
AWS
re:Invent
NOV. 28 – DEC. 2, 2022 | LAS VEGAS, NV

LFS304

Learn how to use the new Amazon Omics

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Agenda

- Introduction to AWS for Health
- Overview of Amazon Omics
- Technical hands-on session
- Omics sequence storage
- Omics workflows
- Omics analytics storage

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AWS for Health



Omics



Healthcare



Life sciences

Accelerating innovations from benchtop to bedside

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Solving for customer pain points



Petabyte-scale storage is challenging and costly.

Workflow management is hard at scale, but I want to use the tools and workflow languages I am comfortable with.

I want to use general-purpose data science methods and tools on genomics data, but it's hard to make that bridge.

Compliance (both privacy/security and QMS) is critical.

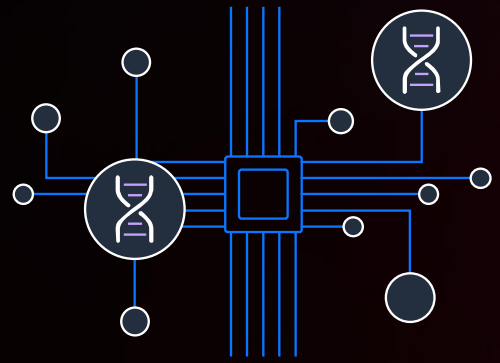
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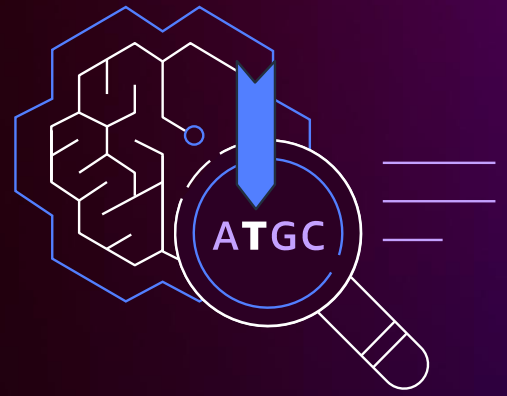
What is Amazon Omics?



Genomics-aware object storage



Scalable compute to process genomic data



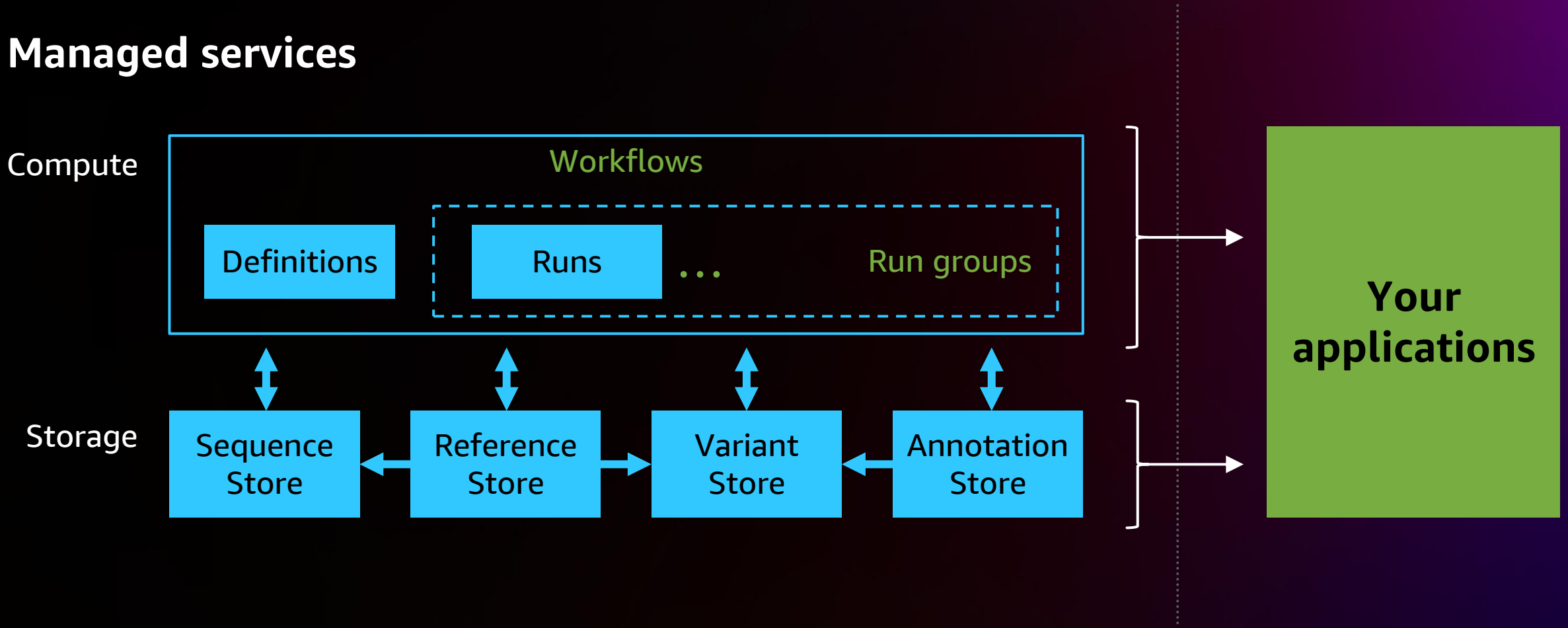
Query-ready variants and annotations

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What is Amazon Omics?

