implement a consistent and effective tagging strategy. The following sections assume basic knowledge of AWS resources, tagging, detailed billing, and AWS Identity and Access Management (IAM).

Each tag has two parts:

- A tag key (for example, CostCenter, Environment, or Project). Tag keys are case sensitive.
- A *tag value* (for example, 111122223333 or Production). Like tag keys, tag values are case sensitive.

You can use tags to categorize resources by purpose, owner, environment, or other criteria. For more information, see AWS Tagging Strategies.

You can add, change, or remove tags one resource at a time from each resource's service console, service API, or the AWS CLI.

To enable tagging, make sure TagResources is authorized. You can authorize TagResources by attaching an IAM policy like the following example.

{

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```
"Version": "2012-10-17",
    "Statement": [
        {
            "Effect": "Allow",
            "Action": "omics:Create*",
            "Resource": "*"
        },
        }
            "Effect": "Allow",
            "Action": "omics:Start*",
            "Resource": "*"
        },
        {
            "Effect": "Allow",
            "Action": "omics:Tag*",
            "Resource": "*"
        },
        {
            "Effect": "Allow",
            "Action": "omics:Untag*",
            "Resource": "*"
        },
        {
            "Effect": "Allow",
            "Action": "omics:List*",
            "Resource": "*"
        }
    ]
}
```

### **Best practices**

As you create a tagging strategy for AWS resources, follow best practices:

- Do not store Personally Identifiable Information (PII), Protected Health Information(PHI) or other sensitive information in tags.
- Use a standardized, case-sensitive format for tags, and apply it consistently across all resource types.
- Consider tag guidelines that support multiple purposes, like managing resource access control, cost tracking, automation, and organization.

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Use automated tools to help manage resource tags. <u>AWS Resource Groups</u> and the <u>Resource Groups Tagging API</u> enable programmatic control of tags, making it possible to automatically manage, search, and filter tags and resources.

- Tagging is more effective when you use more tags.
- Tags can be edited or modified as user needs change. However to update access control tags, you must also update the policies that reference those tags to control access to your resources.

#### **Tagging requirements**

Tags have the following requirements:

- Keys can't be prefixed with aws:.
- Keys must be unique per tag set.
- A key must be between 1 and 128 allowed characters.
- A value must be between 0 and 256 allowed characters.
- Values do not need to be unique per tag set.
- Allowed characters for keys and values are Unicode letters, digits, white space, and any of the following symbols: \_ . : / = + - @.
- · Keys and values are case sensitive.

#### Adding a tag to an HealthOmics resource

Adding tags to a resource can help you identify and organize your AWS resources and manage access to them. First, you add one or more tags (key-value pairs) to a resource. You can use up to 50 tags per resource. There are also restrictions on the characters that you can use in the key and value fields.

After you add tags, you can create IAM policies to manage access to the AWS resource based on these tags. You can use the HealthOmics console or the AWS CLI to add tags to a resource. Adding tags to a repository can impact access to that repository. Before you add a tag to a data store, review any IAM policies that might use tags to control access to resources such as data stores.

Service tags are autogenerated for both a subject and a sample id for sequence stores.

Follow these steps to use the AWS CLI to add a tag to an HealthOmics resource. For example, to add tags to a sequence store while it is being created, you would use the following command in the

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AWS CLI. The name of the sequence store is MySequenceStore, and the two added tags with keys are key1 and key2 with values as value1 and value2 respectively

```
aws omics create-sequence-store --name "MySequenceStore" --tags key1=value1,key2=value2
```

The output does not list the tags. It returns the following response.

To add tags to an existing resource, you would run the following example command:

```
aws omics tag-resource --resource-arn arn:aws:omics:us-
west-2:555555555555sequenceStore/2275234794 --tags key1=value1,key2=value2
```

If successful, this command returns no response.

#### Listing tags for a resource

Follow these steps to use the AWS CLI to view a list of the AWS tags for an HealthOmics resource. If no tags have been added, the returned list is empty.

At the terminal or command line, run the list-tags-for-resource command as shown in the following example.

```
aws omics list-tags-for-resource --resource-arn arn:aws:omics:us-
west-2:5555555555555sequenceStore/2275234794
```

You will receive a list of tags in response, in JSON format.

```
{
    "tags": {
```

Listing tags for a resource 79

### Removing tags from a data store

You can remove one or more tags associated with a resource. Removing a tag does not delete the tag from other AWS resources that are associated with that tag.

At the terminal or command line, run the untag-resource command, specifying the Amazon Resource Name (ARN) of the resource where you want to remove tags and the tag key of the tag you want to remove.

```
aws omics untag-resource --resource-arn arn:aws:omics:us-
west-2:555555555555sequenceStore/2275234794 --tag-keys key1,key2
```

If successful, this command does not return a response. To verify the tags associated with the resource, run the list-tags-for-resource command.

# AWS HealthOmics and interface VPC endpoints (AWS PrivateLink)

You can establish a private connection between your VPC and AWS HealthOmics by creating an *interface VPC endpoint*. Interface endpoints are powered by <u>AWS PrivateLink</u>, a technology that you can use to privately access AWS HealthOmics API operations without an internet gateway, NAT device, VPN connection, or AWS Direct Connect connection. Instances in your VPC don't require public IP addresses to communicate with AWS HealthOmics API operations. Traffic between your VPC and AWS HealthOmics doesn't go outside the Amazon network.

Each interface endpoint is represented by one or more <u>Elastic Network Interfaces</u> in your subnets.

For more information, see <u>Interface VPC endpoints (AWS PrivateLink)</u> in the *Amazon VPC User Guide*.

VPC endpoint policies are supported for AWS HealthOmics for all Regions except Israel (Tel Aviv). By default, full access to AWS HealthOmics is allowed through the endpoint.

#### **Considerations for AWS HealthOmics VPC endpoints**

Before you set up an interface VPC endpoint for AWS HealthOmics, make sure that you review Interface endpoint properties and limitations in the *Amazon VPC User Guide*.

AWS HealthOmics supports making calls to all HealthOmics Storage API actions from your VPC.

VPC endpoint policies are not supported for AWS HealthOmics by default, but you can create a VPC endpoint for full AWS HealthOmics access for the HealthOmics Storage operations. For more information, see Controlling access to services with VPC endpoints in the Amazon VPC User Guide.

#### Creating an interface VPC endpoint for AWS HealthOmics

You can create a VPC endpoint for the AWS HealthOmics service by using the Amazon VPC console or the AWS Command Line Interface (AWS CLI). For more information, see <a href="Creating an interface">Creating an interface</a> endpoint in the Amazon VPC User Guide.

Create a VPC endpoint for AWS HealthOmics by using the following service names:

com.amazonaws.region.storage-omics

- com.amazonaws.*region*.control-storage-omics
- com.amazonaws.region.analytics-omics
- com.amazonaws.*region*.workflows-omics
- com.amazonaws.region.tags-omics

If you turn on private DNS for the endpoint, you can make API requests to AWS HealthOmics by using its default DNS name for the Region, for example, omics.us-east-1.amazonaws.com.

For more information, see <u>Accessing a service through an interface endpoint</u> in the *Amazon VPC User Guide*.

#### Creating a VPC endpoint policy for AWS HealthOmics

You can attach an endpoint policy to your VPC endpoint that controls access to AWS HealthOmics. The policy specifies the following information:

- The principal that can perform actions
- The actions that can be performed
- The resources on which actions can be performed

For more information, see <u>Controlling access to services with VPC endpoints</u> in the *Amazon VPC User Guide*.

#### **Example: VPC endpoint policy for AWS HealthOmics actions.**

The following is an example of an endpoint policy for AWS HealthOmics. When attached to an endpoint, this policy grants access to AWS HealthOmics actions for all principals on all resources.

API

```
"Resource":"*"
}
]
}
```

CLI

```
aws ec2 modify-vpc-endpoint \
    --vpc-endpoint-id vpce-id \
    --region us-west-2 \
    --policy-document \
    "{\"Statement\":[{\"Principal\":\"*\",\"Effect\":\"Allow\",\"Action\":
[\"omics:List*\"],\"Resource\":\"*\"}]}"
```

## Special considerations for accessing read sets using Amazon S3 URIs

To access read sets through Amazon S3 URIs when you're using a private connection, you must set up the PrivateLink interface endpoints on the sequence store. After you set them up, the endpoints will be in the format com.amazonaws.region.storage-omics and com.amazonaws.region.control-storage-omics.

Follow the guide <u>Gateway endpoints for Amazon S3</u> to configure your gateway endpoints. The Amazon S3 bucket is owned by HealthOmics, so you don't have to create or adjust the bucket policy. Gateway endpoints rely on the policy attached to the user or role that accesses the data, but you can also configure endpoints with more restrictive policies. These policies can include restrictions on access based on the Amazon S3 Access Point ARN and Amazon S3 actions.

## **HealthOmics Storage**

Use HealthOmics Storage to store, retrieve, organize, and share genomics data efficiently and at low cost. HealthOmics Storage understands the relationships between different data objects, so that you can define which read sets originated from the same source data. This provides you with data provenance.

Data that's stored in ACTIVE state is retrievable immediately. Data that hasn't been accessed for 30 days or more is stored in ARCHIVE state. To access archived data, you can reactivate it through the API operations or console.

With the HealthOmics Storage API operations, you can perform the following actions:

- Create, manage, and delete sequence and reference stores
- Create and manage read sets
- Import, export, and work with read sets
- Share and access read sets with collaborators through Amazon S3 URI access
- Create, manage, and import references
- Copy read sets to local file systems for analysis
- Tag AWS resources such as sequence stores, read sets, and references
- List and read files through Amazon S3 API operations by using the Amazon S3 URI

HealthOmics sequence stores are designed to preserve the content integrity of files. However, bitwise equivalence of imported data files and exported files isn't preserved because of the compression during active and archive tiering.

During ingestion, HealthOmics generates an entity tag, or *HealthOmics ETag*, to make it possible to validate the content integrity of your data files. Sequencing portions are identified and captured as an ETag at the source level of a read set. The ETag calculation doesn't alter the actual file or genomic data. After a read set is created, the ETag should not change throughout the lifecycle of the read set source. This means that reimporting the same file will result in the same ETag value being calculated.

#### ETag calculation and data provenance

An AWS HealthOmics entity tag or HealthOmics ETag is a hash of the ingested content in a sequence store. This simplifies data retrieval and processing while maintaining the content integrity of the ingested data files. The ETag reflects changes to the semantic content of the object, not its metadata. The specified read set type and algorithm determine how the ETag is calculated. The ETag calculation doesn't alter the actual file or genomic data. When the file type schema of the read set permits it, the sequence store updates fields that are linked to data provenance.

Files have a bitwise identity and a semantic identity. The bitwise identity means that the bits of a file are identical, and a semantic identity means that the contents of a file are identical. Semantic identity is resilient to metadata changes and compression changes as it captures the content integrity of the file.

Read sets in AWS HealthOmics sequence stores undergo compression/decompression cycles and data provenance tracking throughout an object's lifecycle. During this processing, the bitwise identity of an ingested file may change and is expected to change each time a file is activated; however, the semantic identity of the file is maintained. The semantic identity is captured as a HealthOmics entity tag, or ETag that is calculated during sequence store ingestion and available as read set metadata.

An AWS HealthOmics entity tag or HealthOmics ETag is a hash of the ingested content's semantic identity in a sequence store. This simplifies data retrieval and processing, while maintaining the content integrity of the ingested data files. The ETag reflects changes to the semantic content of the object, not its metadata. The specified read set type and algorithm determine how the ETag is calculated. The ETag calculation doesn't alter the actual file or genomic data.

When the file type schema of the read set permits it, the sequence store updates fields that are linked to data provenance. For uBAM, BAM, and CRAM files, a new @CO or Comment tag is added to the header. The comment contains the sequence store ID and ingestion timestamp.

When accessing a file using the Amazon S3 URI, Amazon S3 API operations may also return checksum and Amazon S3 ETag values. The Amazon S3 ETag and checksum values differ from the HealthOmics ETags because they represent the file's bitwise identity. To learn more about descriptive metadata and Objects, see the Amazon S3 API Object documentation. These values can change with each activation cycle of a read set and can be used to validate the reading of a file. However, they should not be cached and used as a file identity validation throughout the file's

lifecycle because they do not remain consistent. In contrast, the AWS HealthOmics ETag remains consistent through the read set's lifecycle.

#### How ETags are calculated

The ETag is generated from a hash of the ingested file contents. The ETag algorithm family is set to MD5up by default, but it can be configured differently during sequence store creation. When the ETag is calculated, the algorithm and the calculated hashes are added to the read set. The supported MD5 algorithms for file types are as follows.

- FASTQ\_MD5up Calculates the MD5 hash of an uncompressed, complete FASTQ read set source.
- BAM MD5up Calculates the MD5 hash of the alignment section of an uncompressed BAM or uBAM read set source as represented in the SAM, based on the linked reference, if one is available.
- CRAM\_MD5up Calculates the MD5 hash of the alignment section of the uncompressed CRAM read set source as represented in the SAM, based on the linked reference.

#### Note

MD5 hashing is known to be vulnerable to collisions. Because of this, two different files might have the same ETag if they were manufactured to exploit the known collision.

The following algorithms are supported for the SHA256 family. The algorithms are calculated as follows:

- FASTQ SHA256up Calculates the SHA-256 hash of an uncompressed, complete FASTQ read set source.
- BAM\_SHA256up Calculates the SHA-256 hash of the alignment section of an uncompressed BAM or uBAM read set source as represented in the SAM, based on the linked reference, if one is available.
- CRAM SHA256up Calculates the SHA-256 hash of the alignment section of an uncompressed CRAM read set source as represented in the SAM, based on the linked reference.

The following algorithms are supported for the SHA512 family. The algorithms are calculated as follows:

How ETags are calculated

• FASTQ\_SHA512up – Calculates the SHA-512 hash of an uncompressed, complete FASTQ read set source.

- BAM\_SHA512up Calculates the SHA-512 hash of the alignment section of an uncompressed BAM or uBAM read set source as represented in the SAM, based on the linked reference, if one is available.
- CRAM\_SHA512up Calculates the SHA-512 hash of the alignment section of an uncompressed CRAM read set source as represented in the SAM, based on the linked reference.

#### **Creating reference and sequence stores**

Reference and sequence stores are AWS resources that you can use to manage your genomic data through the API, AWS CLI, and console. The first step is for you to create a reference store to hold reference genomes that are used to map your read sets.

#### **Creating and managing reference stores**

The following example shows you how to create a reference store by using the AWS CLI. You can have one reference store per AWS Region. Reference stores support storage of FASTA files with the extensions .fasta, .fa, .fas, .fsa, .faa, .fna, .ffn, .frn, .mpfa, .seq, .txt. The bgzip version of these extensions is also supported. In the following example, replace reference store name with the name you've chosen for your reference store.

```
aws omics create-reference-store --name "reference store name"
```

You receive a JSON response with the reference store ID and name, the ARN, and the timestamp of when your reference store was created.

You can use the reference store ID in additional AWS CLI commands. You can retrieve the list of reference store IDs linked to your account by using the **list-reference-stores** command, as shown in the following example.

```
aws omics list-reference-stores
```

In response, you receive the name of your newly created reference store.

After you create a reference store, you can create import jobs to load genomic reference files into it. To do so, you must use or create an IAM role to access the data. The following is an example policy.

```
{
    "Version": "2012-10-17",
    "Statement": [
        {
             "Effect": "Allow",
             "Action": [
                 "s3:GetObject",
                 "s3:GetBucketLocation"
            ],
             "Resource": [
                 "arn:aws:s3:::DOC-EXAMPLE-BUCKET1",
                 "arn:aws:s3:::DOC-EXAMPLE-BUCKET1/*"
            ]
         }
      ]
   }
}
```

You must also have a trust policy similar to the following example.

```
{
```

You can now import a reference genome. This example uses Genome Reference Consortium Human Build 38 (hg38), which is open access and available from the <u>Registry of Open Data on AWS</u>. The bucket that hosts this data is based in US East (Ohio). To use buckets in other AWS Regions, you can copy the data to an Amazon S3 bucket hosted in your Region. Use the following AWS CLI command to copy the genome to your Amazon S3 bucket.

```
aws s3 cp s3://broad-references/hg38/v0/Homo_sapiens_assembly38.fasta s3://DOC-EXAMPLE-BUCKET
```

You can then begin your import job. Replace *reference store ID*, *role ARN*, and *source file path* with your own input.

```
aws omics start-reference-import-job --reference-store-id reference store ID --role-
arn role ARN --sources source file path
```

After the data is imported, you receive the following response in JSON.

```
{
    "id": "7252016478",
    "referenceStoreId": "3242349265",
    "roleArn": "arn:aws:iam::111122223333:role/OmicsReferenceImport",
    "status": "CREATED",
    "creationTime": "2022-07-01T21:15:13.727Z"
}
```

You can monitor the status of a job by using the following command. In the following example, replace *reference store ID* and *job ID* with your reference store ID and the job ID that you want to learn more about.

```
aws omics get-reference-import-job --reference-store-id reference store ID --id job ID
```

In response, you receive a response with the details for that reference store and its status.

You can also find the reference that was imported by listing your references and filtering them based on the reference name. Replace *reference store ID* with your reference store ID, and add an optional filter to narrow the list.

```
aws omics list-references --reference-store-id reference store ID --filter name=MyReference
```

In response, you receive the following information.

To learn more about the reference metadata, use the **get-reference-metadata** API operation. In the following example, replace **reference store ID** with your reference store ID and **reference ID** with the reference ID that you want to learn more about.

```
aws omics get-reference-metadata --reference-store-id reference store ID --id reference ID
```

You receive the following information in response.

```
{
    "id": "1234567890",
    "arn": "arn:aws:omics:us-west-2:5555555555555:referenceStore/referencestoreID/
reference/referenceID",
    "referenceStoreId": "1234567890",
    "md5": "7ff134953dcca8c8997453bbb80b6b5e",
    "status": "ACTIVE",
    "name": "MyReference",
    "creationTime": "2022-07-02T00:15:19.787Z",
    "updateTime": "2022-07-02T00:15:19.787Z",
    "files": {
        "source": {
            "totalParts": 31,
            "partSize": 104857600,
            "contentLength": 3249912778
        },
        "index": {
            "totalParts": 1,
            "partSize": 104857600,
            "contentLength": 160928
        }
    }
}
```

You can also download parts of the reference file by using **get-reference**. In the following example, replace **reference store ID** with your reference store ID and **reference ID** with the reference ID that you want to download from.

