Amazon Omics

Transform genomic and biological data into insights

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Today’s Agenda

Background

Introducing Amazon Omics

Features

Use cases

Demo

Customers

Resources

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Computing as a utility

Focus on applications and not infrastructure

Pay as you go, and only for what you use

On Demand and fit for purpose

AWS Global Infrastructure

The AWS Cloud spans 99 Availability Zones within 31 geographic regions around the world, with announced plans for 12 more Availability Zones and 4 more AWS Regions in Canada, Israel, New Zealand, and Thailand.

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Customer benefits of AWS

Security Availability Performance Scalability Flexibility

Low cost

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Customer obsessed

90%

of our roadmap originates with customer requests and are designed to meet specific needs

96%

of R1 Research Institutions are using AWS

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How AWS can help your research

Science, not servers
Use compute when you need
It to do large-scale analysis

Collaboration
Access data sets that span institutions

Share effort
Leverage work done by
other scientists to save time

Reproduce research
A common platform for
reproducing scientific analyses

State-of-the-art analytics
Use data science methods
in your research

Security
A collection of tools to protect
data and privacy

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Why AWS for genomics

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Challenges

Genomic and biological data has the potential to transform how we treat disease—but its scale is complicated and costly to manage

- Tens of millions of whole genomes to be sequenced and stored in the next 5 years
- Multiple specialized tools and workflow languages required
- Special security, privacy, and compliance requirements mandated

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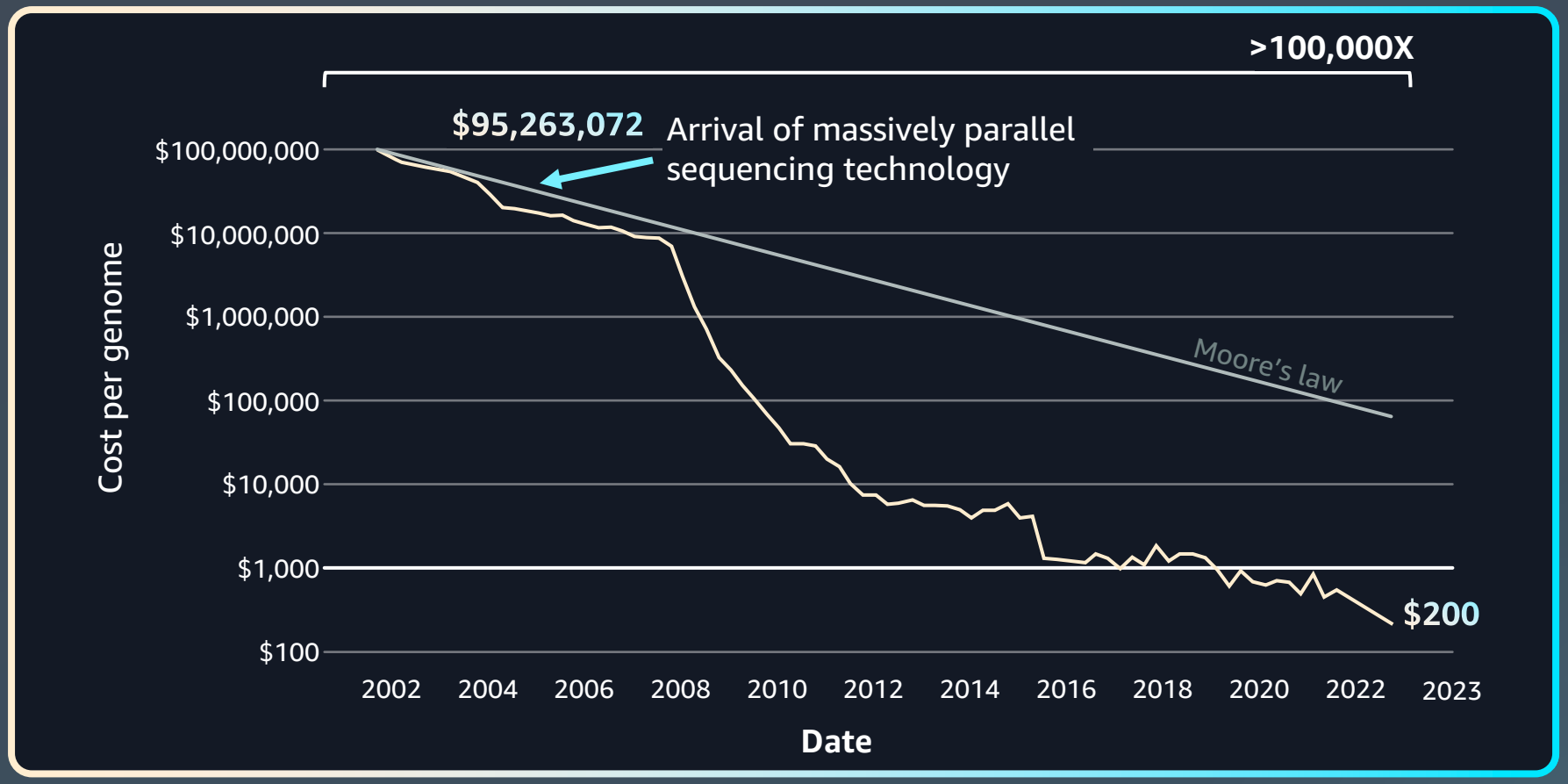
Cost per human genome