Managed services



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What we are going to build

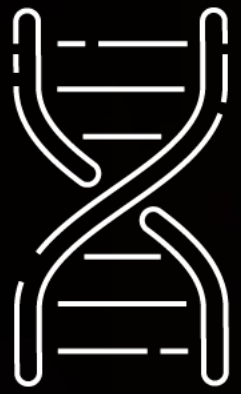
END-TO-END GENOMIC DATA JOURNEY USING AMAZON OMICS



Create sequence and reference stores from existing Amazon S3

Create and run workflows to process sequence data

Create and query variant and annotation stores







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Let’s build!

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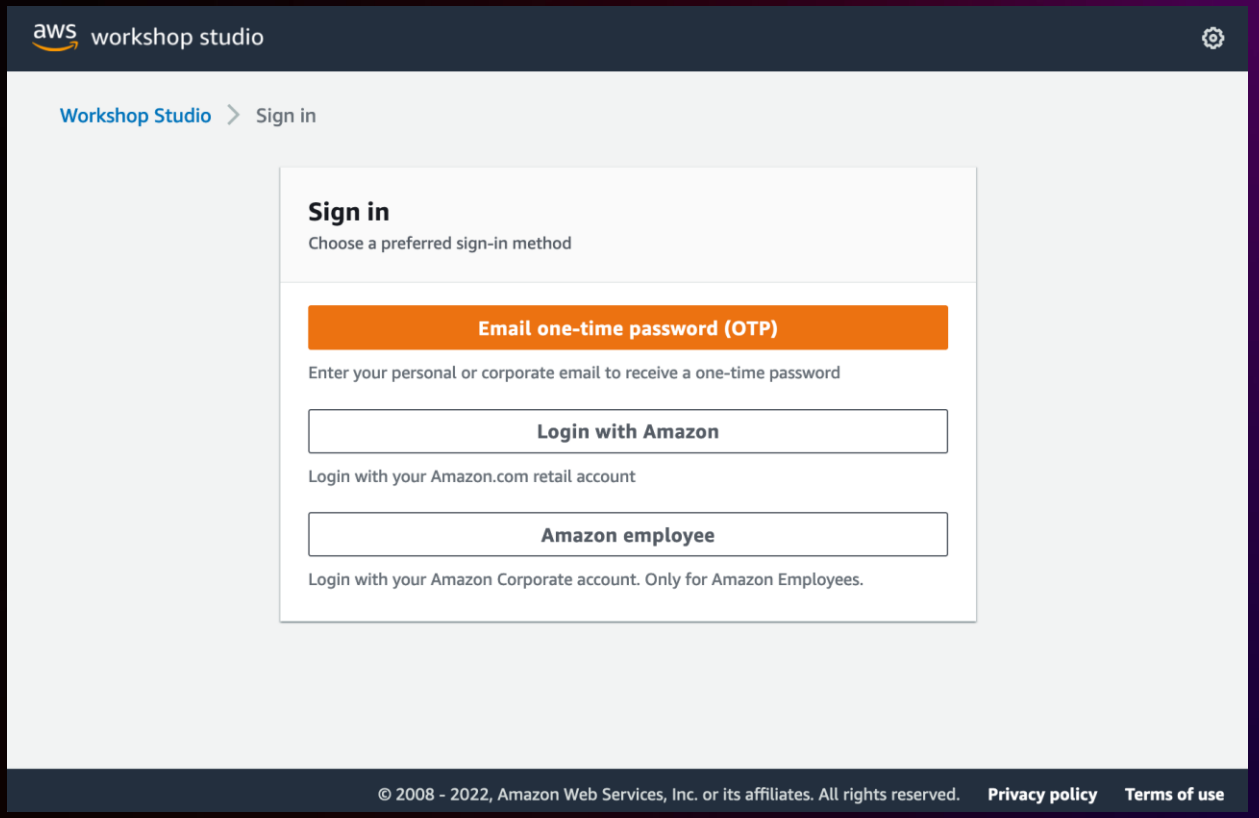
Getting started with this workshop

- As a participant, you will have access to an AWS account with any optional pre-provisioned infrastructure and IAM policies needed to complete this workshop
- The AWS account will only be available for the duration of this workshop; you will lose access to the account thereafter
- The optional pre-provisioned infrastructure will be deployed to a specific Region; check your workshop content to determine whether other Regions will be used
- Be sure to review the terms and conditions of the event; do not upload any personal or confidential information in the account

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Sign in via your preferred method