```
aws omics get-reference --reference-store-id reference store ID --id reference ID -- part-number 1 outfile.fa
```

#### **Creating and managing sequence stores**

HealthOmics sequence stores support storage of genomic files in the unaligned formats of FASTQ (gzip-only) and uBAM. It also supports the aligned formats of BAM and CRAM. Imported files are stored as read sets, which are an AWS resource. This means that you can add tags and control access through IAM. Aligned read sets require a reference genome to align genomic sequences, but it's optional for unaligned read sets.

To store read sets, you first create a sequence store. When you create a sequence store, you can specify an optional Amazon S3 bucket as a fallback location. The fallback location is used for storing any files that fail to create a read set during a direct upload. Fallback locations are available for sequence stores created after May 15, 2023. You specify the fallback location when you create the sequence store. You can't add a fallback location after the sequence store is created.

In the following example, replace *sequence store name* with the name you chose for your sequence store.

```
aws omics create-sequence-store --name sequence store name --fallback-location "s3://DOC-EXAMPLE-BUCKET"
```

You receive the following response in JSON, which includes the ID number for your newly created sequence store.

```
{
    "id": "3936421177",
    "arn": "arn:aws:omics:us-west-2:111122223333:sequenceStore/3936421177",
    "name": "sequence_store_example_name",
    "creationTime": "2022-07-13T20:09:26.038Z"
    "fallbackLocation" : "s3://DOC-EXAMPLE-BUCKET"
}
```

You can also view all sequence stores associated with your account by using the **list-sequence-stores** command, as shown in the following.

```
aws omics list-sequence-stores
```

You receive the following response.

Additionally, you can use **get-sequence-store** to learn more about a sequence store by using its ID, as shown in the following.

```
aws omics get-sequence-store --id sequence store ID
```

```
"arn": "arn:aws:omics:us-west-2:123456789012:sequenceStore/sequencestoreID",
  "creationTime": "2024-01-12T04:45:29.857Z",
  "description": null,
  "fallbackLocation": null,
  "id": "2015356892",
  "name": "MySequenceStore",
  "s3Access": {
    "s3AccessPointArn": "arn:aws:s3:us-
west-2:123456789012:accesspoint/592761533288-2015356892",
    "s3Uri": "s3://592761533288-2015356892-ajdpi90jdas90a79fh9a8ja98jdfa9jf98-
s3alias/592761533288/sequenceStore/2015356892/"
  },
  "sseConfig": {
    "keyArn": "arn:aws:kms:us-west-2:123456789012:key/eb2b30f5-635d-4b6d-b0f9-
d3889fe0e648",
    "type": "KMS"
```

```
}
```

#### **Deleting reference and sequence stores**

Both reference and sequence stores can be deleted. Sequence stores can only be deleted if they don't contain read sets, and reference stores can only be deleted if they don't contain references. Deleting a sequence or reference store also deletes any tags associated with that store.

The following example shows how to delete a reference store by using the AWS CLI. If the action is successful, you won't receive a response. In the following example, replace *reference store ID* with your reference store ID.

```
aws omics delete-reference-store --id reference store ID
```

The following example shows you how to delete a sequence store. You don't receive a response if the action succeeds. In the following example, replace *sequence store ID* with your sequence store ID.

```
aws omics delete-sequence-store --id sequence store ID
```

You can also delete a reference in a reference store, as shown in the following example. References can only be deleted if they are not being used in a read set, variant store, or annotation store. In the following example, replace *reference store ID* with your reference store ID, and replace *reference ID* with the ID for the reference you want to delete.

```
aws omics delete-reference --id reference ID --reference-store-id reference store ID
```

#### **Sequence store imports**

After you create your sequence store, you can create import jobs to load files into the data store. You can upload your files from an Amazon S3 bucket, or you can upload directly by using the synchronous API operations. Your S3 bucket must be in the same Region as your sequence store. You can use a command similar to the following to move files into your S3 bucket.

You can upload any combination of aligned and unaligned read sets into your sequence store, however, if any of the read sets in your import are aligned, you must include a reference genome.

```
aws s3 cp s3://1000genomes/phase1/data/HG00100/alignment/
HG00100.chrom20.ILLUMINA.bwa.GBR.low_coverage.20101123.bam s3://your-bucket
aws s3 cp s3://1000genomes/phase3/data/HG00146/sequence_read/SRR233106_1.filt.fastq.gz
s3://your-bucket
aws s3 cp s3://1000genomes/phase3/data/HG00146/sequence_read/SRR233106_2.filt.fastq.gz
s3://your-bucket
aws s3 cp s3://1000genomes/data/HG00096/alignment/
HG00096.alt_bwamem_GRCh38DH.20150718.GBR.low_coverage.cram s3://your-bucket
aws s3 cp s3://gatk-test-data/wgs_ubam/NA12878_20k/NA12878_A.bam s3://your-bucket
```

The sample BAM and CRAM used in this example require different genome references, Hg19 and Hg38. To learn more or to access these references, see <u>The Broad Genome References</u> in the Registry of Open Data on AWS.

You can reuse the IAM access policy that you used to create the Reference store. You must also create a manifest file in JSON to model the import job in import.json (see the following example). If you are creating a sequence store in the console, you don't have to specify the sequenceStoreId or roleARN, so your manifest file will start with the sources input.

#### **API** manifest

This example code is used to import three read sets by using the API: one FASTQ, one BAM, and one CRAM.

```
"description": "BAM for HG00100",
            "generatedFrom": "1000 Genomes"
        },
        {
            "sourceFiles":
            {
                "source1": "s3://DOC-EXAMPLE-BUCKET/SRR233106_1.filt.fastq.gz",
                "source2": "s3://DOC-EXAMPLE-BUCKET/SRR233106_2.filt.fastq.gz"
            },
            "sourceFileType": "FASTQ",
            "subjectId": "mySubject",
            "sampleId": "mySample",
            // NOTE: there is no reference arn required here
            "name": "HG00146",
            "description": "FASTQ for HG00146",
            "generatedFrom": "1000 Genomes"
        },
        {
            "sourceFiles":
            {
                "source1": "s3://DOC-EXAMPLE-BUCKET/
HG00096.alt_bwamem_GRCh38DH.20150718.GBR.low_coverage.cram"
            },
            "sourceFileType": "CRAM",
            "subjectId": "mySubject",
            "sampleId": "mySample",
            "referenceArn": "arn:aws:omics:us-
west-2:555555555555:referenceStore/0123456789/reference/0000000001",
            "name": "HG00096",
            "description": "CRAM for HG00096",
            "generatedFrom": "1000 Genomes"
        },
        {
            "sourceFiles":
                "source1": "s3://DOC-EXAMPLE-BUCKET/NA12878_A.bam"
            },
            "sourceFileType": "UBAM",
            "subjectId": "mySubject",
            "sampleId": "mySample",
            // NOTE: there is no reference arn required here
            "name": "NA12878_A",
            "description": "uBAM for NA12878",
            "generatedFrom": "GATK Test Data"
```

```
}
}
```

#### Console manifest

This example code is used to import a single read set by using the console.

```
Г
    {
        "sourceFiles":
            "source1": "s3://DOC-EXAMPLE-BUCKET/
HG00100.chrom20.ILLUMINA.bwa.GBR.low_coverage.20101123.bam"
        "sourceFileType": "BAM",
        "subjectId": "mySubject",
        "sampleId": "mySample",
        "name": "HG00100",
        "description": "BAM for HG00100",
        "generatedFrom": "1000 Genomes"
    },
    {
        "sourceFiles":
        {
            "source1": "s3://DOC-EXAMPLE-BUCKET/SRR233106_1.filt.fastq.gz",
            "source2": "s3://DOC-EXAMPLE-BUCKET/SRR233106_2.filt.fastq.qz"
        },
        "sourceFileType": "FASTQ",
        "subjectId": "mySubject",
        "sampleId": "mySample",
        "name": "HG00146",
        "description": "FASTQ for HG00146",
        "generatedFrom": "1000 Genomes"
    },
    {
        "sourceFiles":
        {
            "source1": "s3://your-bucket/
HG00096.alt_bwamem_GRCh38DH.20150718.GBR.low_coverage.cram"
        },
        "sourceFileType": "CRAM",
        "subjectId": "mySubject",
        "sampleId": "mySample",
```

```
"name": "HG00096",
        "description": "CRAM for HG00096",
        "generatedFrom": "1000 Genomes"
    },
    {
        "sourceFiles":
        {
            "source1": "s3://DOC-EXAMPLE-BUCKET/NA12878_A.bam"
        },
        "sourceFileType": "UBAM",
        "subjectId": "mySubject",
        "sampleId": "mySample",
        "name": "NA12878_A",
        "description": "uBAM for NA12878",
        "generatedFrom": "GATK Test Data"
    }
]
```

Alternatively, you can upload the manifest file in YAML format.

To start the import job, use the following AWS CLI command.

```
aws omics start-read-set-import-job --cli-input-json file://import.json
```

You receive the following response, which indicates successful job creation.

```
{
    "id": "3660451514",
    "sequenceStoreId": "3936421177",
    "roleArn": "arn:aws:iam::111122223333:role/OmicsImport",
    "status": "CREATED",
    "creationTime": "2022-07-13T22:14:59.309Z"
}
```

After the import job starts, you can monitor its progress with the following command. In the following example, replace *sequence store id* with your sequence store ID, and replace *job import ID* with the import ID.

```
aws omics get-read-set-import-job --sequence-store-id sequence store id --id job import ID
```

The following shows the statuses for all import jobs associated with the specified sequence store ID.

```
{
    "id": "1234567890",
    "sequenceStoreId": "1234567890",
    "roleArn": "arn:aws:iam::111122223333:role/OmicsImport",
    "status": "RUNNING",
    "statusMessage": "The job is currently in progress."
    "creationTime": "2022-07-13T22:14:59.309Z",
    "sources": [
        {
            "sourceFiles":
            {
                "source1": "s3://DOC-EXAMPLE-BUCKET/
HG00100.chrom20.ILLUMINA.bwa.GBR.low_coverage.20101123.bam"
            },
            "sourceFileType": "BAM",
            "status": "IN_PROGRESS",
            "statusMessage": "The job is currently in progress."
            "subjectId": "mySubject",
            "sampleId": "mySample",
            "referenceArn": "arn:aws:omics:us-
west-2:111122223333:referenceStore/3242349265/reference/8625408453",
            "name": "HG00100",
            "description": "BAM for HG00100",
            "generatedFrom": "1000 Genomes"
        },
        {
            "sourceFiles":
            {
                "source1": "s3://DOC-EXAMPLE-BUCKET/SRR233106_1.filt.fastq.qz",
                "source2": "s3://DOC-EXAMPLE-BUCKET/SRR233106_2.filt.fastq.gz"
            },
            "sourceFileType": "FASTQ",
            "status": "IN_PROGRESS",
            "statusMessage": "The job is currently in progress."
            "subjectId": "mySubject",
            "sampleId": "mySample",
            "name": "HG00146",
            "description": "FASTQ for HG00146",
            "generatedFrom": "1000 Genomes"
        },
```

```
{
            "sourceFiles":
                "source1": "s3://DOC-EXAMPLE-BUCKET/
HG00096.alt_bwamem_GRCh38DH.20150718.GBR.low_coverage.cram"
            },
            "sourceFileType": "CRAM",
            "status": "IN_PROGRESS",
            "statusMessage": "The job is currently in progress."
            "subjectId": "mySubject",
            "sampleId": "mySample",
            "referenceArn": "arn:aws:omics:us-
west-2:111122223333:referenceStore/3242349265/reference/1234568870",
            "name": "HG00096",
            "description": "CRAM for HG00096",
            "generatedFrom": "1000 Genomes"
        },
        {
            "sourceFiles":
            {
                "source1": "s3://DOC-EXAMPLE-BUCKET/NA12878_A.bam"
            },
            "sourceFileType": "UBAM",
            "status": "IN_PROGRESS",
            "statusMessage": "The job is currently in progress."
            "subjectId": "mySubject",
            "sampleId": "mySample",
            "name": "NA12878_A",
            "description": "uBAM for NA12878",
            "generatedFrom": "GATK Test Data"
        }
    ]
}
```

After the job completes, you can use the **list-read-sets** API operation to find the imported sequence files. In the following example, replace *sequence store id* with your sequence store ID.

```
aws omics list-read-sets --sequence-store-id sequence store id
```

You receive the following response.

```
{
    "readSets": [
        {
            "id": "0000000001",
            "arn": "arn:aws:omics:us-west-2:111122223333:sequenceStore/01234567890/
readSet/0000000001",
            "sequenceStoreId": "1234567890",
            "subjectId": "mySubject",
            "sampleId": "mySample",
            "status": "ACTIVE",
            "name": "HG00100",
            "description": "BAM for HG00100",
            "referenceArn": "arn:aws:omics:us-
west-2:111122223333:referenceStore/01234567890/reference/0000000001",
            "fileType": "BAM",
            "sequenceInformation": {
                "totalReadCount": 9194,
                "totalBaseCount": 928594,
                "generatedFrom": "1000 Genomes",
                "alignment": "ALIGNED"
            },
            "creationTime": "2022-07-13T23:25:20Z"
            "creationType": "IMPORT",
            "etag": {
                "algorithm": "BAM_MD5up",
                "source1": "d1d65429212d61d115bb19f510d4bd02"
            }
        },
        {
            "id": "00000000002",
            "arn": "arn:aws:omics:us-west-2:111122223333:sequenceStore/0123456789/
readSet/0000000002",
            "sequenceStoreId": "0123456789",
            "subjectId": "mySubject",
            "sampleId": "mySample",
            "status": "ACTIVE",
            "name": "HG00146",
            "description": "FASTQ for HG00146",
            "fileType": "FASTQ",
            "sequenceInformation": {
                "totalReadCount": 8000000,
                "totalBaseCount": 1184000000,
                "generatedFrom": "1000 Genomes",
```

```
"alignment": "UNALIGNED"
            },
            "creationTime": "2022-07-13T23:26:43Z"
            "creationType": "IMPORT",
            "etag": {
                "algorithm": "FASTQ_MD5up",
                "source1": "ca78f685c26e7cc2bf3e28e3ec4d49cd"
            }
        },
            "id": "0000000003",
            "arn": "arn:aws:omics:us-west-2:111122223333:sequenceStore/0123456789/
readSet/0000000003",
            "sequenceStoreId": "0123456789",
            "subjectId": "mySubject",
            "sampleId": "mySample",
            "status": "ACTIVE",
            "name": "HG00096",
            "description": "CRAM for HG00096",
            "referenceArn": "arn:aws:omics:us-
west-2:111122223333:referenceStore/0123456789/reference/0000000001",
            "fileType": "CRAM",
            "sequenceInformation": {
                "totalReadCount": 85466534,
                "totalBaseCount": 24000004881,
                "generatedFrom": "1000 Genomes",
                "alignment": "ALIGNED"
            },
            "creationTime": "2022-07-13T23:30:41Z"
            "creationType": "IMPORT",
            "etag": {
                "algorithm": "CRAM_MD5up",
                "source1": "66817940f3025a760e6da4652f3e927e"
            }
        },
            "id": "0000000004",
            "arn": "arn:aws:omics:us-west-2:111122223333:sequenceStore/0123456789/
readSet/0000000004",
            "sequenceStoreId": "0123456789",
            "subjectId": "mySubject",
            "sampleId": "mySample",
            "status": "ACTIVE",
            "name": "NA12878_A",
```

```
"description": "uBAM for NA12878",
            "fileType": "UBAM",
            "sequenceInformation": {
                "totalReadCount": 20000,
                "totalBaseCount": 5000000,
                "generatedFrom": "GATK Test Data",
                "alignment": "ALIGNED"
            },
            "creationTime": "2022-07-13T23:30:41Z"
            "creationType": "IMPORT",
            "etag": {
                "algorithm": "BAM_MD5up",
                "source1": "640eb686263e9f63bcda12c35b84f5c7"
            }
        }
    ]
}
```

To view more details about a given read set, use the **get-read-set-metadata** API operation. In the following example, replace **sequence store id** with your sequence store ID, and replace **read set id** with your read set ID.

```
aws omics get-read-set-metadata --sequence-store-id sequence store id --id read set id
```

You receive the following response.

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```
"contentLength": 10818,
      "partSize": 104857600,
      "s3Access": {
        "s3Uri": "s3://accountID-sequence store ID-ajdpi90jdas90a79fh9a8ja98jdfa9jf98-
s3alias/592761533288/sequenceStore/2015356892/readSet/9515444019/
import_source1.fastq.qz"
},
      "totalParts": 1
    },
    "source2": {
      "contentLength": 10818,
      "partSize": 104857600,
      "s3Access": {
        "s3Uri": "s3://accountID-sequence store ID-ajdpi90jdas90a79fh9a8ja98jdfa9jf98-
s3alias/592761533288/sequenceStore/2015356892/readSet/9515444019/
import_source1.fastq.qz"
      },
      "totalParts": 1
    }
  },
  "id": "9515444019",
  "name": "paired-fastq-import",
  "sampleId": "sampleId-paired-fastq-import",
  "sequenceInformation": {
    "alignment": "UNALIGNED",
    "generatedFrom": null,
    "totalBaseCount": 30000,
    "totalReadCount": 200
  },
  "sequenceStoreId": "2015356892",
  "status": "ACTIVE",
  "statusMessage": null,
  "subjectId": "subjectId-paired-fastq-import"
}
```

You can use **get-read-set** to download in parallel by downloading individual parts. These parts are similar to Amazon S3 parts. The following is an example of how to download part 1 from a read set. In the following example, replace *sequence store id* with your sequence store ID, and replace *read set id* with your read set ID.

```
aws omics get-read-set --sequence-store-id sequence store id --id read set id --part-number 1 outfile.bam
```

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You can also use the HealthOmics Transfer Manager to download files for a HealthOmics reference or read set. You can download the HealthOmics Transfer Manager <a href="https://example.com/here-based-set-bas

#### Direct upload to a sequence store

The HealthOmics Transfer Manager is recommended for adding files to your sequence store. You can also upload your read sets directly to a sequence store through the direct upload API operations.

Direct upload read sets exist first in PROCESSING\_UPLOAD state. This means that the file parts are currently being uploaded, and you can access the read set metadata. After the parts are uploaded and the checksums are validated, the read set becomes ACTIVE and behaves the same as an imported read set.

If the direct upload fails, the read set status is shown as UPLOAD\_FAILED. You can configure an Amazon S3 bucket as a fallback location for any files that fail to upload. The file parts for those read sets are transferred to the fallback location. Fallback locations are available on sequence stores created after May 15, 2023. You must also have an IAM policy that grants you read access to that Amazon S3 location.

To begin, start a multipart upload. You can do this by using the AWS CLI, as shown in the following example.