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Amazon Omics

A purpose-built service to help healthcare and life science organizations and their software partners **store, query, and analyze** genomics, transcriptomic, and other omics data and then **generate insights** from that data **to improve health and advance scientific discoveries**

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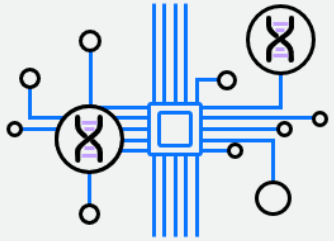
Benefits

Multiomic and multimodal analysis

Population-level scale



Fully managed bioinformatics computation

Built-in security, privacy, and compliance

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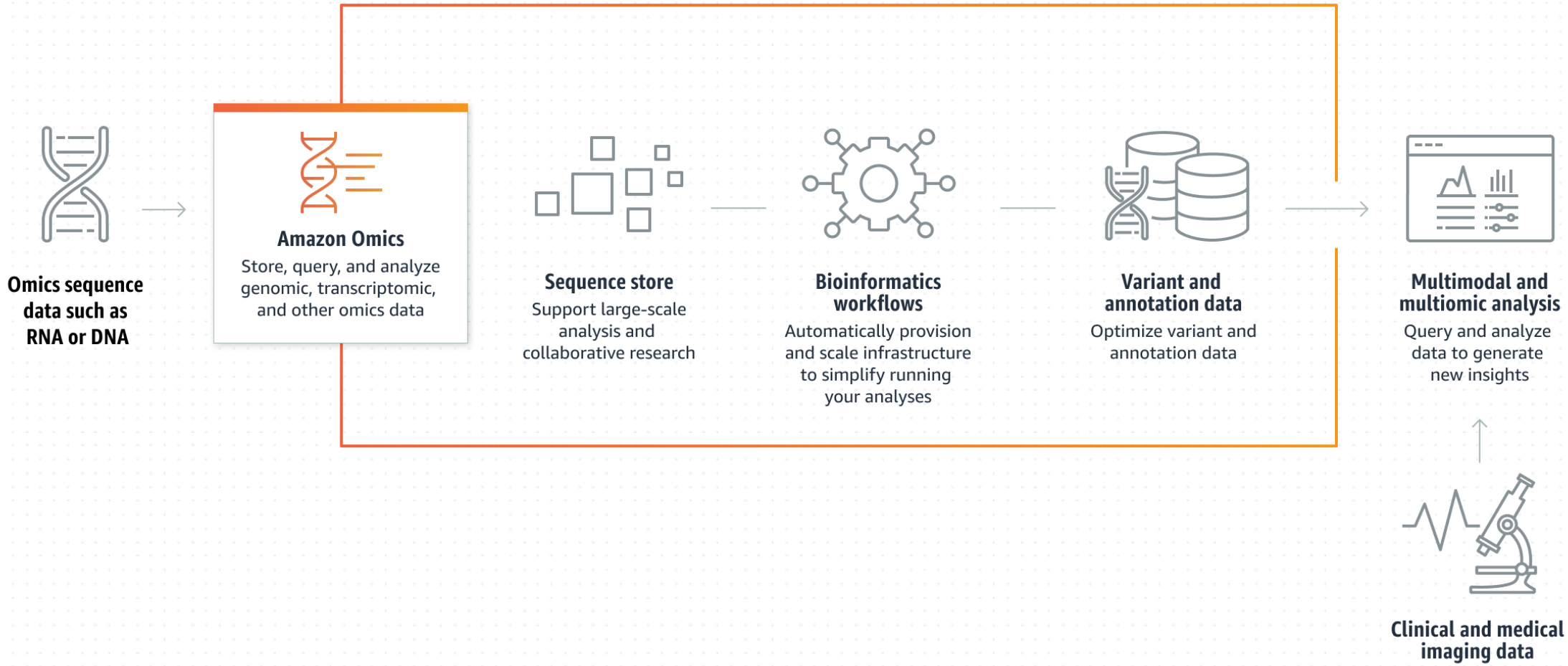
How it works