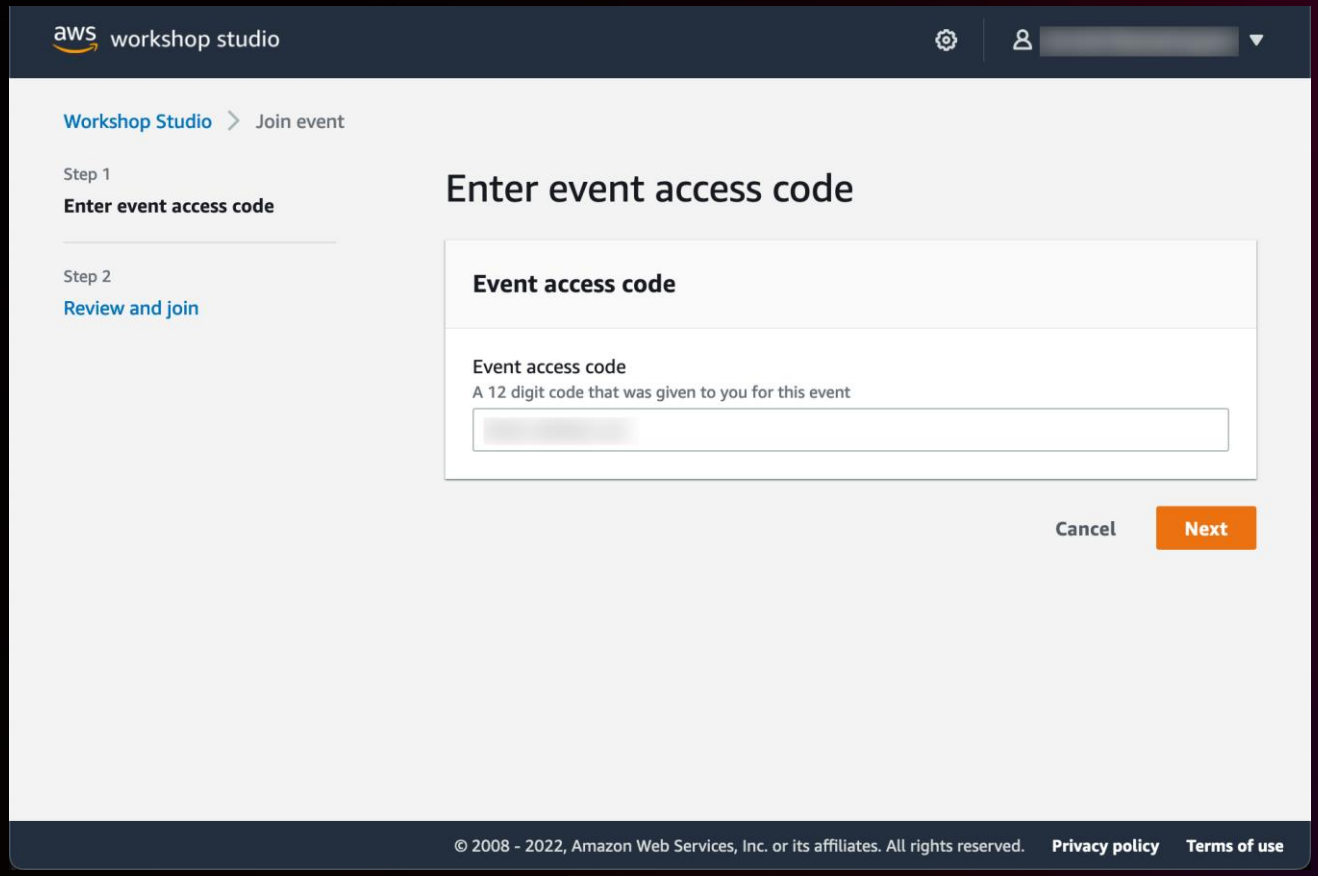
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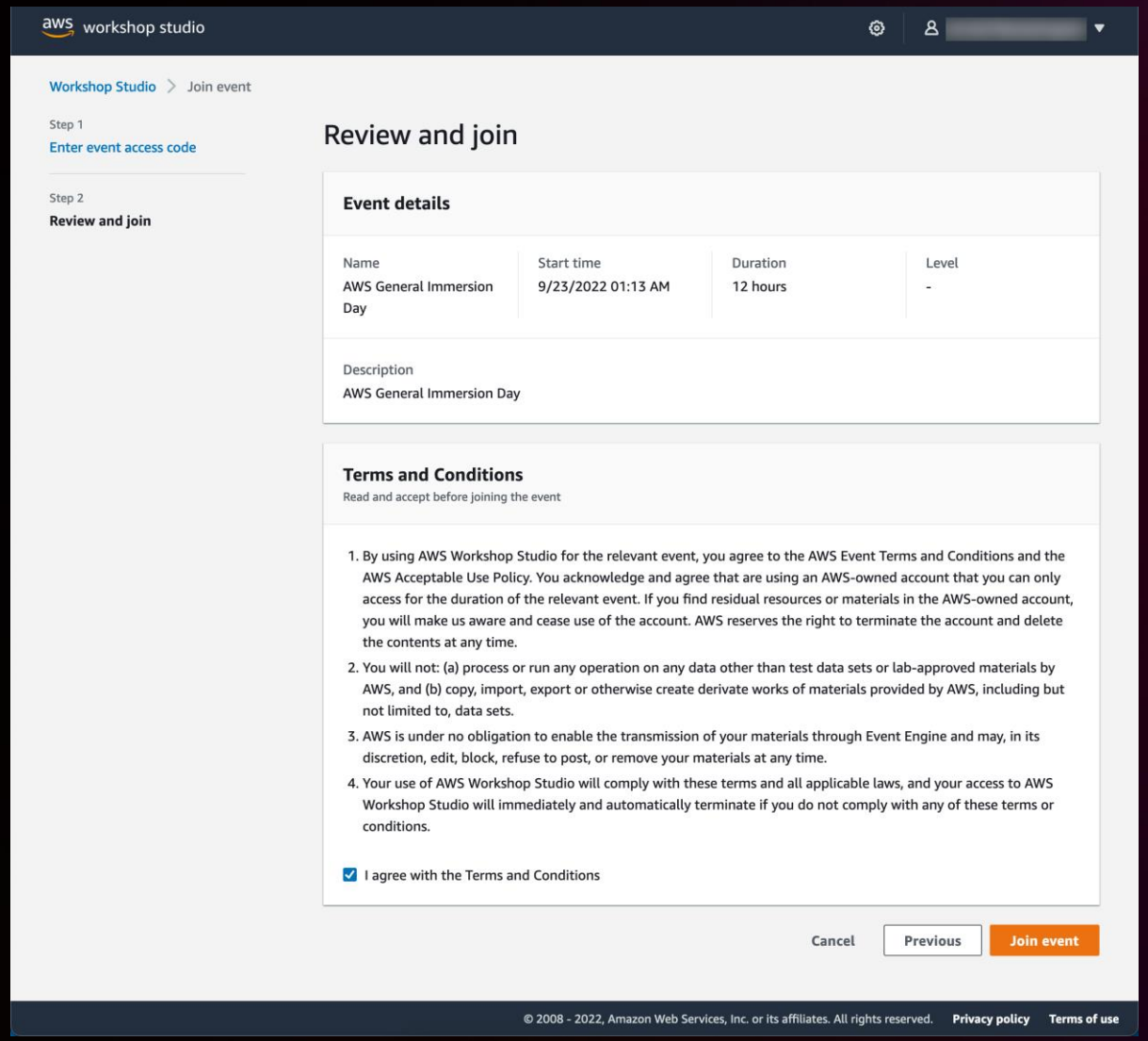
Enter event access code