First, you create the parts by separating your data, as shown in the following example.

```
split -b 100MiB SRR233106_1.filt.fastq.gz source1_part_
```

After your source files are in parts, you can then create a multipart read set upload, as shown in the following. In the following example, replace *sequence store ID* and the other parameters with your sequence store ID and other values.

```
aws omics create-multipart-read-set-upload \
    --sequence-store-id sequence store ID \
    --name upload name \
    --source-file-type FASTQ \
    --subject-id subject ID \
    --sample-id sample ID \
    --description "FASTQ for HG00146" "description of upload" \
```

```
--generated-from "1000 Genomes""source of imported files"
```

In the response, you will get the uploadID and other metadata. Use the uploadID for the next step of the upload process.

```
{
    "sequenceStoreId": "1504776472",
    "uploadId": "7640892890",
    "sourceFileType": "FASTQ",
    "subjectId": "mySubject",
    "sampleId": "mySample",
    "generatedFrom": "1000 Genomes",
    "name": "HG00146",
    "description": "FASTQ for HG00146",
    "creationTime": "2023-11-20T23:40:47.437522+00:00"
}
```

Next, add your read sets to the upload. If your file is small enough, you only have to perform this step once. For larger files, you perform this step for each part of your file. If you upload a new part by using a previously used part number, it overwrites the previously uploaded part.

In the following example, replace *sequence store ID*, *upload ID*, and the other parameters with your values.

```
aws omics upload-read-set-part \
    --sequence-store-id sequence store ID \
    --upload-id upload ID \
    --part-source SOURCE1 \
    --part-number part number \
    --payload source1/source1_part_aa.fastq.gz
```

The response is an ID that you can use to check that the uploaded file matches the file you intended.

```
{
    "checksum": "984979b9928ae8d8622286c4a9cd8e99d964a22d59ed0f5722e1733eb280e635"
}
```

Continue uploading the parts of your file, if necessary. To check that your read sets have been uploaded, use the **list-read-set-upload-parts** API operation, as shown in the following. In the

following example, replace *sequence store ID*, *upload ID*, and the *part source* with your own input.

```
aws omics list-read-set-upload-parts \
    --sequence-store-id sequence store ID \
    --upload-id upload ID \
    --part-source SOURCE1
```

The response returns the number of read sets, the size, and the timestamp for when it was most recently updated.

```
{
    "parts": [
        {
            "partNumber": 1,
            "partSize": 104857600,
            "partSource": "SOURCE1",
            "checksum": "MVMQk+vB9C3Ge8ADHkbKq752n3BCUzy141qEkq10D5M=",
            "creationTime": "2023-11-20T23:58:03.500823+00:00",
            "lastUpdatedTime": "2023-11-20T23:58:03.500831+00:00"
        },
        {
            "partNumber": 2,
            "partSize": 104857600,
            "partSource": "SOURCE1",
            "checksum": "keZzVzJNChAqqOdZMvOmjBwrOPM0enPj1UAfs0nvRto=",
            "creationTime": "2023-11-21T00:02:03.813013+00:00",
            "lastUpdatedTime": "2023-11-21T00:02:03.813025+00:00"
        },
        {
            "partNumber": 3,
            "partSize": 100339539,
            "partSource": "SOURCE1",
            "checksum": "TBkNfMsaeDpXzEf3ldlbi0ipFDPaohKHyZ+LF1J4CHk=",
            "creationTime": "2023-11-21T00:09:11.705198+00:00",
            "lastUpdatedTime": "2023-11-21T00:09:11.705208+00:00"
        }
    ]
}
```

To view all active multipart read set uploads, use **list-multipart-read-set-uploads**, as shown in the following. Replace **sequence store ID** with the ID for your own sequence store.

```
aws omics list-multipart-read-set-uploads --sequence-store-id sequence store ID
```

This API only returns multipart read set uploads that are in progress. After the ingested read sets are ACTIVE, or if the upload has failed, the upload will not be returned in the response to the **list-multipart-read-set-uploads** API. To view active read sets, use the **list-read-sets** API. An example response for **list-multipart-read-set-uploads** is shown in the following.

```
{
    "uploads": [
        {
            "sequenceStoreId": "1234567890",
            "uploadId": "8749584421",
            "sourceFileType": "FASTQ",
            "subjectId": "mySubject",
            "sampleId": "mySample",
            "generatedFrom": "1000 Genomes",
            "name": "HG00146",
            "description": "FASTQ for HG00146",
            "creationTime": "2023-11-29T19:22:51.349298+00:00"
        },
        {
            "sequenceStoreId": "1234567890",
            "uploadId": "5290538638",
            "sourceFileType": "BAM",
            "subjectId": "mySubject",
            "sampleId": "mySample",
            "generatedFrom": "1000 Genomes",
            "referenceArn": "arn:aws:omics:us-
west-2:123456789012:referenceStore/8168613728/reference/2190697383",
            "name": "HG00146",
            "description": "BAM for HG00146",
            "creationTime": "2023-11-29T19:23:33.116516+00:00"
        },
        {
            "sequenceStoreId": "1234567890",
            "uploadId": "4174220862",
            "sourceFileType": "BAM",
            "subjectId": "mySubject",
            "sampleId": "mySample",
            "generatedFrom": "1000 Genomes",
```

After you upload all parts of your file, use **complete-multipart-read-set-upload** to conclude the upload process, as shown in the following. Replace **sequence store ID**, **upload ID**, and the parameter for parts with your own values.

```
aws omics complete-multipart-read-set-upload \
    --sequence-store-id sequence store ID \
    --upload-id upload ID \
    --parts '[{"checksum":"gaCBQMe+rpCFZxLpoP6gydBoXaKKDA/
    Vobh5zBDb4W4=","partNumber":1,"partSource":"SOURCE1"}]'
```

The response for **complete-multipart-read-set-upload** is the read set IDs for your imported read sets.

```
{
    "readSetId": "0000000001"
}
```

To stop the upload, use **abort-multipart-read-set-upload** with the upload ID to end the upload process. Replace **sequence store ID** and **upload ID** with your own parameter values.

```
aws omics abort-multipart-read-set-upload \
    --sequence-store-id sequence store ID \
    --upload-id upload ID
```

After the upload is complete, you can retrieve your data from the read set by using **get-read-set**, as shown in the following. If the upload is still processing, **get-read-set** returns limited metadata, and the generated index files are unavailable. Replace **sequence store ID** and the other parameters with your own input.

```
aws omics get-read-set
--sequence-store-id sequence store ID \
```

```
--id read set ID \
--file SOURCE1 \
--part-number 1 myfile.fastq.gz
```

To check the metadata, including the status of your upload, use the **get-read-set-metadata** API operation.

```
aws omics get-read-set-metadata --sequence-store-id sequence store ID --id read set ID
```

The response includes metadata details such as the file type, the reference ARN, the number of files, and the length of the sequences. It will also include the status. Possible statuses are PROCESSING\_UPLOAD, ACTIVE, and UPLOAD\_FAILED.

```
{
    "id": "0000000001",
    "arn": "arn:aws:omics:us-west-2:5555555555555:sequenceStore/0123456789/
readSet/0000000001",
    "sequenceStoreId": "0123456789",
    "subjectId": "mySubject",
    "sampleId": "mySample",
    "status": "PROCESSING_UPLOAD",
    "name": "HG00146",
    "description": "FASTQ for HG00146",
    "fileType": "FASTQ",
    "creationTime": "2022-07-13T23:25:20Z",
    "files": {
        "source1": {
            "totalParts": 5,
            "partSize": 123456789012,
            "contentLength": 6836725,
        },
        "source2": {
            "totalParts": 5,
            "partSize": 123456789056,
            "contentLength": 6836726
        }
    },
    'creationType": "UPLOAD"
}
```

If a read set creation fails, the files can be moved to a fallback Amazon S3 location. This way, you can keep the files in Amazon S3 to re-import after the files issues are resolved. The fallback location can be configured for a sequence store from the console, the AWS CLI, or the API.

### **Exporting read sets**

You can export read sets as a batch export job to an Amazon S3 bucket. To do so, first create an IAM policy that has write access to the bucket, similar to the following IAM policy example.

```
{
    "Version": "2012-10-17",
    "Statement": [
        {
            "Effect": "Allow",
            "Action": [
                "s3:PutObject",
                 "s3:GetBucketLocation"
             ],
             "Resource": [
                "arn:aws:s3:::DOC-EXAMPLE-BUCKET1",
                "arn:aws:s3:::DOC-EXAMPLE-BUCKET1/*"
             ]
        }
    ]
}
```

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After the IAM policy is in place, begin your read set export job. The following example shows you how to do this by using the **start-read-set-export-job** API operation. In the following example, replace all parameters, such as **sequence store ID**, **destination**, **role ARN**, and **sources**, with your input.

```
aws omics start-read-set-export-job
    --sequence-store-id sequence store id \
    --destination valid s3 uri \
    --role-arn role ARN \
    --sources readSetId=read set id_1 readSetId=read set id_2
```

You receive the following response with information on the origin sequence store and the destination S3 bucket.

```
{
    "id": <job-id>,
    "sequenceStoreId": <sequence-store-id>,
    "destination": <destination-s3-uri>,
    "status": "SUBMITTED",
    "creationTime": "2022-10-22T01:33:38.079000+00:00"
}
```

After the job starts, you can check on its status by using the **get-read-set-export-job** API operation, as shown in the following. Replace the **sequence store ID** and **job ID** with your sequence store ID and job ID, respectively.

```
aws omics get-read-set-export-job --id job-id --sequence-store-id sequence store ID
```

You can view all export jobs initialized for a sequence store by using the **list-read-set-export-jobs** API operation, as shown in the following. Replace the **sequence store ID** with your sequence store ID.

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In addition to exporting your read sets, you can also share them by using the Amazon S3 access URIs. To learn more, see Accessing and managing read sets with Amazon S3 URIs.

### Accessing and managing read sets with Amazon S3 URIs

You can use Amazon S3 URI paths to access your active sequence store read sets. Use Amazon S3 API operations to list, share, and download your read sets. This makes it possible for you to collaborate and share your data because you can share access across users in the data owner's account in the Region, or you can share through creating role users outside of the data owner's account that you can assume. Archived read sets are not accessible by using Amazon S3 URIs until they have been activated. When a read set is activated, it is restored to the same URI path each time.

With data loaded into HealthOmics stores, because the Amazon S3 URI is based on Amazon S3 access points, you can directly integrate with industry standard tools that read Amazon S3 URIs, such as the following:

- Visual analysis applications such as Integrative Genomics Viewer (IGV) or UCSC Genome Browser.
- Common workflows with Amazon S3 extensions such as CWL, WDL, and NextFlow.
- Any tool that can authenticate and read from access point Amazon S3 URIs or read presigned Amazon S3 URIs.
- Amazon S3 utilities such as Mountpoint or CloudFront.

Amazon S3 Mountpoint makes it possible for you to use an Amazon S3 bucket as a local file system. To learn more about Mountpoint and to install it for use, see Mountpoint for Amazon S3.

Amazon CloudFront is a content delivery network (CDN) service built for high performance, security, and developer convenience. To learn more about using Amazon CloudFront, seethe <a href="Mazon CloudFront"><u>Amazon CloudFront documentation</u></a>. To set up CloudFront with a sequence store, contact the AWS HealthOmics team.

The data owner root account is enabled for the actions S3:GetObject, S3:GetObjectTagging, and S3:List Bucket on the sequence store prefix. For a user in the account to access the data, you create an IAM policy and attach it to the user or role. For an example policy, see

When a sequence store is created, the creator's root account will automatically have S3:GetObject, S3GetObjectTagging, and S3:ListBucket access. Additionally, if the AWS KMS key on the sequence store is owned by HealthOmics, the root account is added with kms:Decrypt access.

For a user in the account to access the data, you create a policy and attach it to the user or role to allow access to the files using Amazon S3 API operations. To use HealthOmics API operations, you must add AWS HealthOmics permissions to your IAM policy. A policy allowing Amazon S3 API access can be applied at the sequence store level or at a read set level. At the read set level, permission can be restricted either through the prefix or using resource tag filters for sample or subject ID patterns.

The following example gives a user access to a sequence store. You can fine-tune the access with additional conditions or resource-based filters.

```
{
      "Version": "2012-10-17",
      "Statement": [
           {
                "Sid": "S3DirectAccess",
                "Effect": "Allow",
                "Action": [
                    "s3:GetObject",
                    "s3:ListBucket",
                    "s3:GetObjectTagging"
               ],
                "Resource": "*",
"Condition": {
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                                                                                                          114
                    "StringEquals": {
```

To learn more about using IAM policies with HealthOmics, see Service roles for AWS HealthOmics.

Depending on who you are sharing your data with, there are three ways you can use Amazon S3 URIs to share your data. The options are as follows:

- For sharing with users and roles within your account Write a user access policy that includes access to the AWS KMS key and access to the access point. This makes the data accessible for use with the Amazon S3 API operations.
- For sharing with users outside of your account Create a role within the data owner's account
  that has an access policy that allows the user to assume that role. Adding the user with direct
  access is not supported.
- Presigned URLs You can also generate a shareable URL for a file in the sequence store.
   To learn more about creating presigned URLs by using Amazon S3, see <u>Using presigned URL</u> in the Amazon S3 documentation.

You can use the following Amazon S3 API operations on the active read sets to list and retrieve your data. You can access archived read sets through Amazon S3 URIs after they have been activated.

- GetObject— Retrieves an object from Amazon S3.
- <u>HeadObject</u>— The HEAD operation retrieves metadata from an object without returning the object itself. This operation is useful if you only want an object's metadata.
- <u>ListObjects and ListObject v2</u>— Returns some or all (up to 1,000) of the objects in a bucket.
- <u>CopyObject</u>— Creates a copy of an object that is already stored in Amazon S3. HealthOmics supports copying to an Amazon S3 access point, but not writing to an access point.

Health Omics sequence stores maintain the semantic identity of files through ETags. Throughout 115 a lifecycle of a file, the Amazon S3 ETag, which is based on bitwise identity, may change, however,

the HealthOmics ETag will remain the same. To learn more, see ETag calculation and data provenance.

#### Amazon S3 URI structure in HealthOmics storage

All files with Amazon S3 URIs have omics:subjectId and omics:sampleId resource tags. You can use these tags to share access by using IAM policies through a pattern such as "s3:ExistingObjectTag/omics:subjectId": "pattern desired".

The file structure is as follows:

```
.../account_id/sequenceStore/seq_store_id/readSet/read_set_id/files.
```

For files imported into sequence stores from Amazon S3, the sequence store attempts to maintain the original source name. When the names conflict, the system appends read set information to ensure that the file names are unique. For instance, for fastq read sets, if both file names are the same, to make the names unique, sourceX is inserted before .fastq.gz or .fq.gz. For a direct upload, the file names follow the following patterns:

- For FASTQ— read\_set\_name\_sourcex.fastq.gz
- For uBAM/BAM/CRAM— read\_set\_name.file extension with extensions of .bam or .cram.
  An example is NA193948.bam.

For read sets that are BAM or CRAM, index files are automatically generated during the ingestion process. For the index files generated, the proper index extension at the end of the file name is applied. It has the pattern <name of the Source the index is on>.<file index extension>. The index extensions are .bai or .crai.

#### Using Hosted or Local IGV to access read sets

IGV is a genome browser used to analyze BAM and CRAM files. It requires both the file and the index because it only displays a portion of the genome at a time. IGV can be downloaded and used locally, and there are guides to creating an AWS hosted IGV. The public web version is not supported because it requires CORS.

Local IGV relies on the local AWS configuration to access files. Ensure that the role used in that configuration has a policy attached that enables kms:Decrypt and s3:GetObject permissions to the s3 URI of the read sets being accessed. After that, in IGV, you can use "File > load from URL" and paste in the URI for the source and index. Alternatively, presigned URLs can be generated and used

in the same manner, which will bypass the AWS configuration. Note that CORS is not supported with S3 URI access, so requests relying on CORS are not supported.

The example AWS Hosted IGV relies on AWS Cognito to create the correct configurations and permissions inside the environment. Ensure that a policy is created that enableskms:Decrypt and s3:GetObject permissions to the Amazon S3 URI of the read sets being accessed, and add this policy to the role that is assigned to the Cognito user pool. After that, in IGV, you can use "File > load from URL" and enter in the URI for the source and index. Alternatively, presigned URLs can be generated and used in the same manner, which will bypass the AWS configuration.

Note that the sequence store will not appear under the "Amazon" tab because that only displays buckets owned by you in the Region in which the AWS profile is configured.

#### **Using Samtools or HTSlib in AWS HealthOmics**

HTSlib is the core library that is shared by several commonly-used tools such as Samtools, rSamtools, PySam, and others. Use HTSlib version 1.20 or later to get seamless support for Amazon S3 Access Points. For older versions of the HTSlib library, you can use the following workarounds:

- Set the environment variable for the HTS S3 host with: export HTS\_S3\_HOST="s3.region.amazonaws.com".
- Generate a presigned URL for the files that you want to use. If a BAM or CRAM is being used, ensure that a presigned URL is generated for both the file and the index. After that, both files can be used with the libraries.
- Use Mountpoint to mount the sequence store or read set prefix in the same environment where you're using HTSlib libraries. From here, the files can be accessed by using local file paths.

### **Using Mountpoint AWS HealthOmics**

Mountpoint for Amazon S3 is a simple, high-throughput file client for mounting an Amazon S3 bucket as a local file system. With Mountpoint for Amazon S3, your applications can access objects stored in Amazon S3 through file operations such as open and read. Mountpoint for Amazon S3 automatically translates these operations into Amazon S3 object API calls, giving your applications access to the elastic storage and throughput of Amazon S3 through a file interface.

Mountpoint can be installed by using the Mountpoint installation instructions. Mountpoint uses the AWS Profile that's local to the installation and works at an Amazon S3 prefix level. Ensure that the profile being used has a policy that enables s3:GetObject, s3:ListBucket, and kms:Decrypt

permissions to the Amazon S3 URI prefix of the read set(s) or sequence store being accessed. After that, the bucket can be mounted by using the following path:

```
mount-s3 access point arn local path to mount --prefix prefix to sequence store or read set --region region
```

#### **Using CloudFront AWS HealthOmics**

Amazon CloudFront is a content delivery network (CDN) service that's built for high performance, security, and developer convenience. Customers that want to use CloudFront must work with the Service team to turn on the CloudFront distribution. Work with your account team to engage the AWS HealthOmics service team.