Enter 12-digit event access code; if you were given a one-click join link, you can skip this step



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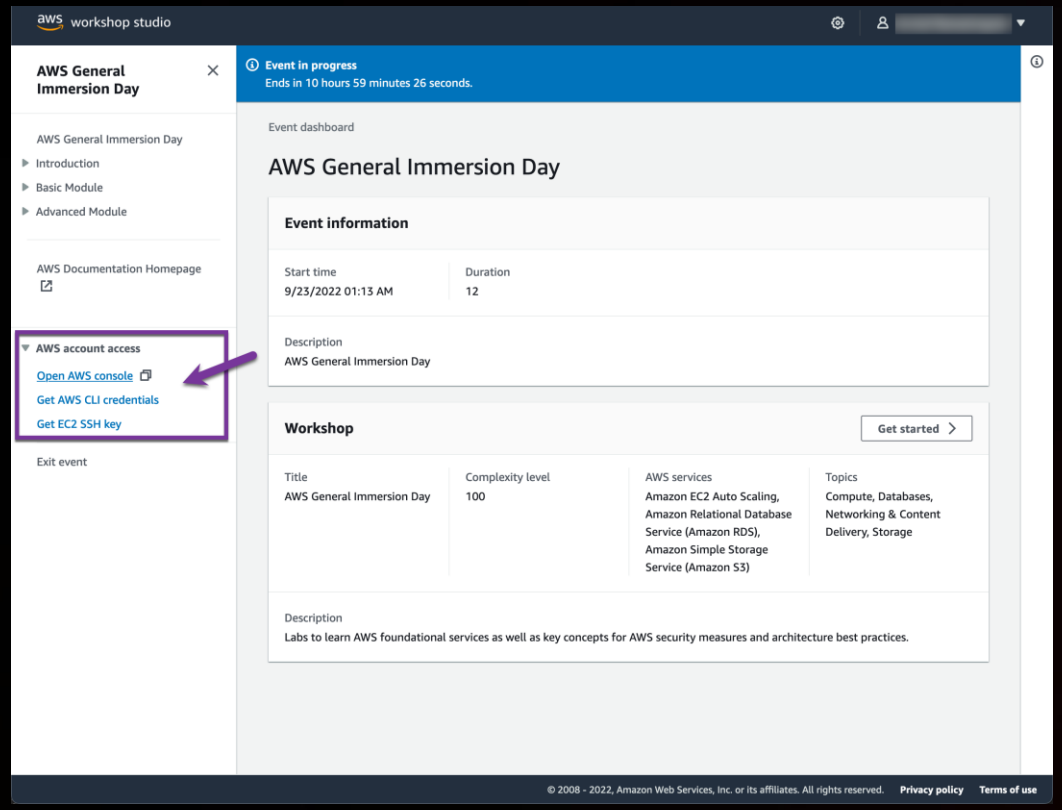
Review terms and join event

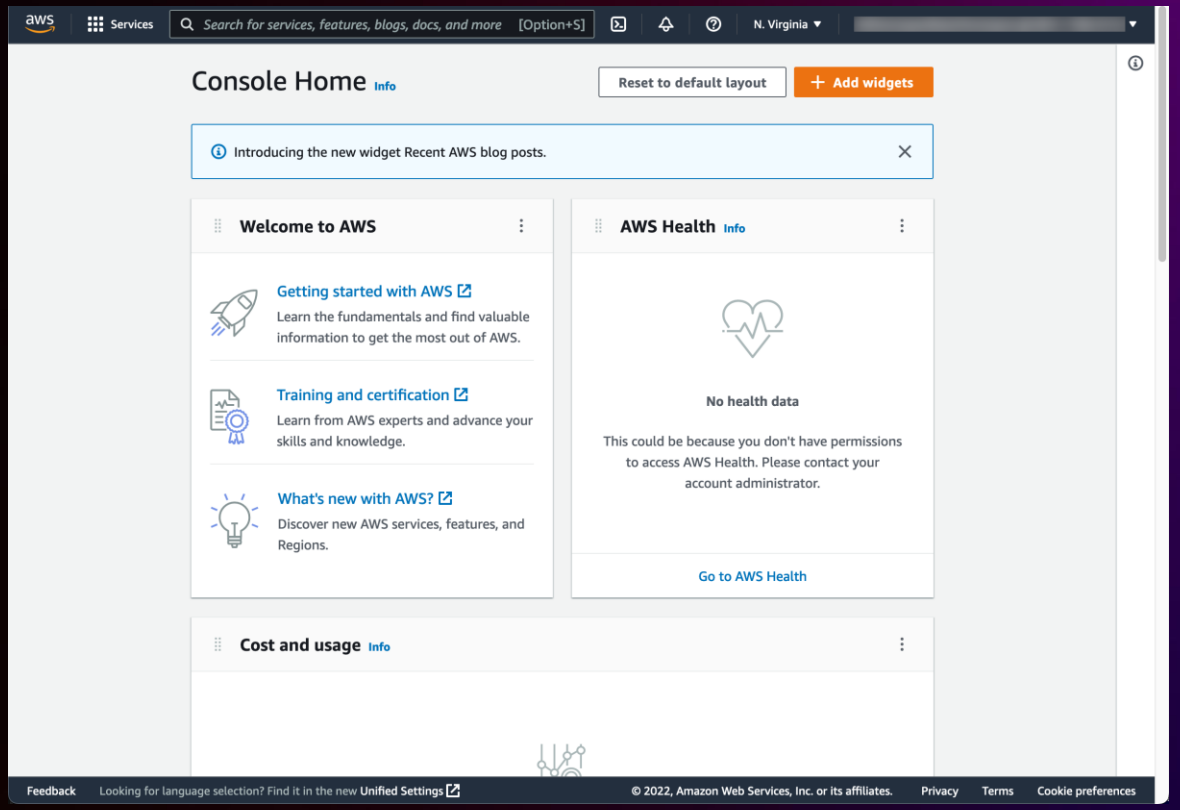


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Access AWS account

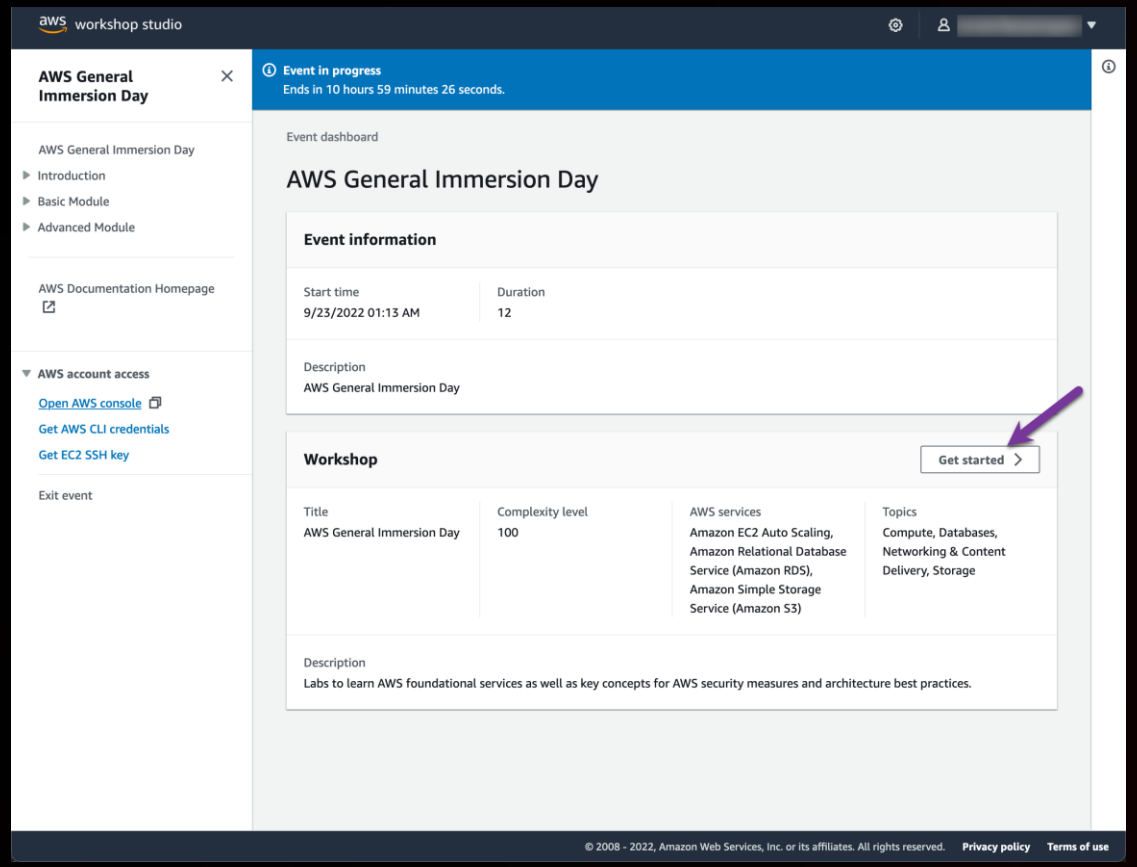