## **Activating read sets**

You can activate read sets that are archived with the **start-read-set-activation-job** API operation, or through the AWS CLI, as shown in the following example. Replace the **sequence store ID** and **read set id** with your sequence store ID and read set IDs.

```
aws omics start-read-set-activation-job
--sequence-store-id sequence store ID \
--sources readSetId=read set ID readSetId=read set id_1 read set id_2
```

You receive a response that contains the activation job information, as shown in the following.

```
{
    "id": "12345678",
    "sequenceStoreId": "1234567890",
    "status": "SUBMITTED",
    "creationTime": "2022-10-22T00:50:54.670000+00:00"
}
```

After the activation job starts, you can monitor its progress with the **get-read-set-activation-job** API operation. The following is an example of how to use the AWS CLI to check your activation job status. Replace *job ID* and *sequence store ID* with your sequence store ID and job IDs, respectively.

```
aws omics get-read-set-activation-job --id job ID --sequence-store-id sequence store ID
```

The response summarizes the activation job, as shown in the following.

```
{
    "id": 123567890,
    "sequenceStoreId": 123467890,
    "status": "SUBMITTED",
    "statusUpdateReason": "The job is submitted and will start soon.",
    "creationTime": "2022-10-22T00:50:54.670000+00:00",
    "sources": [
        {
            "readSetId": <reads set id_1>,
            "status": "NOT_STARTED",
            "statusUpdateReason": "The source is queued for the job."
        },
        {
            "readSetId": <read set id_2>,
            "status": "NOT_STARTED",
            "statusUpdateReason": "The source is queued for the job."
        }
    ]
}
```

You can check the status of an activation job with the **get-read-set-metadata** API operation. Possible statuses are ACTIVE, ACTIVATING, and ARCHIVED. In the following example, replace **sequence store ID** with your sequence store ID, and replace **read set ID** with your read set ID.

```
aws omics get-read-set-metadata --sequence-store-id sequence store ID --id read set ID
```

The following response shows you that the read set is active.

```
"creationTime": "2022-07-13T23:25:20Z",
    "sequenceInformation": {
        "totalReadCount": 1513467,
        "totalBaseCount": 163454436,
        "generatedFrom": "Pulled from SRA",
        "alignment": "ALIGNED"
    },
    "referenceArn": "arn:aws:omics:us-west-2:55555555555:referenceStore/0123456789/
reference/0000000001",
    "files": {
        "source1": {
            "totalParts": 2,
            "partSize": 10485760,
            "contentLength": 17112283,
            "s3Access": {
        "s3Uri": "s3://accountID-sequence store ID-ajdpi90jdas90a79fh9a8ja98jdfa9jf98-
s3alias/592761533288/sequenceStore/2015356892/readSet/9515444019/
import_source1.fastq.qz"
},
         },
        "index": {
            "totalParts": 1,
            "partSize": 53216,
            "contentLength": 10485760
            "s3Access": {
        "s3Uri": "s3://accountID-sequence store ID-ajdpi90jdas90a79fh9a8ja98jdfa9jf98-
s3alias/592761533288/sequenceStore/2015356892/readSet/9515444019/
import_source1.fastq.gz"
},
        }
    },
    "creationType": "IMPORT",
    "etag": {
        "algorithm": "BAM_MD5up",
        "source1": "d1d65429212d61d115bb19f510d4bd02"
    }
}
```

You can view all read set activation jobs by using **list-read-set-activation-jobs**, as shown in the following example. In the following example, replace **sequence store ID** with your sequence store ID.

```
aws omics list-read-set-activation-jobs --sequence-store-id sequence store ID
```

You receive the following response.

# **HealthOmics Analytics**

HealthOmics Analytics supports the storage and analysis of genomic variants and annotations.

With the variant and annotation store API operations, you can perform the following actions:

- Creating and managing variant stores
- Importing variant data and managing import jobs
- Creating and managing annotation stores
- Importing and managing annotation jobs
- Share analytic store data with collaborators
- Tagging AWS resources, such as variant stores and annotation stores

Variant stores support data in VCF formats, and annotation stores support TSV/CSV and GFF3 formats. When your data is in the HealthOmics Analytics data store, access to the VCF files is managed through AWS Lake Formation. You can then query the VCF files by using Amazon Athena. To be supported, queries must use Athena query engine version 3. To read more about Athena query engine versions, see the Amazon Athena documentation.

## Creating and managing variant stores

The following examples show how you can use the API operations to create and manage variant stores. You can also perform these operations with the AWS CLI.

In the following example, the AWS CLI is used to create a variant store.

```
aws omics create-variant-store --name myvariantstore \
   --reference referenceArn="arn:aws:omics:us-
west-2:555555555555:referenceStore/123456789/reference/5987565360"
```

To confirm the creation of your variant store, you will receive the following response.

```
{
    "creationTime": "2022-11-03T18:19:52.296368+00:00",
    "id": "45aeb91d5678",
    "name": "myvariantstore",
    "reference": {
```

To learn more about a variant store, use the **get-variant-store** API.

```
aws omics get-variant-store --name myvariantstore
```

You will receive the following response.

To view all variant stores associated with an account, use the list-variant-stores API.

```
aws omics list-variant-stores
```

You will receive a response that lists all variant stores, along with their IDs, statuses, and other details, as shown in the following example response.

You can also filter the responses for the list-variant-stores API based on statuses or other criteria.

VCF Files imported into analytic stores created on or after May 15, 2023 have defined schemas for Variant Effect Predictor (VEP) annotations. This makes it easier to query and parse imported VCF data. The change does not impact stores created before May 15, 2023, except if the annotation fields parameter is included in the API or CLI call. For these stores, using the annotation fields parameter will cause the request to fail.

## Import jobs and data parsing

The following example shows how to use the AWS CLI to create an import job.

For stores created after May 15, 2023, the following example shows how to add the -- annotation-fields parameter. The annotation fields are defined with the import.

```
aws omics start-variant-import-job \
    --destination-name annotationparsingvariantstore \
    --role-arn arn:aws:iam::123456789012:role/<role_name> \
    --items source=s3://pathToS3/sample.vcf
    --annotation-fields '{"VEP": "CSQ"}'
```

```
"jobId": "981e2286-e954-4391-8a97-09aefc343861"
}
```

Use **get-variant-import-job** to check the status.

```
aws omics get-variant-import-job --job-id 08279950-a9e3-4cc3-9a3c-a574f9c9e229
```

You'll receive a JSON response that shows the status of your import job. VEP annotations in the VCF are parsed for information stored in the INFO column as an ID/Value pair. The default ID for <a href="Ensembl Variant Effect Predictor">Ensembl Variant Effect Predictor</a> annotations INFO column is CSQ, but you can use the -- annotation-fields parameter to indicate a custom value used in the INFO column. Parsing is currently supported for VEP annotations.

For a store created before May 15, 2023 or for VCF files that do not include VEP annotation, no annotation fields will be included in the response.

```
"status": "COMPLETED",
"updateTime": "2023-04-11T17:58:22.676043+00:00",
}
```

The VEP annotations that are a part of VCF files are stored as predefined schema with the following structure. The extras field can be used to store any additional VEP fields that not included in the default schema.

```
annotations struct<
   vep: array<struct<
      allele:string,
      consequence: array<string>,
      impact:string,
      symbol:string,
      gene:string,
      `feature_type`: string,
      feature: string,
      biotype: string,
      exon: struct<rank:string, total:string>,
      intron: struct<rank:string, total:string>,
      hgvsc: string,
      hgvsp: string,
      `cdna_position`: string,
      `cds_position`: string,
      `protein_position`: string,
      `amino_acids`: struct<reference:string, variant: string>,
      codons: struct<reference:string, variant: string>,
      `existing_variation`: array<string>,
      distance: string,
      strand: string,
      flags: array<string>,
      symbol_source: string,
      hgnc_id: string,
      `extras`: map<string, string>
    >>
>
```

The parsing is performed with a best effort approach. If the VEP entry doesn't follow the <u>VEP</u> standard specifications, it won't be parsed and the row in the array will be empty.

For a new variant store, the response for **get-variant-import-job** would include the annotation fields, as shown.

```
aws omics get-variant-import-job --job-id 08279950-a9e3-4cc3-9a3c-a574f9c9e229
```

You'll receive a JSON response that shows the status of your import job.

```
{
    "creationTime": "2023-04-11T17:52:37.241958+00:00",
    "destinationName": "annotationparsingvariantstore",
    "id": "7a1c67e3-b7f9-434d-817b-9c571fd63bea",
    "items": [
    "jobStatus": "COMPLETED",
    "source": "s3://DOC-EXAMPLE-BUCKET/NA12878.2k.garvan.vcf"
    }
 ],
    "roleArn": "arn:aws:iam::123456789012:role/<role_name>",
    "runLeftNormalization": false,
    "status": "COMPLETED",
    "updateTime": "2023-04-11T17:58:22.676043+00:00",
    "annotationFields" : {"VEP": "CSQ"}
  }
}
```

You can use list-variant-import-jobs to see all import jobs and their statuses.

```
aws omics list-variant-import-jobs --ids 7a1c67e3-b7f9-434d-817b-9c571fd63bea
```

The response returned will have information as follows.

```
{
    "variantImportJobs": [
    {
        "creationTime": "2023-04-11T17:52:37.241958+00:00",
        "destinationName": "annotationparsingvariantstore",
        "id": "7a1c67e3-b7f9-434d-817b-9c571fd63bea",
        "roleArn": "arn:aws:iam::55555555555:role/roleName",
        "runLeftNormalization": false,
        "status": "COMPLETED",
        "updateTime": "2023-04-11T17:58:22.676043+00:00",
        "annotationFields": {"VEP": "CSQ"}
    }
}
```

```
]
}
}
```

If necessary, you can cancel an import job with the following command.

```
aws omics cancel-variant-import-job
--job-id edd7b8ce-xmpl-47e2-bc99-258cac95a508
```

### Performing queries on parsed annotation schemas

You can perform queries on your variant store using Amazon Athena. The query would look like the following. In this example, the table\_name is the name of the store, and database\_name is the resource link you've created.

```
SELECT
  "contigname",
  "start",
  "end",
  "referenceallele",
  "vepentries"."allele",
  "vepentries"."impact",
  "vepentries"."symbol",
  "vepentries"."consequence"
FROM
  "<database_name>"."<table_name>"
  CROSS JOIN
     UNNEST("annotations"."vep") AS t(vepentries)
where
  contains("vepentries"."consequence", 'stop_lost') limit 10
```

The response would be the following.

```
referenceallele
#
     contigname
                    start
                              end
                                                          allele
                                                                     impact
                                                                                symbol
 consequence
                                                                     [stop_lost]
1
     chr9
             7798436
                          7798437
                                             G
                                                   HIGH
                                                           DMAC1
2
                                             C
                                                                     [stop_lost,
     chr8
             19960936
                           19960937
                                                  HIGH
                                                           LPL
 NMD_transcript_variant]
3
     chr9
              83055544
                           83055545
                                        C
                                             Α
                                                  HIGH
                                                           RASEF
                                                                     [stop_lost]
                                                                     [stop_lost]
                                             C
4
     chr9
             83055546
                           83055547
                                                   HIGH
                                                           RASEF
```

5	chr9	82982676	82982677	Τ	Α	HIGH	RASEF	[stop_lost]
6	chr9	82982676	82982677	Т	Α	HIGH	RASEF	[stop_lost]
7	chr9	82982676	82982677	T	Α	HIGH	RASEF	[stop_lost]
8	chr9	82982676	82982677	Т	Α	HIGH	RASEF	[stop_lost]
9	chr9	26126010	26126011	Т	Α	HIGH	L0C10050	06422 [stop_lost]
10	chr8	30996593	30996594	Т	G	HIGH	PURG	[stop_lost]

## Creating and managing annotation store examples

An annotation store is a data store representing an annotation database, such as one from a TSV, VCF, or GFF file. If the same reference genome is specified, annotation stores are mapped to the same coordinate system as variant stores during an import. The following examples show how to use the AWS CLI to create and manage an annotation store.

In the following example, the AWS CLI is used to create an annotation store. For all AWS CLI and API operations, the format of your data must be declared.

To confirm the creation of your annotation store, you will receive the following response.

To learn more about an annotation store, use the **get-annotation-store** API.

```
aws omics get-annotation-store --name my_annotation_store
```

You will receive the following response.

```
{
    "id": "eeb019ac79c2",
    "reference": {
        "referenceArn": "arn:aws:omics:us-
west-2:5555555555555:referenceStore/5638433913/reference/5871590330"
    "status": "ACTIVE",
    "storeArn": "arn:aws:omics:us-west-2:55555555555:annotationStore/gffstore",
    "name": "my_annotation_store",
    "creationTime": "2022-11-05T00:05:19.136131+00:00",
    "updateTime": "2022-11-05T00:10:36.944839+00:00",
    "tags": {},
    "storeFormat": "GFF",
    "statusMessage": "",
    "storeSizeBytes": 0,
    "numVersions": 1
}
```

To view all annotation stores associated with an account, use the **list-annotation-stores** API operation.

```
aws omics list-annotation-stores
```

You will receive a response that lists all annotation stores, along with their IDs, statuses, and other details, as shown in the following example response.

}

You can also filter responses based on status or other criteria.

The following example shows how to use the AWS CLI to start an annotation import job.

```
aws omics start-annotation-import-job \
    --destination-name myannostore \
    --version-name myannostore \
    --role-arn arn:aws:iam::123456789012:role/roleName \
    --items source=s3://my-omics-bucket/sample.vcf.gz
    --annotation-fields '{"VEP": "CSQ"}'
```

Annotation stores created before May 15, 2023 return an error message if the **annotation-fields** is included. They don't return output for any API operations involved with annotation store import jobs.

You can then use the **get-annotation-import-job** API operation and the job ID parameter to learn more details about the annotation import job.

```
aws omics get-annotation-import-job --job-id 9e4198fb-fa85-446c-9301-9b823a1a8ba8
```

You will receive the following response, including the annotation fields.

```
{
    "creationTime": "2023-04-11T19:09:25.049767+00:00",
    "destinationName": "parsingannotationstore",
    "versionName": "parsingannotationstore",
    "id": "9e4198fb-fa85-446c-9301-9b823a1a8ba8",
    "items": [
        {
            "jobStatus": "COMPLETED",
            "source": "s3://my-omics-bucket/sample.vep.vcf"
        }
    ],
    "roleArn": "arn:aws:iam::55555555555:role/roleName",
    "runLeftNormalization": false,
    "status": "COMPLETED",
    "updateTime": "2023-04-11T19:13:09.110130+00:00",
    "annotationFields" : {"VEP": "CSQ"}
}
```

To view all annotation store import jobs, use list-annotation-import-jobs.

```
aws omics list-annotation-import-jobs --ids 9e4198fb-fa85-446c-9301-9b823a1a8ba8
```

The response includes the details and statuses of your annotation store import jobs.

```
{
    "annotationImportJobs": [
    {
        "creationTime": "2023-04-11T19:09:25.049767+00:00",
        "destinationName": "parsingannotationstore",
        "versionName": "parsingannotationstore",
        "id": "9e4198fb-fa85-446c-9301-9b823a1a8ba8",
        "roleArn": "arn:aws:iam::555555555555sirole/roleName",
        "runLeftNormalization": false,
        "status": "COMPLETED",
        "updateTime": "2023-04-11T19:13:09.110130+00:00",
        "annotationFields": {"VEP": "CSQ"}
}
]
```

## Creating new versions of annotation stores

You can create new versions of annotation stores to collect different versions of your annotation databases. This helps you organize your annotation data, which is updated regularly.

To create a new version of an existing annotation store, use the **create-annotation-store-version** API as shown in the following example.

```
aws omics create-annotation-store-version \
    --name my_annotation_store \
    --version-name my_version
```

You will get the following response with the annotation store version ID, confirming that a new version of your annotation has been created.

```
{
```

To update the description of an annotation store version, you can use **update-annotation-store-version** to add updates to an annotation store version.

```
aws omics update-annotation-store-version \
    --name my_annotation_store \
    --version-name my_version \
    --description "New Description"
```

You will receive the following response, confirming that the annotation store version has been updated.

```
{
    "storeId": "4934045d1c6d",
    "id": "2a3f4a44aa7b",
    "description":"New Description",
    "status": "ACTIVE",
    "name": "my_annotation_store",
    "versionName": "my_version",
    "creation Time": "2023-07-21T17:20:59.380043+00:00",
    "updateTime": "2023-07-21T17:26:17.892034+00:00"
}
```

To view the details of an annotation store version, use **get-annotation-store-version**.

```
aws omics get-annotation-store-version --name my_annotation_store --version-name
my_version
```

You will receive a response with the version name, status, and other details.

```
{
```

```
"storeId": "4934045d1c6d",
    "id": "2a3f4a44aa7b",
    "status": "ACTIVE",
    "versionArn": "arn:aws:omics:us-west-2:55555555555ss:annotationStore/
my_annotation_store/version/my_version",
    "name": "my_annotation_store",
    "versionName": "my_version",
    "creationTime": "2023-07-21T17:15:49.251040+00:00",
    "updateTime": "2023-07-21T17:15:56.434223+00:00",
    "statusMessage": "",
    "versionSizeBytes": 0
}
```

To view all versions of an annotation store, you can use **list-annotation-store-versions**, as shown in the following example.

```
aws omics list-annotation-store-versions --name my_annotation_store
```

You will receive a response with the following information

```
{
  "annotationStoreVersions": [
     "storeId": "4934045d1c6d",
     "id": "2a3f4a44aa7b",
     "status": "CREATING",
     "versionArn": "arn:aws:omics:us-west-2:55555555555:annotationStore/
my_annotation_store/version/my_version_2",
     "name": "my_annotation_store",
     "versionName": "my_version_2",
     "creation Time": "2023-07-21T17:20:59.380043+00:00",
     "versionSizeBytes": 0
    },
     "storeId": "4934045d1c6d",
     "id": "4934045d1c6d",
     "status": "ACTIVE",
     "versionArn": "arn:aws:omics:us-west-2:55555555555:annotationStore/
my_annotation_store/version/my_version_1",
     "name": "my_annotation_store",
     "versionName": "my_version_1",
     "creationTime": "2023-07-21T17:15:49.251040+00:00",
```

```
"updateTime": "2023-07-21T17:15:56.434223+00:00",
    "statusMessage": "",
    "versionSizeBytes": 0
}
```

If you no longer need an annotation store version, you can use **delete-annotation-store-versions** to delete an annotation store version, as shown in the following example.

```
aws omics delete-annotation-store-versions --name my_annotation_store --versions
my_version
```

If the store version is deleted without errors, you will receive the following response.

```
{
    "errors": []
}
```

If there are errors, you will receive a response with the details of the errors, as shown.

```
{
  "errors": [
     {
         "versionName": "my_version",
         "message": "Version with versionName: my_version was not found."
     }
  ]
}
```

If you try to delete an annotation store version that has an active import job, you will receive a response with an error, as shown.

```
{
  "errors": [
     {
        "versionName": "my_version",
        "message": "version has an inflight import running"
     }
]
}
```

In this case, you can force deletion of the annotation store version, as shown in the following example.

```
aws omics delete-annotation-store-versions --name my_annotation_store --versions
my_version --force
```

### **Creating TSV formatted annotation stores**

The following example creates an annotation store using a tab limited file that contains a header, rows, and comments. The coordinates are CHR\_START\_END\_ONE\_BASED, and it contains the HG19 gene map from the OMIM's Synopsis of the Human Gene Map.

```
aws omics create-annotation-store --name mimgenemap \
  --store-format TSV \
  --reference=referenceArn=arn:aws:omics:us-
west-2:555555555555:referenceStore/6505293348/reference/2310864158 \
  --store-options=tsvStoreOptions='{
    annotationType=CHR_START_END_ONE_BASE,
    formatToHeader={CHR=chromosome, START=genomic_position_start,
 END=genomic_position_end},
    schema=[
      {chromosome=STRING},
      {genomic_position_start=LONG},
      {genomic_position_end=LONG},
      {cyto_location=STRING},
      {computed_cyto_location=STRING},
      {mim_number=STRING},
      {gene_symbols=STRING},
      {gene_name=STRING},
      {approved_gene_name=STRING},
      {entrez_gene_id=STRING},
      {ensembl_gene_id=STRING},
      {comments=STRING},
      {phenotypes=STRING},
      {mouse_gene_symbol=STRING}]}'
```

You can import files with or without a header. To indicate this in a CLI request, use header=false, as shown in the following import job example.

```
--items=source=s3://DOC-EXAMPLE-BUCKET/annotation-examples/hg38_genemap2.txt \
--destination-name output-bucket \
--format-options=tsvOptions='{readOptions={sep="\t",header=false,comment="#"}}'
```

The following example creates an annotation store for a bed file. A bed file is a simple tab delimited file. In this example, the columns are chromosome, start, end, and region name. The coordinates are zero-based, and the data does not have a header.

```
aws omics create-annotation-store \
    --name cexbed --store-format TSV \
    --reference=referenceArn=arn:aws:omics:us-
west-2:555555555555sss:referenceStore/6505293348/reference/2310864158 \
    --store-options=tsvStoreOptions='{
    annotationType=CHR_START_END_ZERO_BASE,
    formatToHeader={CHR=chromosome, START=start, END=end},
    schema=[{chromosome=STRING}, {start=LONG}, {end=LONG}, {name=STRING}]}'
```

You can then import the bed file into the annotation store by using the following the CLI command.

The following example creates an annotation store for a tab delimited file that contains the first few columns of a VCF file, followed by columns with annotation information. It contains genome positions with information on the chromosome, start, reference and alternate alleles, and it contains a header.

```
aws omics create-annotation-store --name gnomadchrx --store-format TSV \
    --reference=referenceArn=arn:aws:omics:us-
west-2:555555555555555:referenceStore/6505293348/reference/2310864158 \
    --store-options=tsvStoreOptions='{
        annotationType=CHR_POS_REF_ALT,
        formatToHeader={CHR=chromosome, POS=start, REF=ref, ALT=alt},
        schema=[
        {chromosome=STRING},
        {start=LONG},
```

```
{ref=STRING},
{alt=STRING},
{filters=STRING},
{ac_hom=STRING},
{ac_het=STRING},
{af_het=STRING},
{af_het=STRING},
{af_het=STRING},
{am=STRING},
```

You would then import the file into the annotation store using the following the CLI command.

The following example shows how a customer can create an annotation store for a mim2gene file. A mim2gene file provides the links between the genes in OMIM and another gene identifier. It is tab delimited and contains comments.

You can then import data into your store as follows.

```
aws omics start-annotation-import-job \
```

#### **Creating VCF formatted annotation stores**

For VCF files, there are two additional inputs, ignoreQualField and ignoreFilterField, that ignore or include those parameters as shown.

You can also cancel an annotation store import, as shown. If the cancellation succeeds, you don't receive a response to this AWS CLI call. However, if the import job ID isn't found or the import job is completed, you receive an error message.

```
aws omics cancel-annotation-import-job --job-id edd7b8ce-xmpl-47e2-bc99-258cac95a508
```



Your metadata import job history for **get-annotation-import-job**, **get-variant-import-job**, **list-annotation-import-jobs**, and **list-variant-import-jobs** is auto-deleted after two years. The variant and annotation data that's imported is not auto-deleted and remains in your data stores.

# **Annotation store import job examples**

For TSV and VCF formats, there are additional parameters that inform the API of how to parse your input.



#### ▲ Important

CSV annotation data that's exported with query engines directly returns information from the dataset import. If the imported data contains formulas or commands, the file might be subject to CSV injection. Therefore, files exported with query engines can prompt security warnings. To avoid malicious activity, turn off links and macros when reading export files.

The TSV parser also performs basic bioinformatics operations, like left normalization and standardization of genomics coordinates, that are listed in the following table.

Format type	Description
Generic	Generic text file. No genomic information.
CHR_POS	Start position - 1, Add end position, which is the same as POS.
CHR_POS_REF_ALT	Contains contig, 1-base position, ref and alt allele information.
CHR_START_END_REF_ALT_ONE_BASE	Contains contig, start, end, ref and alt allele information. Coordinates are 1-based.
CHR_START_END_ZERO_BASE	Contains contig, start, and end positions. Coordinates are 0-based.
CHR_START_END_ONE_BASE	Contains contig, start, and end positions. Coordinates are 1-based.
CHR_START_END_REF_ALT_ZERO_BASE	Contains contig, start, end, ref and alt allele information. Coordinates are 0-based.

A TSV import annotation store request looks like the following example.

```
aws omics start-annotation-import-job \
--destination-name tsv_anno_example \
--role-arn arn:aws:iam::555555555555:role/demoRole \
```

# Deleting a variant or annotation store

Both variant and annotation stores can be deleted. Deleting a variant or annotation store also deletes all the imported data in that store and any associated tags.

The following example shows how to delete a variant store by using the AWS CLI. If the action is successful, the variant store will have a status of DELETING.

```
aws omics delete-variant-store --id <variant-store-id>
```

Similarly, an annotation store can be deleted as shown. If the action is successful, the annotation store will have a status of DELETING. Annotation stores can't be deleted if more than one version exists.

```
aws omics delete-annotation-store --id <annotation-store-id>
```

# **Setting up the Lake Formation console**

In the AWS Lake Formation console, view the permissions by choosing **Data lake permissions** in the primary navigation bar. On the **Data permissions** page, you can view a table that shows the **Resource types**, **Databases**, and **ARN** that's related to a shared resource under **RAM Resource Share**. If you need to accept an AWS Resource Access Manager (AWS RAM) resource share, AWS Lake Formation notifies you in the console.

AWS HealthOmics can implicitly accept the AWS RAM resource shares during store creation. To accept the AWS RAM resource share, the IAM user or role that calls the CreateVariantStore or CreateAnnotationStore API operations must allow the following actions:

• ram: GetResourceShareInvitations - This action allows AWS HealthOmics to find the invitations.

• ram: AcceptResourceShareInvitation - This action allows AWS HealthOmics to accept the invitation by using an FAS token.

Without these permissions, you see an authorization error during store creation.

Here is a sample policy that includes these actions.

```
{
    "Statement": [
    {
        "Effect": "Allow",
        "Action": [
            "omics:*",
            "ram:AcceptResourceShareInvitation",
            "ram:GetResourceShareInvitations"
        ],
        "Resource": "*"
    }
]
```

To make a shared resource that HealthOmics Analytics users can query, the default access controls must be disabled. To learn more about disabling default access controls, see <a href="Changing the default security settings for your data lake">Changing the default security settings for your data lake</a> in the Lake Formation documentation. You can create resource links individually or as a group, so that you can access data in Athena or other AWS services.

# Creating resource links in the AWS Lake Formation console and sharing them with HealthOmics Analytics users

- 1. Open the AWS Lake Formation console: https://console.aws.amazon.com/lakeformation/
- 2. In the primary navigation bar, choose **Databases**.
- 3. In the **Databases** table, choose the **Name** of HealthOmics Analytics data store.
- 4. On the HealthOmics Analytics data store details page, choose **Actions** (▼).
- 5. Choose Create resource link.
- 6. Next, you must provide a **Resource link name**.
- 7. Choose **Create**.
- 8. The new resource link is now listed under **Databases**.

Now, the Lake Formation database administrator needs to grant access to this shared resource using **Grant on target**.

- 1. Open the AWS Lake Formation console: https://console.aws.amazon.com/lakeformation/
- 2. In the primary navigation bar, choose **Databases**.
- 3. On the **Databases** page, Choose the radio button next the **Name** of the resource link you previously created.
- 4. Next, choose **Actions** (▼).
- 5. Then, choose **Grant on target**.
- 6. On the **Grant data permissions** page under **Principals**, choose **IAM users or roles**.
- 7. Under **IAM users or roles** use the **down arrow** (▼) to find the user to which you want to grant access.
- Next, under LF-Tags or catalog resources card, select the Named data catalog resources option.
- 9. Under **Tables-optional** use the **down arrow** (▼) to choose **All Tables** you previously created.
- 10. In the **Table permissions** card, under **Table permissions** choose **Describe** and **Select**.
- 11. Next, choose **Save**.

To view the Lake Formation permissions that you have granted, choose **Data lake permissions** from the primary navigation pane. The table shows all databases and resource links that you have created.

# **Cross-account sharing**

Use cross-account sharing to share your data with collaborators without adding IAM resource share policies. The process starts with a store owner creating an invite that is sent to a subscriber, who then accepts the invite for access. The data shows up as a table in both AWS Glue and Lake Formation.

To create a share, use the **create-share** API operation, as shown in the following example. The principal subscriber is the account that you're sharing your analytic store data with.

```
aws omics create-share --resource-arn "arn:aws:omics:us-
west-2:55555555555:variantStore/omics_dev_var_store"
```

```
--principal-subscriber "123456789012"
--name "my_Share-123"
```

You receive the following response with the share ID and status, confirming that the share has been created.

```
{
    "shareId": "495c21bedc889d07d0ab69d710a6841e-dd75ab7a1a9c384fa848b5bd8e5a7e0a",
    "name": "my_Share-123",
    "status": "PENDING"
}
```

After the share is created, a subscriber can accept it with the accept-share API operation, as shown.

```
aws omics accept-share --share-id "495c21bedc889d07d0ab69d710a6841e-dd75ab7a1a9c384fa848b5bd8e5a7e0a"
```

After the share is accepted, you receive the following response showing that the share is activating.

```
{
    "status": "ACTIVATING"
}
```

To learn more about a share you created, use the **get-share** API, as shown.

```
aws omics get-share --share-id "495c21bedc889d07d0ab69d710a6841e-dd75ab7a1a9c384fa848b5bd8e5a7e0a"
```

This returns a response with share metadata that includes information such as the share Id and status, as shown.

```
"status": "PENDING"
}
```

To view all shares you own, use the **list-shares** API as shown in the following example.

```
aws omics list-shares --resource-owner SELF
```

The response will list the shares you have created, as well as their metadata.

```
{
    "shares": [
       {
         "shareId": "595c1cbd-a008-4eca-a887-954d30c91c6e",
         "name": "mvShare",
         "principalSubscriber": "123456789012",
         "ownerId": "555555555555",
         "status": "PENDING"
       },
         "shareId": "39b65d0d-4368-4a19-9814-b0e31d73c10a",
         "name": "myShare3456",
         "resourceArn": "arn:aws:omics:us-west-2:55555555555:variantStore/store_2",
         "principalSubscriber": "123456789012",
         "ownerId": "55555555555",
         "status": "ACTIVE"
       },
         "shareId": "203152f5-eef9-459d-a4e0-a691668d44ef",
         "name": "myShare4",
         "resourceArn": "arn:aws:omics:us-west-2:555555555555:variantStore/store_3",
         "principalSubscriber": "123456789012",
         "ownerId": "555555555555",
         "status": "ACTIVE"
       }
}
```

To view all shares you have accepted from other accounts, use the **list-shares** API as shown in the following example.

```
aws omics list-shares --resource-owner OTHER
```

The response will list the shares available to you, as well as their metadata.

```
{
     "shares": [
        {
          "shareId": "595c1cbd-a008-4eca-a887-954d30c91c6e",
          "name": "myShare",
          "resourceArn": "arn:aws:omics:us-west-2:123456789012:variantStore/store_1",
          "principalSubscriber": "55555555555",
          "ownerId": "444455556666",
          "status": "PENDING"
        },
        {
          "shareId": "39b65d0d-4368-4a19-9814-b0e31d73c10a",
          "name": "myShare3456",
          "resourceArn": "arn:aws:omics:us-west-2:123456789012:variantStore/store_2",
          "principalSubscriber": "55555555555",
          "ownerId": "111122223333",
          "status": "ACTIVE"
        },
          "shareId": "203152f5-eef9-459d-a4e0-a691668d44ef",
          "name": "myShare4",
          "resourceArn": "arn:aws:omics:us-west-2:123456789012:variantStore/store_3",
          "principalSubscriber": "55555555555",
          "ownerId": "444455556666",
          "status": "ACTIVE"
        }
      ]
 }
```

You can delete a share once it is no longer needed. To do so, use the delete-share API as shown.

```
aws omics delete-share --share-id "495c21bedc889d07d0ab69d710a6841e-dd75ab7a1a9c384fa848b5bd8e5a7e0a"
```

You will receive the following response to confirm that the share has been deleted.

```
{
```

"status": "DELETING"

}

## **HealthOmics Workflows**

With HealthOmics Workflows, you can process and analyze your genomics data using either Ready2Run workflows or private workflows described in CWL, WDL, or Nextflow that you create yourself.

Ready2Run workflows are created by life science publishers. They are ready to run without further definition or configuration. *Private workflows* are created and defined by you. A *run* is a single invocation of a workflow, and a *task* is a single process within the run.

HealthOmics workflows use your requested vCPU and memory to run each task. Before you create a workflow, you must containerize your workflow tools and create corresponding private image repositories in Amazon Elastic Container Registry (Amazon ECR). You can run a workflow on genomics data that's stored in an Amazon S3 bucket or in an HealthOmics sequence store.

With the workflow API operations, you can perform the following actions:

- Creating, retrieving, and managing workflows
- Starting and managing runs, including cancellation and deletion
- Tracking the status of ongoing runs
- Creating and managing run groups
- Tagging AWS resources such as workflows, runs, and run groups

Workflows written in WDL versions 1.0 and 1.1, Nextflow v22.04.0, or CWL versions 1.0, 1.1, 1.2 are supported. To learn more about workflow languages, see the specifications for <u>WDL</u>, <u>Nextflow</u>, or <u>CWL</u>.

For full examples of how to use HealthOmics workflows, see <u>AWS HealthOmics Github tutorials</u> or the AWS workshop end to end tutorial for AWS HealthOmics.

# Setting up private workflows

If you're not using a Ready2Run workflow, the first step to using HealthOmics Workflows is to create a private workflow. Whether you create your own workflow or use a Ready2Run workflow, you need to specify the following:

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- An input S3 bucket or HealthOmics storage URI.
- An output S3 bucket.
- An Amazon ECR container image stored in an Amazon ECR repository. It is recommended that
  you define your Amazon ECR container image URIs as parameters in your workflow so that access
  can be verified before the run begins. It also makes it easier to run a workflow in a new Region by
  simply changing the Region parameter.
- An IAM policy that grants the workflow access to the preceding resources. For instance, if your workflow definition file specifies an output S3 bucket, you must also grant write access to that bucket through an IAM policy or role.

The following is a comprehensive example of an IAM role that grants permission to access those resources. This policy also includes access to some Amazon CloudWatch logs that can help with troubleshooting or tracking the use of AWS actions and resources. The CloudWatch permissions aren't required to run a workflow.

```
{
    "Version": "2012-10-17",
    "Statement": [
        {
            "Effect": "Allow",
            "Action": [
                 "s3:GetObject"
            ],
            "Resource": [
                 "arn:aws:s3:::DOC-EXAMPLE-BUCKET1/*"
            ]
        },
        {
            "Effect": "Allow",
            "Action": [
                 "s3:ListBucket"
            ],
            "Resource": [
                 "arn:aws:s3:::DOC-EXAMPLE-BUCKET1"
            ]
        },
            "Effect": "Allow",
             "Action": [
                 "s3:PutObject"
```

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```
],
            "Resource": [
                "arn:aws:s3:::DOC-EXAMPLE-BUCKET2/*"
            ]
        },
        {
            "Effect": "Allow",
            "Action": [
                "logs:DescribeLogStreams",
                "logs:CreateLogStream",
                "logs:PutLogEvents"
            ],
            "Resource": [
                "arn:aws:logs:us-west-2:123456789012:log-group:/aws/omics/
WorkflowLog:log-stream:*"
            ]
        },
        {
            "Effect": "Allow",
            "Action": [
                "logs:CreateLogGroup"
            ],
            "Resource": [
                "arn:aws:logs:us-west-2:{{accountId}}:log-group:/aws/omics/
WorkflowLog: *"
        },
          "Effect": "Allow",
          "Action": [
              "ecr:BatchGetImage",
              "ecr:GetDownloadUrlForLayer",
              "ecr:BatchCheckLayerAvailability"
          ],
          "Resource": [
              "arn:aws:ecr:us-west-2:{{accountId}}:repository/*"
          ]
      }
    ]
}
```

The role must authorize the service to assume it, which can be done by adding the following trust policy.

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#### Workflow definition files

Workflow definitions must be written in WDL, Nextflow, or CWL. All parameters must be declared in the workflow definition file. Parameters include input and output locations, Amazon ECR container repositories, and runtime parameters such as allocated memory or CPU.

Note that the storage requirements to perform runs may be more than expected due to internal file system usage, so allow for more allocated memory than anticipated in your workflow definition file.

Output files must also be declared in the workflow definition file. If intermediate files are produced by the workflow run and you want to copy them to the output location, you must declare them as workflow outputs. For HealthOmics Storage URIs and input Amazon S3 URIs, access must be granted through an IAM policy. They must also be in the same Region as the workflow run.

HealthOmics storage workflow inputs must be in ACTIVE status. Input with an ARCHIVED status will not be imported and will cause a workflow to fail.

The following is an example WDL workflow that reads the contents of an INPUT file and writes them into a RESULT file.