ACCESS THE AWS CONSOLE, OR GENERATE AWS CLI CREDENTIALS AS NEEDED

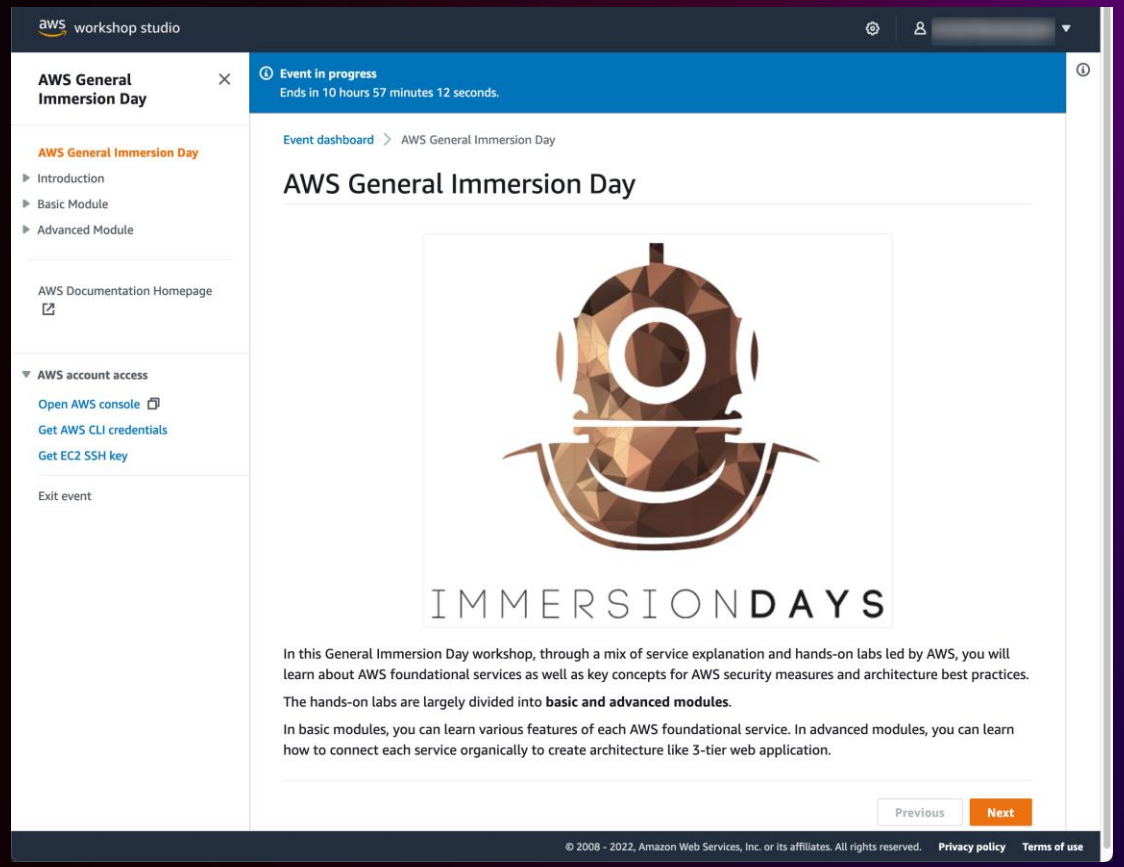




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Get started with the workshop