```
version 1.0

workflow TestFlow {
   input {
     File input_txt_file
   }
```

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```
# Copies input file data to output.
    call TxtFileCopyTask{
        input:
            input_txt_file = input_txt_file,
    }
    output {
        File output_txt_file = TxtFileCopyTask.output_txt_file
    }
}
# Task definitions.
task TxtFileCopyTask {
    input {
        File input_txt_file
    }
    command {
        cat ~{input_txt_file} > outfile.txt
    }
    output {
        File output_txt_file = "outfile.txt"
    }
    runtime {
        cpu: 2
        memory: "4 GiB"
        docker: "ACCOUNT-ID.dkr.ecr.us-west-2.amazonaws.com/ubuntu:latest"
    }
}
```

You must zip the workflow definition file and any dependencies, such as subworkflows, before you can use the file to create a workflow with the **create-workflow** API operation.

```
{
    "input_txt_file": {
        "description": "Input file to be copied",
        "required": true
    }
}
```

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## **Parameter templates**

When creating a workflow you must create a parameter template JSON file if the workflow has required inputs. Each input is a named object where the name must match the name of the workflow input exactly. Each object must have a description string which will be displayed in the service console's start run page. Each object may declare a boolean optional value indicating if the value is optional for all runs. If a parameter is not marked as optional, the default value is true, as shown in the following example parameter template in JSON.

```
{
  "myRequiredParameter1": {
    "description": "this parameter is required",
},
  "myRequiredParameter2": {
    "description": "this parameter is also required",
    "optional": false
},
  "myOptionalParameter": {
    "description": "this parameter is optional",
    "optional": true
}
```

A parameter template is not required for workflows written in CWL. HealthOmics auto-detects all the workflow inputs for CWL workflows.

After you define your workflow and the parameters, you can create a workflow using the CLI as shown. If you are including multiple workflow definition files, use the --main parameter to specify which file is the main definition file for your workflow. You can also specify the accelerator used.

```
aws omics create-workflow
--name Test --main multi_workflow/workflow2.wdl
--definition-zip fileb://definition.zip
--parameter-template file://params_sample_description.json
--accelerators GPU
```

You should receive the following response to confirm that the workflow has been created.

```
{
    "arn": "arn:aws:omics:us-west-2:....",
    "id": "1234567",
```

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```
"status": "CREATING",
"tags": {
    "resourceArn": "arn:aws:omics:us-west-2:...."
}
```

Larger zip files containing the workflow definition can be loaded from an Amazon S3 bucket using the --definition-uri parameter, as shown.

#### Workflow run parameters and inputs

When starting a workflow run, a JSON file containing the details of any inputs must be included if the parameter template declares any required parameters. The JSON file will contain the exact name of the inputs and an appropriate JSON value.

Reference the input JSON file in the AWS CLI by adding --inputs file://
<input\_file.json> to your start-run request.

An HealthOmics reference store object can be referred to with a URI like the following. Use your own *account ID*, *reference store ID*, and *reference ID* where indicated.

```
omics://account ID.storage.us-west-2.amazonaws.com/reference store id/reference/id
```

Some workflows will require both the SOURCE and INDEX files for the reference genome. The previous URI is the default short form and will default to the SOURCE file. In order to specify either file, you can use the long URI form, as follows.

```
omics://account ID.storage.us-west-2.amazonaws.com/reference store id/reference/id/
source
omics://account ID.storage.us-west-2.amazonaws.com/reference store id/reference/id/
index
```

Using a sequence read set would have a similar pattern, as shown.

```
aws omics create-workflow \
    --name workflow name \
    --main sample workflow.wdl \
    --definition-uri omics://account ID.storage.us-
west-2.amazonaws.com/sequence_store_id/readSet/id \
    --parameter-template file://parameters_sample_description.json
```

Some read sets, such as those based on FASTQ, can contain paired reads. In the following examples, they're referred to as SOURCE1 and SOURCE2. Formats such as BAM and CRAM will only have a SOURCE1 file. Some read sets will contain INDEX files such as bai or crai files. The preceding URI is the default short form and will default to the SOURCE1 file. To specify the exact file or index, you can use the long URI form, as follows.

```
omics://123456789012.storage.us-west-2.amazonaws.com/<sequence_store_id>/readSet/<id>/
source1
omics://123456789012.storage.us-west-2.amazonaws.com/<sequence_store_id>/readSet/<id>/
source2
omics://123456789012.storage.us-west-2.amazonaws.com/<sequence_store_id>/readSet/<id>/
index
```

The following is an example of an input JSON file that uses two Omics Storage URIs.

```
{
    "input_fasta": "omics://123456789012.storage.us-west-2.amazonaws.com/
<reference_store_id>/reference/<id>",
    "input_cram": "omics://123456789012.storage.us-west-2.amazonaws.com/
<sequence_store_id>/readSet/<id>"
}
```

Reference the input JSON file in the AWS CLI by adding --inputs file:// <input\_file.json> to your **start-run** request.

# Verifying the status of your workflow

After you create your workflow, you can verify the status and view other details of the workflow using **get-workflow**, as shown.

```
aws omics get-workflow --id 1234567
```

The response gives you your workflow details, including the status, as shown.

```
{
    "arn": "arn:aws:omics:us-west-2:....",
    "id": "1234567",
    "status": "ACTIVE",
    "type": "PRIVATE",
    "name": "workflow_name"
    "creationTime": "2022-07-06T00:27:05.542459"
```

}

Before a run can be started, the status must be listed as ACTIVE.

#### **Workflow tasks**

Workflow tasks are the individual processes within a run. Each task has a unique identifier. For a private workflow, HealthOmics workflows will use your defined compute specifications to run your task. Input files to the workflow and workflow tasks are staged to a scratch volume that is dedicated to the workflow run. They are read-only, which prevents tasks modifying potential inputs to other tasks in a workflow. The directories included as input are also read-only. All inputs are made available to the tasks' working directory as symbolic links. They're only accessible if they're declared in the workflow definition file. Many genomics applications assume that index files will be in the same location as a sequence file (such as a companion bai file for a bam file). To make sure indexes are present, you must specify them as tasks inputs.

Because workflow tasks can't connect to the public internet, they can't download resources by using http, https, or ftp. Required resources should be included as workflow inputs from Amazon S3 or an HealthOmics sequence store. They also should be present in the container images that are used to run workflow tasks. Workflow tasks may interact with Amazon S3 as long as the IAM role used to run the workflow has been granted access.

#### Memory and computation considerations for tasks

Private workflow tasks are run on HealthOmics instances by using the smallest instance that can accommodate the requested CPU and memory. We recommend choosing the most sensible combination of CPU and memory for your needs. For example, if you need 64 GiB of RAM, then the most cost-effective type is omics.r.2xlarge. This type allocates eight vCPUs. If your task allocation only calls for one vCPU, the task container only gets one, even though eight are available on the host machine. Unless this will adversely affect the workflow, you might want to set a larger number of vCPUs. All tasks reserve a small amount of memory for management and logging agents, so the full memory allocation might not always be available to the application in the task.

Container resource allocations are hard limits. Tasks that run out of memory or attempt to use additional vCPUs can be immediately shut down by the host, potentially without warning.

#### Running Java applications in a private workflow task

When running Java applications in a workflow task, the image used to run the task must contain Java 1.8–212 or later. Versions before this might attempt to allocate heap memory that's not

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available to the container. By default, for versions after 1.8-212, the heap allocation of the JVM will be 25% of the memory available to the container. If you use a -Xmx flag to request a larger amount, be aware that not all memory consumed by Java is heap memory. Allocating 100% of the available memory to the JVM heap causes the task to fail.

#### **Including task inputs in Amazon ECR images**

All executables, libraries, and scripts needed to run a workflow task should be provided by the Amazon ECR image that's used to run the task.

It's best practice to avoid using scripts, binaries, and libraries that are external to a tasks container image. This is especially important when using nf-core workflows that use a bin directory as part of the workflow package. While this directory will be available to the workflow task, it is mounted as a read-only directory. Required resources in this directory should be copied into the task image and made available at runtime or when building the container image used for the task.

#### **Debugging workflow tasks**

The following are best practices and considerations for debugging your tasks and workflows.

- Task logs rely on STDOUT and STDERR being produced by the task. If the application used in the task doesn't produce either of these, then there won't be a task log. To assist with debugging, use applications in verbose mode.
- To view the commands being run in a task along with their interpolated values, use the set -x Bash command. This can help determine if the task is using the correct inputs and identify where errors might have kept the task from running as intended.
- Use the echo command to output the values of variables to STDOUT or STDERR. This helps you confirm that they're being set as expected.
- Use commands like 1s -1 <name\_of\_input\_file> to confirm that inputs are present and are of the expected size. If they aren't, this might reveal a problem with a prior task producing empty outputs due to a bug.
- Use the command df -Ph . | awk 'NR==2 {print \$4}' in a tasks script to determine the space currently available to the task and help identify situations where you might need to run the workflow with additional storage allocation.

Including any of the preceding commands in a task script assumes that the task container also includes these commands and that they are on the path of the container environment.

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# **Using custom IAM permissions for runs**

You can include any workflow, run, or run group referenced by the StartRun request in an authorization request. To do so, list the desired combination of workflows, runs, or run groups in the IAM policy. For example, you can limit the use of a workflow to a specific run or run group. You can also specify that a workflow only be used with a run group.

The following is an example IAM policy that allows a single workflow with a single run group.

```
{
    "Version": "2012-10-17",
    "Statement": [
        {
            "Effect": "Allow",
            "Action": [
                 "omics:StartRun"
            ],
            "Resource": [
                "arn:aws:omics:us-west-2:123456789012:workflow/1234567",
                "arn:aws:omics:us-west-2:123456789012:runGroup/2345678"
            ]
        },
            # Optionally, allow user to rerun a failed run.
            "Effect": "Allow",
            "Action": [
                "omics:StartRun"
            ],
            "Resource": [
                "arn:aws:omics:us-west-2:123456789012:run/*",
                "arn:aws:omics:us-west-2:123456789012:runGroup/2345678"
            ]
        },
            "Effect": "Allow",
            "Action": [
                "omics:GetRun",
                "omics:ListRunTasks",
                "omics:GetRunTask",
                "omics:CancelRun",
                "omics:DeleteRun"
            ],
            "Resource": [
```

```
"arn:aws:omics:us-west-2:123456789012:run/*"

]
},

]
```

# Writing workflow definition files

You can write your own workflow definition file for private workflows. Workflows written in WDL, Nextflow, and CWL are supported.

# Writing workflows in WDL

The following tables show how inputs in WDL map to the matching primitive type or complex JSON type. Type coercion is limited and whenever possible, types should be explicit.

#### **Primitive types**

WDL type	JSON type	Example WDL	Example JSON key and value	Notes
Boolean	boolean	Boolean b	"b": true	The value must be lower case and unquoted.
Int	integer	Int i	"i": 7	Must be unquoted.
Float	number	Float f	"f": 42.2	Must be unquoted.
String	string	String s	"s": "characte rs"	JSON strings that are a URI must be mapped to a WDL file to be imported.
File	string	File f	"f": "s3:// BUCKET-	Amazon S3 and HealthOmics

WDL type	JSON type	Example WDL	Example JSON key and value	Notes
			NAME/path/ to/file"	storage URIs are imported as long as the IAM role provided for the workflow has read access to these objects. No other URI schemes are supported (such as file://, https://, and ftp://). The URI must specify an object. It cannot be a directory meaning it can not end with a /.

WDL type	JSON type	Example WDL	Example JSON key and value	Notes
Directory	string	Directory d	"d": "s3:// bucket/ path/"	The Directory type is not included in WDL 1.0 or 1.1, so you will need to add version development to the header of the WDL file. The URI must be a Amazon S3 URI and with a prefix that ends with a '/'. All contents of the directory will be recursively copied to the workflow as a single download. The Directory should only contain files related to the workflow. It is not recommend ed to include a Directory with many objects or subdirectories as it may add

WDL type	JSON type	Example WDL	Example JSON key and value	Notes
				delays while downloading.

Complex types in WDL are data structures comprised of primitive types. Data structures such as lists will be converted to arrays.

#### **Complex types**

WDL type	JSON type	Example WDL	Example JSON key and value	Notes
Array	array	Array[Int] nums	"nums": [1, 2, 3]	The members of the array must follow the format of the WDL array type.
Pair	object	Pair[Stri ng, Int] str_to_i	<pre>"str_to_i ": {"left": "a", "right": 1}</pre>	Each value of the pair must use the JSON format of its matching WDL type.
Мар	object	<pre>Map[Int, String] int_to_st ring</pre>	<pre>"int_to_s tring": { 2: "hello", 1: "goodbye" }</pre>	Each entry in the map must use the JSON format of its matching WDL type.
Struct	object	<pre>struct SampleBam AndIndex { String sample_na</pre>	<pre>"b_and_i": { sample_na me: "NA12878" , bam:</pre>	The names of the struct members must exactly match the names of

WDL type	JSON type	Example WDL	Example JSON key and value	Notes
		me File bam File bam_index } SampleBam AndIndex b_and_i	"s3://BUC KET-NAME/ NA12878.b am", bam_index : "s3://BUC KET-NAME/ NA12878.b am.bai" }	the JSON object keys. Each value must use the JSON format of the matching WDL type.
0bject	N/A	N/A	N/A	The WDL Object type is outdated and should be replaced by Struct in all cases.

The HealthOmics workflow engine does not support qualified or name-spaced input parameters. Handling of qualified parameters and their mapping to WDL parameters is not specified by the WDL language and can be ambiguous. For these reasons, all input parameters should be declared at the top level (main) workflow and passed down to subworkflow calls by using standard WDL mechanisms.

In addition to standard cpu, memory, and container task runtime directives, HealthOmics also supports acceleratorCount and acceleratorType. The acceleratorType is dependent on the number of GPUs. It can be either a G5 instance, such as nvidia-tesla-a10g, or a G4 instance, such as nvidia-tesla-t4 or nvidia-tesla-t4-a10g. G4 instances are not supported in the Israel (Tel Aviv) region.

A workflow definition file written in WDL that has the accelerator count and type defined in the parameters would look like the following.

version 1.1

```
workflow hello_gpu {
    call hello {}
    call nvidia_smi {}
}
task hello {
    command {
        echo "hello"
    }
    runtime {
    }
    output {
        String out = read_string( stdout() )
}
task nvidia_smi {
    command {
        nvidia-smi
    runtime {
        # Note: you will need to provision the following container image in your ECR
 Private registry
        container: "111122223333.dkr.ecr.us-west-2.amazonaws.com/nvidia/cuda:10.0-
devel-centos7"
        acceleratorCount: 1
        acceleratorType: "nvidia-tesla-t4-a10g"
    }
    output {
        String out = read_string( stdout () )
    }
}
```

# Writing workflows in Nextflow

Nextflow DSL2 is based on the Groovy programming language, so parameters are dynamic and type coercion is possible using the same rules as Groovy. Parameters and values supplied by the input JSON are available in the parameters (params) map of the workflow.

When an Amazon S3 or HealthOmics URI is used to construct a Nextflow file or path object, it makes the matching object available to the workflow, as long as read access is granted. The use

of prefixes or directories is allowed for Amazon S3 URIs. HealthOmics does not currently support the use of glob patterns such as "s3://BUCKET-NAME/path/\*.gz" in Amazon S3 URIs or HealthOmics Storage URIs because POSIX glob pattern behavior is undefined. Glob patterns may be used within the workflow definition in the creation of path or file channels.

HealthOmics supports Nextflow task directives accelerator and type. The acceleratorType is dependent on the number of GPUs. It can be either a G5 instance, such as nvidia-tesla-a10g, or a G4 instance, such as nvidia-tesla-t4 or nvidia-tesla-t4-a10g. G4 instances are not supported in the Israel (Tel Aviv) region.

A workflow definition file written in Nextflow that has the accelerator count and type defined in the parameters would look like the following.

```
nextflow.enable.dsl = 2
process hello {
    output:
        stdout emit: out
    script:
        echo "hello"
        .....
}
process nvidia_smi {
    // Note: you will need to provision the following container image in your ECR
 Private registry
    container "111122223333.dkr.ecr.<aws-region>.amazonaws.com/nvidia/cuda:10.0-devel-
centos7"
    accelerator 1, type: 'nvidia-tesla-t4-a10g'
    output:
        stdout emit: out
    script:
        nvidia-smi
        11 11 11
```

```
workflow HELLO_GPU {
   hello()
   nvidia_smi()
}

workflow {
   HELLO_GPU()
}
```

## Writing workflows in CWL

Workflows written in Common Workflow Language, or CWL, offer similar functionality to workflows written in WDL and Nextflow. CWL versions 1.0, 1.1, and 1.2 are supported. You can use Amazon S3 or HealthOmics storage URIs as input parameters. We recommend that you declare Amazon ECR containers in the workflow as input parameters for validation of the Amazon ECR permissions. The workflow should be hosted in the same Region as your workflow.

If input is defined in a secondaryFile in a sub workflow, the same definition must also be present in the main workflow.

To run a workflow in CWL, the following changes are required.

- All Docker container URIs should be replaced with Amazon ECR URIs.
- All the workflow files are declared in the main workflow as input, and all variables are explicitly defined.
- All JavaScript code is strict mode complaint.

CWL workflows should be defined for each container used. It is not recommended to hardcode the dockerPull entry with a fixed Amazon ECR URI.

HealthOmics workflows do not support operation processes. To learn more about operations processes in CWL workflows, see the CWL documentation.

HealthOmics supports GPU acceleration in CWL using cwltool:CUDARequirement extension syntax. The accelerator type is determined by cudaComputeCapability. It can be either a G5 instance, such as nvidia-tesla-a10g, or a G4 instance, such as nvidia-tesla-t4. If either G4 or G5 can be used for a

task, nvidia-tesla-t4-a10g is recommended. G4 instances are not supported in the Israel (Tel Aviv) region.

It's recommended that you input your container URIs as parameters. This makes it possible to check the permissions for access to the container or for you to use the workflow in multiple Regions without having to recreate the workflow in each new Region.

The following is an example of a workflow written in CWL.

```
cwlVersion: v1.2
class: Workflow
inputs:
  in_file:
    type: File
    secondaryFiles: [.fai]
  out_filename: string
  docker_image: string
outputs:
  copied_file:
    type: File
    outputSource: copy_step/copied_file
steps:
  copy_step:
    in:
      in_file: in_file
      out_filename: out_filename
      docker_image: docker_image
    out: [copied_file]
    run: copy.cwl
```

The following file defines the copy.cwl task.

```
cwlVersion: v1.2
class: CommandLineTool
baseCommand: cp
```

```
inputs:
  in_file:
    type: File
    secondaryFiles: [.fai]
    inputBinding:
      position: 1
  out_filename:
    type: string
    inputBinding:
      position: 2
  docker_image:
    type: string
outputs:
  copied_file:
    type: File
    outputBinding:
        glob: $(inputs.out_filename)
requirements:
  InlineJavascriptRequirement: {}
  DockerRequirement:
    dockerPull: "$(inputs.docker_image)"
```

The following is an example of a workflow written in CWL with a GPU requirement.

```
cwlVersion: v1.2
class: CommandLineTool
baseCommand: ["/bin/bash", "docm_haplotypeCaller.sh"]
$namespaces:
    cwltool: http://commonwl.org/cwltool#
requirements:
    cwltool:CUDARequirement:
        cudaDeviceCountMin: 1
        cudaComputeCapability: "nvidia-tesla-t4"
        cudaVersionMin: "1.0"

inputs: []
outputs: []
```

```
- class: InlineJavascriptRequirement
- class: InitialWorkDirRequirement
  listing:
  - entryname: 'docm_haplotypeCaller.sh'
    entry: |
        nvidia-smi --query-gpu=gpu_name,gpu_bus_id,vbios_version --format=csv
```

## **Workflow definition file examples**

The following examples are private workflow definitions for converting from CRAM to BAM in WDL. The CRAM to BAM workflow defines two tasks and uses tools from the genomes-in-the-cloud container, which is shown in the example and is publicly available.

Note that HealthOmics workflows require Amazon ECR containers to be in the same account and Region as the account calling the service. Amazon ECR containers should be included as parameters in your workflow to validate access. Workflows written in WDL don't support output.

To allow HealthOmics to access the Amazon ECR container, add the following policy to your account in the section that covers Amazon ECR repository permissions.

```
{
  "Version": "2008-10-17",
  "Statement": [
    {
        "Effect": "Allow",
        "Principal": {
            "Service": "omics.amazonaws.com"
        },
        "Action": [
            "ecr:BatchGetImage",
            "ecr:GetDownloadUrlForLayer"
        ]
    }
}
```

You can include the Amazon ECR container as a parameter by including it in your workflow as shown. This is recommended so that the access permissions to your image are checked when you start the run. The following file defines all parameters for your workflow.

```
"input_cram": "s3://DOC-EXAMPLE-BUCKET1/inputs/NA12878.cram",
    "ref_dict": "s3://DOC-EXAMPLE-BUCKET1/inputs/Homo_sapiens_assembly38.dict",
    "ref_fasta": "s3://DOC-EXAMPLE-BUCKET1/inputs/Homo_sapiens_assembly38.fasta",
    "ref_fasta_index": "s3://DOC-EXAMPLE-BUCKET1/inputs/
Homo_sapiens_assembly38.fasta.fai",
    "sample_name": "NA12878",

    "gotc_docker":"<account_id>.dkr.ecr.<region>.amazonaws.com/genomes-in-the-cloud:2.4.7-1603303710"
}
```

Then specify which files to use in your run. The following example is for when your files are stored in an Amazon S3 bucket.

```
"input_cram": "s3://DOC-EXAMPLE-BUCKET1/inputs/NA12878.cram",
    "ref_dict": "s3://DOC-EXAMPLE-BUCKET1/inputs/Homo_sapiens_assembly38.dict",
    "ref_fasta": "s3://DOC-EXAMPLE-BUCKET1/inputs/Homo_sapiens_assembly38.fasta",
    "ref_fasta_index": "s3://DOC-EXAMPLE-BUCKET1/inputs/
Homo_sapiens_assembly38.fasta.fai",
    "sample_name": "NA12878"
}
```

If you want to specify files from a sequence store, indicate that as shown in the following example, using the URI for the sequence store.

```
{
    "input_cram": "omics://429915189008.storage.us-west-2.amazonaws.com/111122223333/
readSet/4500843795/source1",
    "ref_dict": "s3://DOC-EXAMPLE-BUCKET1/inputs/Homo_sapiens_assembly38.dict",
    "ref_fasta": "s3://DOC-EXAMPLE-BUCKET1/inputs/Homo_sapiens_assembly38.fasta",
    "ref_fasta_index": "s3://DOC-EXAMPLE-BUCKET1/inputs/
Homo_sapiens_assembly38.fasta.fai",
    "sample_name": "NA12878"
}
```

You can then define your workflow in WDL as shown in the following.

```
version 1.0
```

```
workflow CramToBamFlow {
    input {
        File ref_fasta
        File ref_fasta_index
        File ref_dict
        File input_cram
        String sample_name
        String gotc_docker = "<account>.dkr.ecr.us-west-2.amazonaws.com/genomes-in-the-
cloud:latest"
    }
    #Converts CRAM to SAM to BAM and makes BAI.
    call CramToBamTask{
         input:
            ref_fasta = ref_fasta,
            ref_fasta_index = ref_fasta_index,
            ref_dict = ref_dict,
            input_cram = input_cram,
            sample_name = sample_name,
            docker_image = gotc_docker,
     }
     #Validates Bam.
     call ValidateSamFile{
        input:
           input_bam = CramToBamTask.outputBam,
           docker_image = gotc_docker,
     }
     #Outputs Bam, Bai, and validation report to the FireCloud data model.
     output {
         File outputBam = CramToBamTask.outputBam
         File outputBai = CramToBamTask.outputBai
         File validation_report = ValidateSamFile.report
      }
}
#Task definitions.
task CramToBamTask {
    input {
       # Command parameters
       File ref_fasta
       File ref_fasta_index
       File ref_dict
       File input_cram
       String sample_name
       # Runtime parameters
       String docker_image
```

```
}
   #Calls samtools view to do the conversion.
   command {
       set -eo pipefail
       samtools view -h -T ~{ref_fasta} ~{input_cram} |
       samtools view -b -o ~{sample_name}.bam -
       samtools index -b ~{sample_name}.bam
      mv ~{sample_name}.bam.bai ~{sample_name}.bai
    }
    #Runtime attributes:
    runtime {
        docker: docker_image
    }
    #Outputs a BAM and BAI with the same sample name
     output {
         File outputBam = "~{sample_name}.bam"
         File outputBai = "~{sample_name}.bai"
    }
}
#Validates BAM output to ensure it wasn't corrupted during the file conversion.
task ValidateSamFile {
   input {
      File input_bam
      Int machine_mem_size = 4
      String docker_image
   }
   String output_name = basename(input_bam, ".bam") + ".validation_report"
   Int command_mem_size = machine_mem_size - 1
   command {
       java -Xmx~{command_mem_size}G -jar /usr/gitc/picard.jar \
       ValidateSamFile \
       INPUT=~{input_bam} \
       OUTPUT=~{output_name} \
       MODE=SUMMARY \
       IS_BISULFITE_SEQUENCED=false
    }
    runtime {
    docker: docker_image
   #A text file is generated that lists errors or warnings that apply.
```

```
output {
    File report = "~{output_name}"
}
```

Workflows written in Nextflow require a **publishDir** directive to export the task contents to your output Amazon S3 bucket. Use it every time any of the task outputs need to be kept on workflow run completion. It also must point to the folder in the following example, **/mnt/workflow/pubdir**. For security reasons, this is the only folder authorized to export files to your Amazon S3 bucket.

```
nextflow.enable.dsl=2
workflow {
    CramToBamTask(params.ref_fasta, params.ref_fasta_index, params.ref_dict,
 params.input_cram, params.sample_name)
    ValidateSamFile(CramToBamTask.out.outputBam)
}
process CramToBamTask {
    container "<account>.dkr.ecr.us-west-2.amazonaws.com/genomes-in-the-cloud"
    publishDir "/mnt/workflow/pubdir"
    input:
        path ref_fasta
        path ref_fasta_index
        path ref_dict
        path input_cram
        val sample_name
    output:
        path "${sample_name}.bam", emit: outputBam
        path "${sample_name}.bai", emit: outputBai
    script:
        set -eo pipefail
        samtools view -h -T $ref_fasta $input_cram |
        samtools view -b -o ${sample_name}.bam -
```

```
samtools index -b ${sample_name}.bam
        mv ${sample_name}.bam.bai ${sample_name}.bai
}
process ValidateSamFile {
    container "<account>.dkr.ecr.us-west-2.amazonaws.com/genomes-in-the-cloud"
    publishDir "/mnt/workflow/pubdir"
    input:
        file input_bam
    output:
        path "validation_report"
    script:
    11 11 11
        java -Xmx3G -jar /usr/gitc/picard.jar \
        ValidateSamFile \
        INPUT=${input_bam} \
        OUTPUT=validation_report \
        MODE=SUMMARY \
        IS_BISULFITE_SEQUENCED=false
    11 11 11
}
```

# Ready2Run workflows

You can also use Ready2Run to analyze your data. Ready2Run workflows are preconfigured workflows published by health science publishers. Ready2Run workflows might be open source workflows such as nf-core or GATK best practices, or they might be subscription-based. When you use a Ready2Run workflow, your workflow is preconfigured and can't be edited. This means you can't edit the storage capacity of the workflow. Ready2Run workflows can't be added to run groups.

Ready2Run workflows are well-suited for the following people:

 New bioinformaticians — You can focus on the analysis of pipeline output and generating results, without needing to set up the underlying infrastructure.

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- Experienced bioinformaticians You can use established workflows to replicate results.
- Software developers You can integrate your applications directly with the AWS HealthOmics SDK.

Use Ready2Run workflows in the console or by using the same API operations that you would use with a private workflow.

If you're running a licensed workflow offered by Sentieon, the IAM role used to start the run must allow read access to s3://sentieon-omics-license-<region>.

All Ready2Run workflows provide logs, including CloudWatch logs that you can use for troubleshooting.

### Using Ready2Run workflows in the console

Using Ready2Run workflows in the console is similar to using a private workflow. One key difference is that you can't download the details for a workflow.

#### To use a Ready2Run workflow in the console

- Select the workflow that you want to use. In the details tab, view the name, price per run, description, workflow language type, run storage capacity, status, creation date, and parameters with descriptions. The details tab also tells you whether the workflow requires a subscription.
- 2. From the specify run details page, add a run name and description. These are useful for identifying specific runs later. You can also add an IAM role to access data in an Amazon S3 bucket, assign run priority to your run, add tags, and specify maximum run storage capacity.
- 3. Upload a JSON file from an Amazon S3 bucket with the parameter names to be used in your run. You can either start your run directly or add additional details through the **Create Run** page. If you upload a JSON file, the console will parse the file and perform inline validation. You can then manually add the values for your parameters as needed.
- 4. From the add parameters page, search for or manually enter the URI of the Amazon S3 bucket or HealthOmics storage where your file is located.
- 5. (Optional) Add tags to help identify and manage your run.
- 6. Review the run details and choose **Start run** to begin your run.

### Using Ready2Run workflows with the CLI or API

Most of the API operations for creating runs and workflows behave similarly for both Ready2Run and private workflows.

To find available Ready2Run workflows, use **list-workflows** with the --type= SERVICE parameter to return a list of all service workflows.

```
aws omics list-workflows --type READY2RUN
```

After you identify the workflow to run from the **list-workflows** response, you can use **get-workflow** with the --id parameter to get more details.

```
aws omics get-workflow --type READY2RUN --id workflow id
```

To run a Ready2Run workflow, you can use **start-run** API operation with the --workflow-type = READY2RUN parameter, as shown.

```
aws-omics start-run \
    --workflow-type READY2RUN \
    --workflow-id workflow id \
    --output-uri DOC-EXAMPLE-BUCKET \
    --role-arn arn:aws:iam::1234567892012:role/service-role/OmicsWorkflow-20221004T164236 \
    --parameters file:///path/to/parameters.json
```

To monitor your run, you can use the **get-run** API operation, as shown.

```
aws-omics get-run --id run id
```

# **Running workflows**

After you create your workflow, you can perform runs either individually or as part of a run group. You can also queue runs to be processed. The following topics show how to create an optional run group to limit computational resources and reduce costs. There are also examples of how to start runs and get information on ongoing runs.

#### **Starting runs**

To run a workflow, you first need an input data file in an Amazon Simple Storage Service (Amazon S3) bucket or a HealthOmics sequence store. For this example, create a text file called hello\_world.txt and upload it to an S3 bucket. Create a file called params\_sample\_content.json with the definition of the input file to use, as shown.

```
{
    "input_txt_file": "s3://DOC-EXAMPLE-BUCKET1/hello_world.txt"
}
```

Then, use the **start-run** API operation with the IAM role and S3 bucket that you created. Although the default retention mode is RETAIN, this example sets the retention mode to REMOVE. If the quota for maximum runs has been met, the earliest runs with REMOVE retention mode are deleted first. This makes room for new runs to start—even if the maximum runs limit is met—as long as there are runs with REMOVE retention mode that can be removed.

When the parameter is set to REMOVE, the run metadata is removed after the run completes and the metadata has been sent to Amazon CloudWatch.

```
aws omics start-run/
    --workflow-id workflow id \
    --role-arn arn:aws:iam::1234567892012:role/service-role/
OmicsWorkflow-20221004T164236 \
    --name workflow name \
    --retention-mode REMOVE
```

In response, you get the following output. The uuid is unique to the run, and along with runOutputUri can be used to track where output data is written.

```
{
    "arn": "arn:aws:omics:us-west-2:....:run/1234567",
    "id": "1234567",
    "uuid":"96c57683-74bf-9d6d-ae7e-f09b097db14a",
    "runOutputUri":"s3://bucket/folder/8405154/96c57683-74bf-9d6d-ae7e-f09b097db14a"
    "status": "PENDING"
}
```

You can also use the **start-run** API with a GPU workflow ID, as shown.

```
aws omics start-run --workflow-id workflow id \
     --role-arn arn:aws:iam::1234567892012:role/service-role/
OmicsWorkflow-20221004T164236 \
     --name GPUTestRunModel \
     --output-uri s3://DOC-EXAMPLE-BUCKET1
```

You can create a workflow with the accelerators parameter defined, as shown.

```
aws omics create-workflow --name workflow name \
    --definition-uri s3://DOC-EXAMPLE-BUCKET1/GPUWorkflow.zip \
    --accelerators GPU
```

You can then use the ID in the response with the **get-run** API to check the status of a run, as shown.

```
aws omics get-run --id run id
```

The response from this API operation tells you the status of the workflow run. Possible statuses are PENDING, STARTING, RUNNING, and COMPLETED. When a run is COMPLETED, you can find an output file called outfile.txt in your output S3 bucket, in a folder named after the run ID.

The **get-run** API operation also returns other details, such as whether the workflow is Ready2Run or PRIVATE, the workflow engine, and accelerator details. The following example shows the response for **get-run** for a run of a private workflow, described in WDL with a GPU accelerator and no tags assigned to the run.

```
{
    "arn": "arn:aws:omics:us-west-2:123456789012:run/7830534",
    "id": "7830534",
    "uuid": "96c57683-74bf-9d6d-ae7e-f09b097db14a",
    "runOutputUri":"s3://bucket/folder/8405154/96c57683-74bf-9d6d-ae7e-f09b097db14a"
    "status": "COMPLETED",
    "workflowId": "4074992",
    "workflowType": "PRIVATE",
    "roleArn": "arn:aws:iam::123456789012:role/service-role/
OmicsWorkflow-20221004T164236",
    "name": "RunGroupMaxGpuTest",
    "runGroupId": "9938959",
    "digest":
 "sha256:a23a6fc54040d36784206234c02147302ab8658bed89860a86976048f6cad5ac",
    "accelerators": "GPU",
    "outputUri": "s3://DOC-EXAMPLE-BUCKET1",
```

```
"startedBy": "arn:aws:sts::123456789012:assumed-role/Admin/<role_name>",
    "creationTime": "2023-04-07T16:44:22.262471+00:00",
    "startTime": "2023-04-07T16:56:12.504000+00:00",
    "stopTime": "2023-04-07T17:22:29.908813+00:00",
    "tags": {}
}
```

You can see the status of all runs with the list-runs API operation, as shown.

```
aws omics list-runs
```

To see all the tasks completed for a specific run, use the **list-run-tasks** API.

```
aws omics list-run-tasks --id task ID
```

To get the details of any specific task, use the get-run-task API.

```
aws omics get-run-task --id <run_id> --task-id task ID
```

After the run completes, the metadata is sent to CloudWatch under the stream manifest/run/<run ID>/<run UUID>.

The following is an example of the manifest.

```
{
    "arn": "arn:aws:omics:us-east-1:123456789012:run/1695324",
    "creationTime": "2022-08-24T19:53:55.284Z",
    "resourceDigests": {
      "s3://omics-data/broad-references/hg38/v0/Homo_sapiens_assembly38.dict":
 "etag:3884c62eb0e53fa92459ed9bff133ae6",
      "s3://omics-data/broad-references/hg38/v0/Homo_sapiens_assembly38.fasta":
 "etag:e307d81c605fb91b7720a08f00276842-388",
      "s3://omics-data/broad-references/hq38/v0/Homo_sapiens_assembly38.fasta.fai":
 "etag:f76371b113734a56cde236bc0372de0a",
      "s3://omics-data/intervals/hg38-mjs-whole-chr.500M.intervals":
 "etag:27fdd1341246896721ec49a46a575334",
      "s3://omics-data/workflow-input-lists/dragen-gvcf-list.txt":
 "etag:e22f5aeed0b350a66696d8ffae453227"
    },
    "digest":
 "sha256:a5baaff84dd54085eb03f78766b0a367e93439486bc3f67de42bb38b93304964",
    "engine": "WDL",
```

```
"main": "gatk4-basic-joint-genotyping-v2.wdl",
    "name": "1044-gvcfs",
    "outputUri": "s3://omics-data/workflow-output",
    "parameters": {
      "callset_name": "cohort",
      "input_gvcf_uris": "s3://omics-data/workflow-input-lists/dragen-gvcf-list.txt",
      "interval_list": "s3://omics-data/intervals/hg38-mjs-whole-chr.500M.intervals",
      "ref_dict": "s3://omics-data/broad-references/hg38/v0/
Homo_sapiens_assembly38.dict",
      "ref_fasta": "s3://omics-data/broad-references/hg38/v0/
Homo_sapiens_assembly38.fasta",
      "ref_fasta_index": "s3://omics-data/broad-references/hg38/v0/
Homo_sapiens_assembly38.fasta.fai"
    },
    "roleArn": "arn:aws:iam::123456789012:role/OmicsServiceRole",
    "startedBy": "arn:aws:sts::123456789012:assumed-role/admin/ahenroid-Isengard",
    "startTime": "2022-08-24T20:08:22.582Z",
    "status": "COMPLETED",
    "stopTime": "2022-08-24T20:08:22.582Z",
    "storageCapacity": 9600,
    "uuid": "a3b0ca7e-9597-4ecc-94a4-6ed45481aeab",
    "workflow": "arn:aws:omics:us-east-1:123456789012:workflow/1558364",
    "workflowType": "PRIVATE"
  },
    "arn": "arn:aws:omics:us-east-1:123456789012:task/1245938",
    "cpus": 16,
    "creationTime": "2022-08-24T20:06:32.971290",
    "image": "123456789012.dkr.ecr.us-west-2.amazonaws.com/gatk",
    "imageDigest":
 "sha256:8051adab0ff725e7e9c2af5997680346f3c3799b2df3785dd51d4abdd3da747b",
    "memory": 32,
    "name": "geno-123",
    "run": "arn:aws:omics:us-east-1:123456789012:run/1695324",
    "startTime": "2022-08-24T20:08:22.278Z",
    "status": "SUCCESS",
    "stopTime": "2022-08-24T20:08:22.278Z",
    "uuid": "44c1a30a-4eee-426d-88ea-1af403858f76"
  },
  . . .
```

Run metadata isn't deleted if it's not present in the CloudWatch logs. You can also use the run ID to rerun workflow runs using the CLI tool. Learn more and download the tool from the <u>AWS</u> HealthOmics Tool Github repository.

The following is an example of using the tool to rerun a workflow run, using the run ID. You can retrieve an ID for a run the CloudWatch logs.