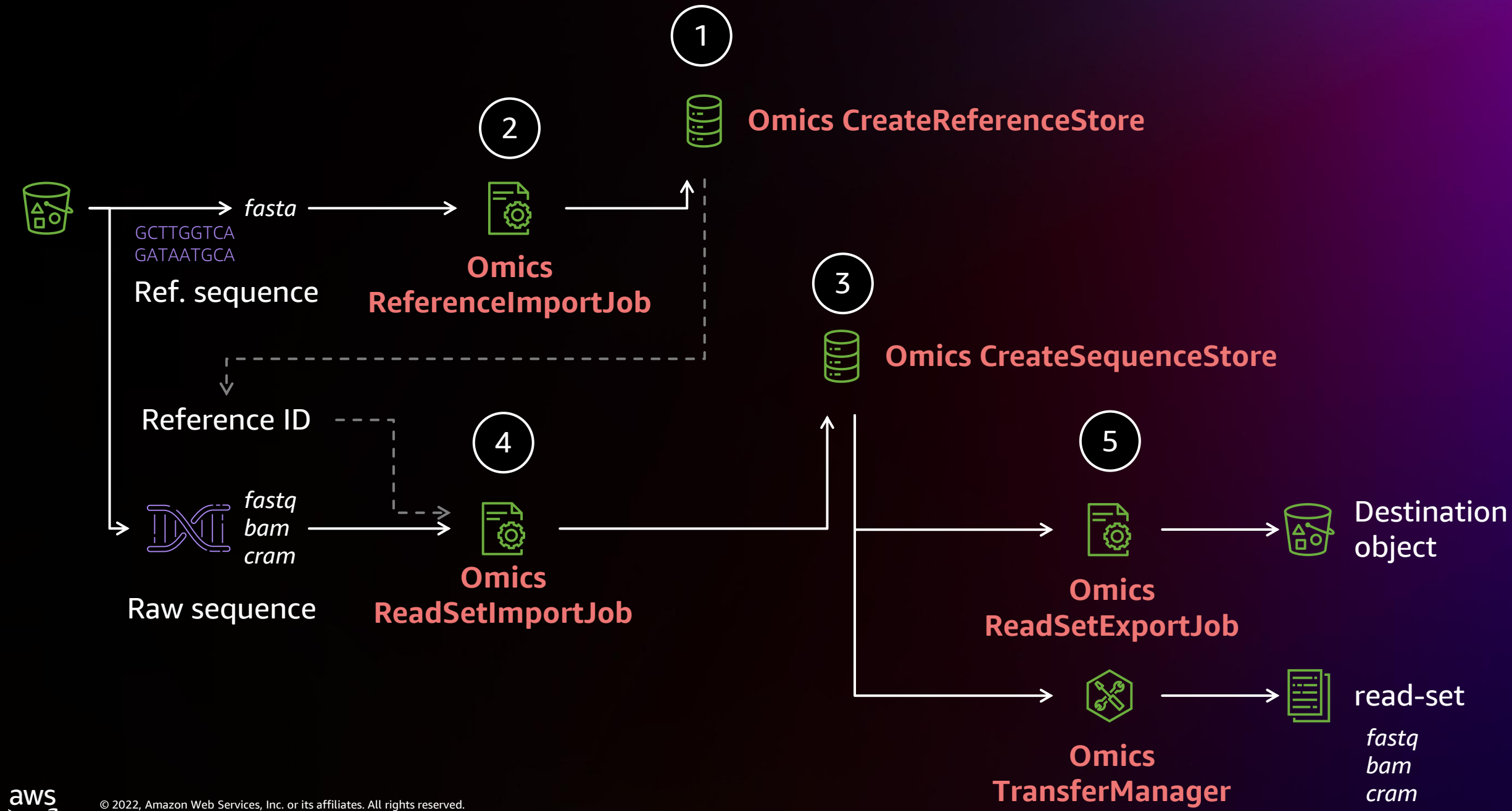




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Omics sequence storage

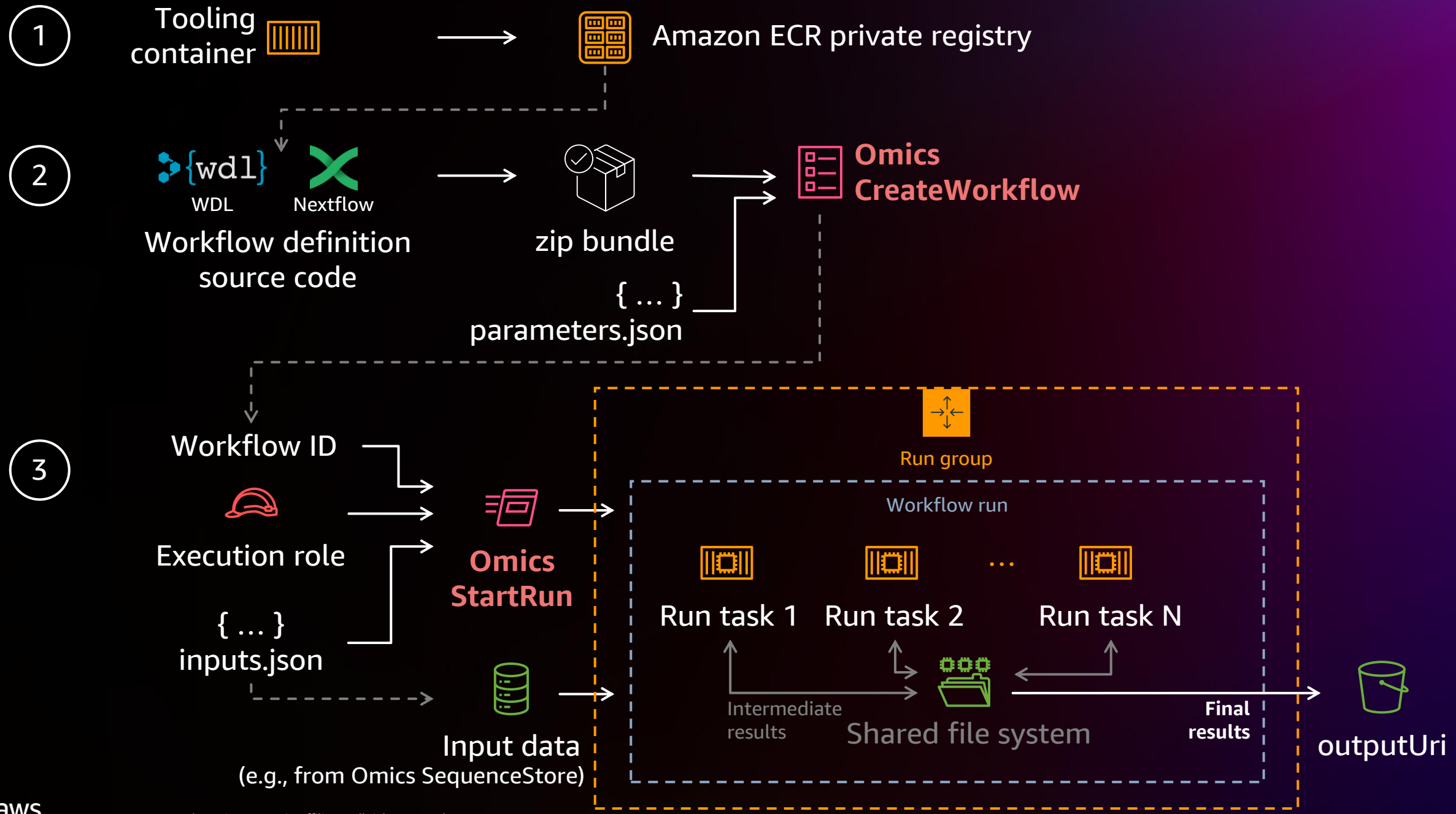
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Omics workflows