```
omics-rerun 9876543 --name workflow name --retention-mode REMOVE
```

If the run exists in CloudWatch, you receive a response similar to the following.

```
Original request:
  "workflowId": "9679729",
  "roleArn": "arn:aws:iam::123456789012:role/DemoRole",
  "name": "sample_rerun",
  "parameters": {
    "image": "123456789012.dkr.ecr.us-west-2.amazonaws.com/default:latest",
    "file1": "omics://123456789012.storage.us-west-2.amazonaws.com/8647780323/
readSet/6389608538"
  },
  "outputUri": "s3://workflow-output-bcf2fcb1"
}
StartRun request:
  "workflowId": "9679729",
  "roleArn": "arn:aws:iam::123456789012:role/DemoRole",
  "name": "new test",
  "parameters": {
    "image": "123456789012.dkr.ecr.us-west-2.amazonaws.com/default:latest",
    "file1": "omics://123456789012.storage.us-west-2.amazonaws.com/8647780323/
readSet/6389608538"
  },
  "outputUri": "s3://workflow-output-bcf2fcb1"
}
StartRun response:
  "arn": "arn:aws:omics:us-west-2:123456789012:run/9171779",
  "id": "9171779",
  "status": "PENDING",
  "tags": {}
```

}

If the workflow no longer exists, you receive an error message.

#### Creating and working with run groups

Run groups are optional. They limit the compute resources that are used during each run. You can set the maximum vCPU, maximum duration, or maximum concurrent runs to help limit your use of compute resources.

To create a run group, use the **create-run-group** API operation to create a run group named TestRunGroup. The following example uses a maximum of 20 CPUs, 10 GPUs, and a max duration of 600 minutes as parameters.

```
aws omics create-run-group --name TestRunGroup \
--max-cpus 20 \
--maxgpus 10 \
--max-duration 600
```

The response from this API operation is the newly created RunGroup ID. Use this ID to get additional details with the **get-run-group** API operation, as shown. To run the example, replace the **run group id** with the ID of the run group you want to learn more about

```
aws omics get-run-group --id run group id
```

The response shows the setting for maximum GPUs for the run group, and shows whether any tags are assigned.

```
{
    "arn": "arn:aws:omics:us-west-2:123456789012:runGroup/9938959",
    "id": "9938959",
    "name": "RunGroupMaxGpuTest",
    "maxGpus": 2,
    "creationTime": "2023-04-07T16:39:47.641002+00:00",
    "tags": {}
}
```

You can also use the **list-run-group** API operation to view all created run groups.

```
aws omics list-run-groups
```

### **Deleting workflows and runs**

When you no longer need a workflow, run, or run group, you can delete it by using the AWS CLI, API, or console. Workflows can only be deleted when they are listed in ACTIVE or FAILED status, and deleting a workflow does not affect any ongoing runs that are using the workflow.

The following example shows how you can use the AWS CLI command to delete a workflow. You won't receive a response. To run the example, replace the *workflow id* with the ID of the workflow you want to delete.

```
aws omics delete-workflow --id workflow id
```

In addition to deleting a run, you can also cancel a run. To cancel a run, its status must be PENDING, STARTING, RUNNING, or STOPPING. The following AWS CLI command shows how you can cancel a run. To run the example, replace the *run id* with the ID of the run you would like to cancel. If successful, there is no response.

```
aws omics cancel-run --id run id
```

The following AWS CLI command deletes a run. Runs can only be deleted if they are complete or canceled. To run the example, replace the xun id with the ID of the run you want to delete. There is no response if the run is successfully deleted.

```
aws omics delete-run --id run id
```

You can also delete run groups. Run groups can only be deleted if there are no runs associated with that run group with the status of PENDING, STARTING, RUNNING, or STOPPING.

The following example shows how you can use the AWS CLI to delete a run group. You will not receive a response. To run the example, replace the *run group id* with the ID of the run group you want to delete.

```
aws omics delete-run-group --id run group id
```

# Using the CloudWatch Logs for troubleshooting

The CloudWatch Logs include run, task, and engine logs, which can be used to get updates on run progress or troubleshoot failed runs. The engine logs provide a detailed log of the data

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processing steps and analyses, and can be used to identify and correct errors. They are also useful for improving reproducibility and maintaining compliance with regulatory requirements.

#### To view the CloudWatch Logs for workflows using the console

- 1. Open the AWS HealthOmics console https://console.aws.amazon.com/omics/.
- 2. On the AWS HealthOmics home page, choose

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in the upper left corner of the screen to open the navigation pane. Select Runs.

- 3. Select the run from the runs list, which is organized by run ID.
- 4. When the run details page opens, choose **View Cloudwatch logs** to view the run logs. This links you to the CloudWatch console.
- 5. From the tasks page, select **View Logstream** to be linked to the engine logs for a further breakdown of errors.

Tasks logs can also be found in your AWS account log under the /aws/omics/WorkflowLog log group. Engine logs are only generated for failed workflow runs, and are organized in the log stream by run ID and engine, run/{run-id}/task/{task-id}.

# **Troubleshooting**

The following documentation can help you troubleshoot problems you might have with your HealthOmics data stores and workflows.

#### **Topics**

- Why can't I run my workflow?
- Why can't I create a reference store?
- Why can't I create a sequence store?
- Why can't I create a workflow?
- Why did my task fail?
- Why can't I import my BAM, CRAM or FASTQ files?
- Why can't I import my VCF or gVCF files?
- Why can't I see my annotation store or variant store in Athena?
- Why can't I access my data store in Athena?
- Why do I get a "Request Too Long" error message when I try to create a workflow?
- Error and status messages for run failures

# Why can't I run my workflow?

If you're getting an error to run your workflow while using the AWS CLI, check to make sure you have the latest version installed. Also make sure you have the correct Amazon ECR and IAM permissions to access your input data.

A full log of failed tasks can be found in your engine logs, which can be accessed either through the console or your AWS account. Tasks logs can also be found in your AWS account log under the /aws/omics/WorkflowLog log group. Engine logs are only generated for failed workflow runs, and are organized in the log stream by Run ID and engine, run/{run-id}/task/{task-id}.

# Why can't I create a reference store?

If your receive an error that states "You don't have permissions for this action with the credentials you sent," check your IAM permissions. Confirm you have permission in your policy for the role you are using to use the CreateReferenceStore action.

# Why can't I create a sequence store?

If you receive an error that states "You don't have permissions for this action with the credentials you sent," check your IAM permissions. Confirm you have permission in your policy for the role you are using to use the CreateSequenceStore action.

## Why can't I create a workflow?

If the error states "Zip file contains multiple workflow definition files", you must specify the main/master WDL or Nextflow file to use. You can do this by entering in the main path to the file as part of the command --main folder\_name/file\_name through the AWS CLI. You can also do this using the main workflow's definition file field for the console.

# Why did my task fail?

Make sure that you have the appropriate service role. This role must be able to read from the Amazon Simple Storage Service location(s) or sequence store where your data resides. It must also have the appropriate trust policy for HealthOmics to assume the role. You can find more information in the CloudWatch Logs.

# Why can't I import my BAM, CRAM or FASTQ files?

Make sure that you have the appropriate service role. This role must be able to read from any Amazon S3 location where your data resides. If you are using a customer managed key(CM-CMK), you must also use the AWS KMS decrypt permissions. It must also have the appropriate trust policy for HealthOmics to assume the role.

# Why can't I import my VCF or gVCF files?

Make sure that you have the appropriate service role. This role must be able to read from any Amazon S3 location where your data resides. If you are using a customer managed key(CM-CMK), you must also use the AWS KMS decrypt permissions. It must also have the appropriate trust policy for HealthOmics to assume the role.

# Why can't I see my annotation store or variant store in Athena?

In Lake Formation, be sure to create a resource link based on the store that was shared with you. Once you create a resource link that you have permission to access, the store should be visible in Athena.

## Why can't I access my data store in Athena?

If your annotation or variant store is visible but you are receiving an error message saying that access is denied, check which query engine version you're using. Only queries run using engine version 3 are supported. To read more about Athena query engine versions, see the <a href="Managerial"><u>Amazon</u></a>
Athena documentation.

# Why do I get a "Request Too Long" error message when I try to create a workflow?

If you're creating a workflow with a definition zip embedded in the request, you may get a "Request Too Long" error message. You can work around this issue by uploading the definition zip to an Amazon S3 bucket.

# Error and status messages for run failures

To learn more about why a run a has failed, use the **GetRun** API operation.

To troubleshoot a run failure, refer to the following error codes and statuses. This table lists failures and messages.

#### Service error messages

Failure reason	Detailed error description
INSTANCE_RESERVATION_FAILED	There isn't enough instance capacity to complete the workflow run. Wait and try the workflow run again.
IMPORT_FAILED	The workflow run didn't finish because of a transient error while importing <i>uri</i> . Try the workflow run again.

Failure reason	Detailed error description
EXPORT_FAILED	The workflow run didn't finish because of a transient error while exporting the run output. Try the workflow run again.
INVALID_URI_INPUT	The URI structure isn't a valid <i>uri</i> . Check the URI structure and try again.
INVALID_S3_INPUT	The URI does not exist: <i>uri</i> . Check that the URI path exists and confirm that the role can access the object.
INVALID_OMICS_STORAGE_INPUT	The HealthOmics storage URI does not exist: <pre>uri</pre> . Check that the read set path exists and confirm that the role can access the read set.
INACTIVE_OMICS_STORAGE_RESOURCE	The HealthOmics storage URI is not in ACTIVE state. Activate the read set and try again. To learn more about activating read sets, see <a href="Activating read sets">Activating read sets</a> .
MODIFIED_INPUT_RESOURCE	The input URI was modified after the run started.
ECR_PERMISSION_ERROR	HealthOmics doesn't have permission to access the image URI. Confirm that the Amazon ECR private repository exists and has granted access to the HealthOmics service principal.
INVALID_ECR_IMAGE_URI	The Amazon ECR image URI structure isn't valid. Check for a valid URI and try again.
INVALID_TASK_RESOURCE_VALUE	The requested GPU, CPU, or memory is either too high for available compute capacity, or is less than the minimum value of 1 for task <i>ID</i> .

Failure reason	Detailed error description
OUT_OF_MEMORY_ERROR	The workflow task <i>ID</i> ran out of memory.  Increase the memory value in the workflow definition and try the run again.
RUN_TASK_FAILED	The workflow run failed because the task failed. To debug task failure, use the <b>GetRunTask</b> API operation and the Amazon CloudWatch Logs stream.
IMPORT_FAILED	The import failed. Check that the input file exists and the run role can access input.
EXPORT_FAILED	The export failed. Check that the output bucket exists and the run role has write permission to the bucket.
ASSUME_ROLE_FAILED	HealthOmics doesn't have permission to assume the role. Specify the HealthOmics principal in the trust relationship for the role.
UNSUPPORTED_INPUT_SIZE	The total input size is too high. Decrease the input size and try again.
SERVICE_ERROR	The workflow run didn't finish because of a transient service error. Try the workflow run again.

# **Quotas for AWS HealthOmics**

Your AWS account has default quotas, formerly referred to as limits, for each AWS service. Unless otherwise noted, each quota is Region-specific. You can request increases for some quotas, and other quotas cannot be increased.

To view the quotas for AWS HealthOmics, open the <u>Service Quotas console</u>. In the navigation pane, choose **AWS services** and select **AWS HealthOmics**.

To request a quota increase, see <u>Requesting a Quota Increase</u> in the <u>Service Quotas User Guide</u>. If the quota is not yet available in Service Quotas, use the <u>limit increase form</u>.

## **HealthOmics Storage quotas**

Your AWS account has the following quotas related to AWS HealthOmics for storage and reference stores. Where noted, some quotas can be adjusted by request.

Maximum resource or throttling per account or region	Default	Adjustable Yes/No
Sequence stores	20	Yes
Reference stores	1	No
Read sets per sequence store	1,000,000	Yes
References per reference store	50	Yes
Concurrent sequence or reference import jobs	5	Yes
Concurrent sequence or reference export jobs	5	Yes
Read sets per activation job	20	No
Concurrent activation jobs	25	Yes

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Maximum resource or throttling per account or region	Default	Adjustable Yes/No
Read set sources per import job	100	No
Read sets per export job	100	No
Max part size for a direct upload (MiB)	100	No
Max parts in file for direct upload	10000	No
CreateSequenceStore, CreateReferenceStore, DeleteSequenceStore, DeleteReferenceStore	1 TPS	Yes
GetSequenceStore, ListSeque nceStores	5 TPS	Yes
GetReadSet	30 TPS	Yes
GetReadSetMetadata, ListReadSets	5 TPS	Yes
BatchDeleteReadSet	1 TPS	Yes
StartReadSetImportJob, GetReadSetImportJob, ListReadSetImportJobs	5 TPS	Yes
StartReadSetExportJob, GetReadSetExportJob, ListReadSetExportJobs	5 TPS	Yes

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Maximum resource or throttling per account or region	Default	Adjustable Yes/No
GetReferenceStore, ListRefer enceStores	5 TPS	Yes
StartReferencetImportJob, GetReferenceImportJob, ListReferenceImportJobs	5 TPS	Yes
ListReferences, GetRefere nceMetadata	5 TPS	Yes
DeleteReference	1 TPS	Yes
StartReadsetActivationJob	1 TPS	Yes
ListReadsetActivationJobs, GetReadSetActivationJob	5 TPS	Yes
GetReference	10 TPS	Yes
CreateMultipartReadSetUploa d, CompleteMultipartR eadSetUpload, AbortMult ipartReadSetUpload	1 TPS	No
UploadReadSetPart	10 TPS	Yes
ListMultipartReadSetUploads, ListReadSetUploadParts	5 TPS	Yes
TagResource, UntagResource, ListTagsForResource	5 TPS	Yes

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# **HealthOmics Analytics quotas**

Your AWS account has the following quotas related to AWS HealthOmics for analytics. Where noted, some quotas can be adjusted by request.

Maximum resource or throttling per account or region	Default	Adjustable Yes/No
Variant stores	10	Yes
Annotation stores	10	Yes
Concurrent variant or annotation store import jobs	5	Yes
Files per variant store import job	1000	Yes
Files per annotation store import job	1	No
Variant store import job file size (GB)	20	Yes
Annotation store import file size (GB)	20	Yes
Maximum number of annotation store versions	10	Yes
Maximum number of shares for annotation or variant store	10	Yes
CreateVariantStore, DeleteVar iantStore, GetVariantStore, ListVariantStores, UpdateVar iantStore	1 TPS	No

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Maximum resource or throttling per account or region	Default	Adjustable Yes/No
StartVariantImportJob, CancelVariantImportJob, GetVariantImportJob, ListVariantImportJobs	1 TPS	No
CreateAnnotationStore, DeleteAnnotationStore, GetAnnotationStore, ListAnnotationStores, UpdateAnnotationStore	1 TPS	No
StartAnnotationImportJob, ListAnnotationImportJobs, GetAnnotationImportJob, CancelAnnotationImportJob	1 TPS	No

# **HealthOmics Workflows quotas**

Your AWS account has the following quotas related to AWS HealthOmics for workflows. All quotas can be adjusted by request.

Maximum resource or throttling per account or region	Default	Adjustable Yes/No
Number of workflows	100	Yes
Number of workflow runs (active or inactive)	5000	Yes
Number of run groups	10	Yes
Active workflow runs	10	Yes

Maximum resource or throttling per account or region	Default	Adjustable Yes/No
Concurrent tasks per run	25	Yes
Concurrent active CPUs	3000	Yes
Workflow run duration (hours)	168	Yes
Run storage capacity (GB)	9600	Yes
StartRun	0.1 TPS	Yes
CreateWorkflow	5	Yes
CancelRun, DeleteRun , GetRun, GetRunTask, ListRunTasks, ListRuns	10 TPS	Yes
CreateRunGroup, DeleteRun Group, GetRunGroup, ListRunGroups, UpdateRun Group	10 TPS	Yes
DeleteWorkflow, GetWorkfl ow, ListWorkflows, UpdateWorkflow	10 TPS	Yes

# **Document history for the AWS HealthOmics User Guide**

The following table describes the documentation releases for AWS HealthOmics.

Change	Description	Date
New Features	AWS HealthOmics added support for S3 access to reference and sequence stores, and support for SHA256 ETags.	April 15, 2024
New Features	AWS HealthOmics added entity tags (ETags) for sequence stores.	October 6, 2023
New Features	AWS HealthOmics added annotation store versioning and analytic store sharing.	August 15, 2023
New Features	AWS HealthOmics added Common Workflow Language (CWL) as a supported language for HealthOmics workflows.	June 30, 2023
New Features	AWS HealthOmics added new Ready2Run workflows, GPU support for workflows, data parsing for annotation stores, direct upload into HealthOmi cs storage, and integration with EventBridge.	May 15, 2023
New managed policy	AWS HealthOmics added a new managed policy that provides full access. To learn	February 23, 2023

	more, see <u>AWS managed</u> <u>policies</u> .	
New managed policy	AWS HealthOmics added a new managed policy that limits access to read only. To learn more, see AWS managed policies.	November 29, 2022
<u>Initial release</u>	Initial release of the AWS HealthOmics User Guide	November 29, 2022