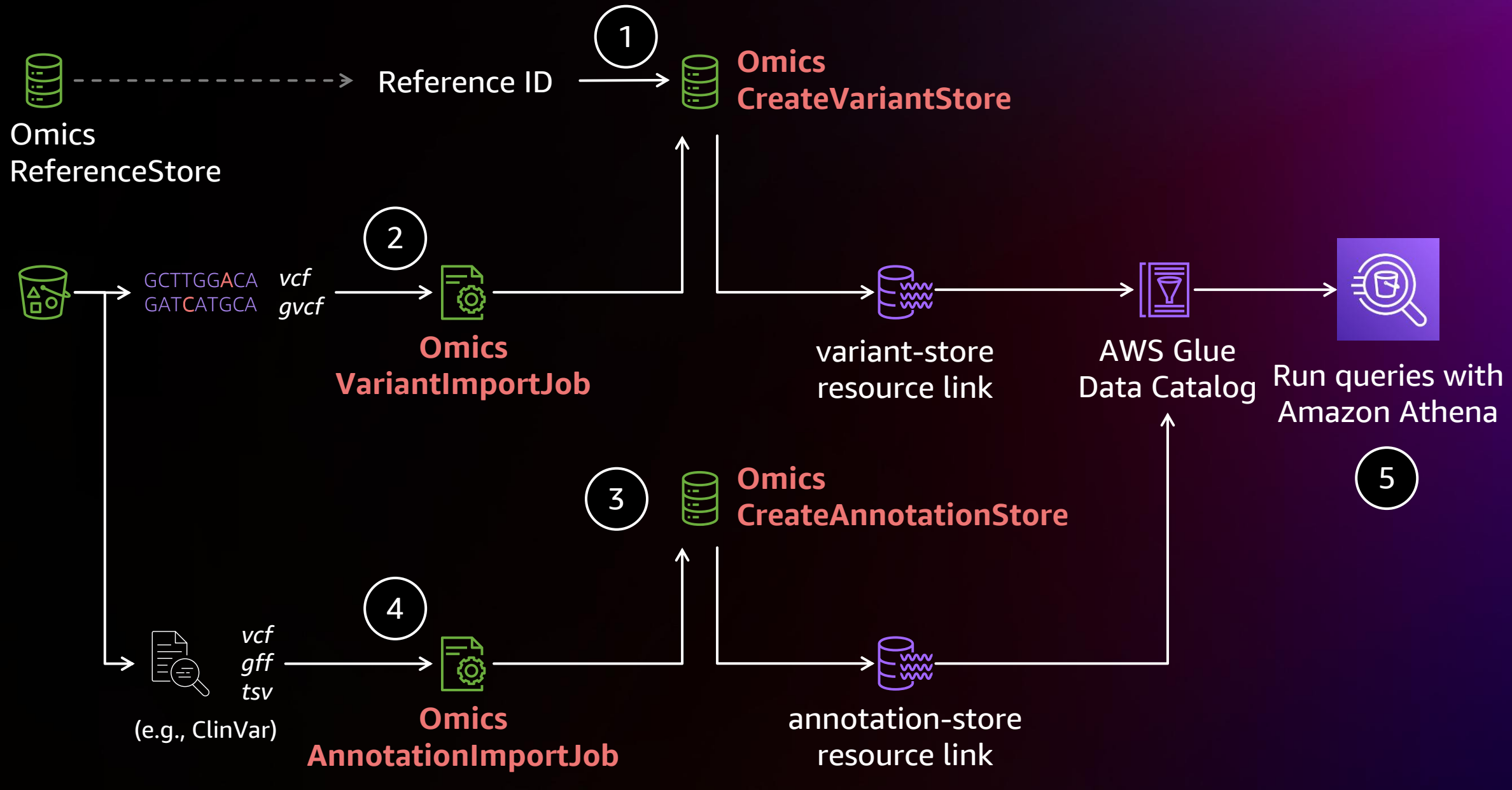
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Omics analytics storage

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Wrap-up

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Recap – What we built

End-to-end genomic data journey using Amazon Omics



Stored raw sequencing data

GCTTGGTCA
GATAATGCA
fasta
Ref. sequence
Omics ReferenceStore

fastq
bam
cram
Raw sequence
Omics SequenceStore

Ran analysis workflows

{wdl}
Workflow Definition Language
Nextflow
Omics workflow

Stored and queried variant data

GCTTGGACA
GATCATGCA
vcf
gvcf
Variants
Omics VariantStore

vcf
gff
tsv
Annotations
Omics AnnotationStore

Query and interpret with Amazon Athena

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AWS and Children's National Hospital

Customer obsession = Patient obsession

Collect pins at the AWS for Health Lounge from child artists who are also patients at Children's National Hospital – learn about their inspiration and their why





Artwork provided by Zahara, age 13, patient with sickle cell

"I drew a cherry blossom because I love going to the DC Cherry Blossom Festival in the spring. The care I get at Children's National allows me to do all the things I love and have lots of fun!"

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AWS for Health Lounge

Connect with **AWS health experts** and other attendees in our dedicated lounge space for **informal discussions and networking**

Explore **AWS healthcare and life sciences demos** and new additions to the AWS for Health portfolio!



Location: AWS Industries Networking Lounges, Caesars Forum, Main Hall

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Thank you!



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