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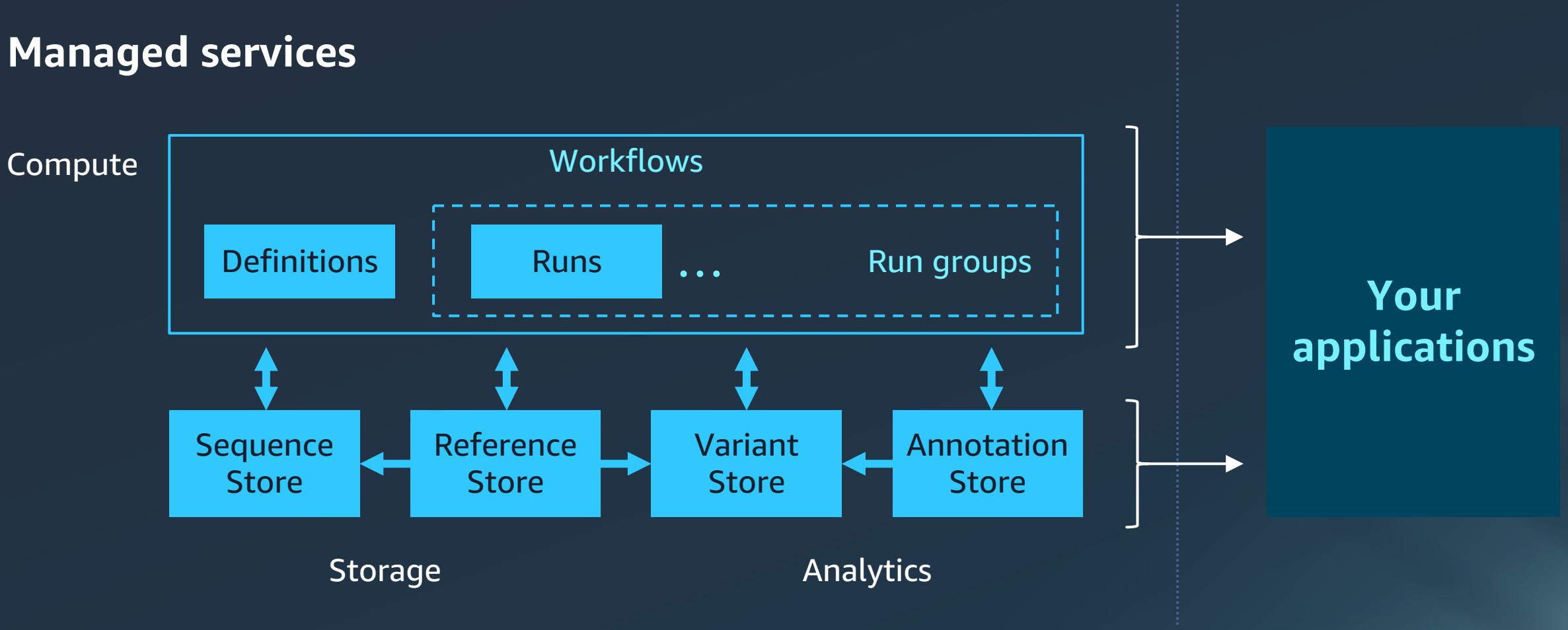
What is Amazon Omics?

Managed services



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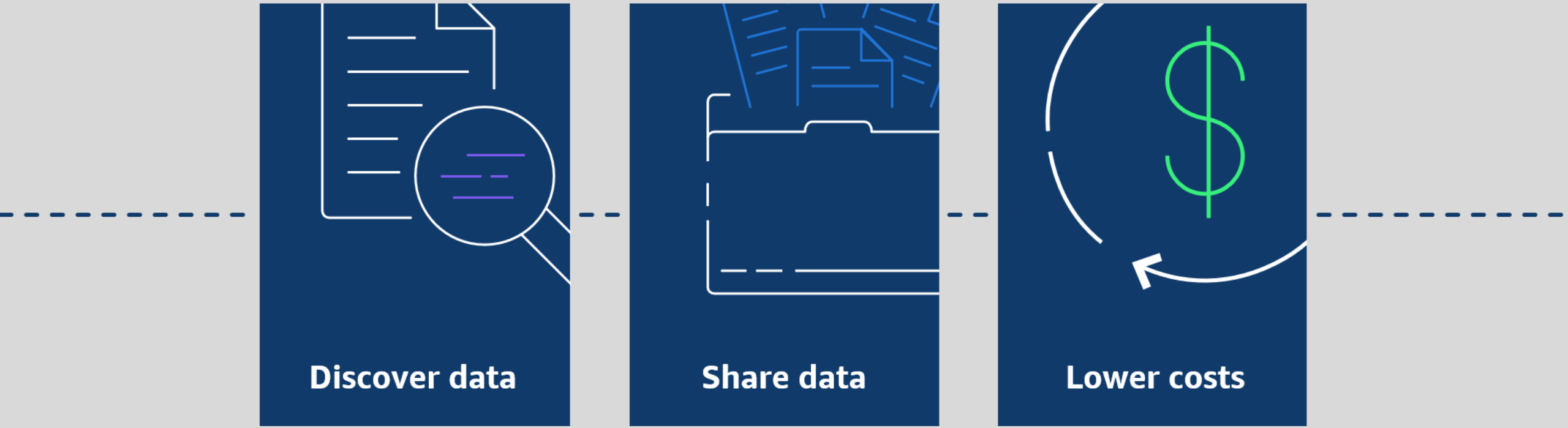
Storage



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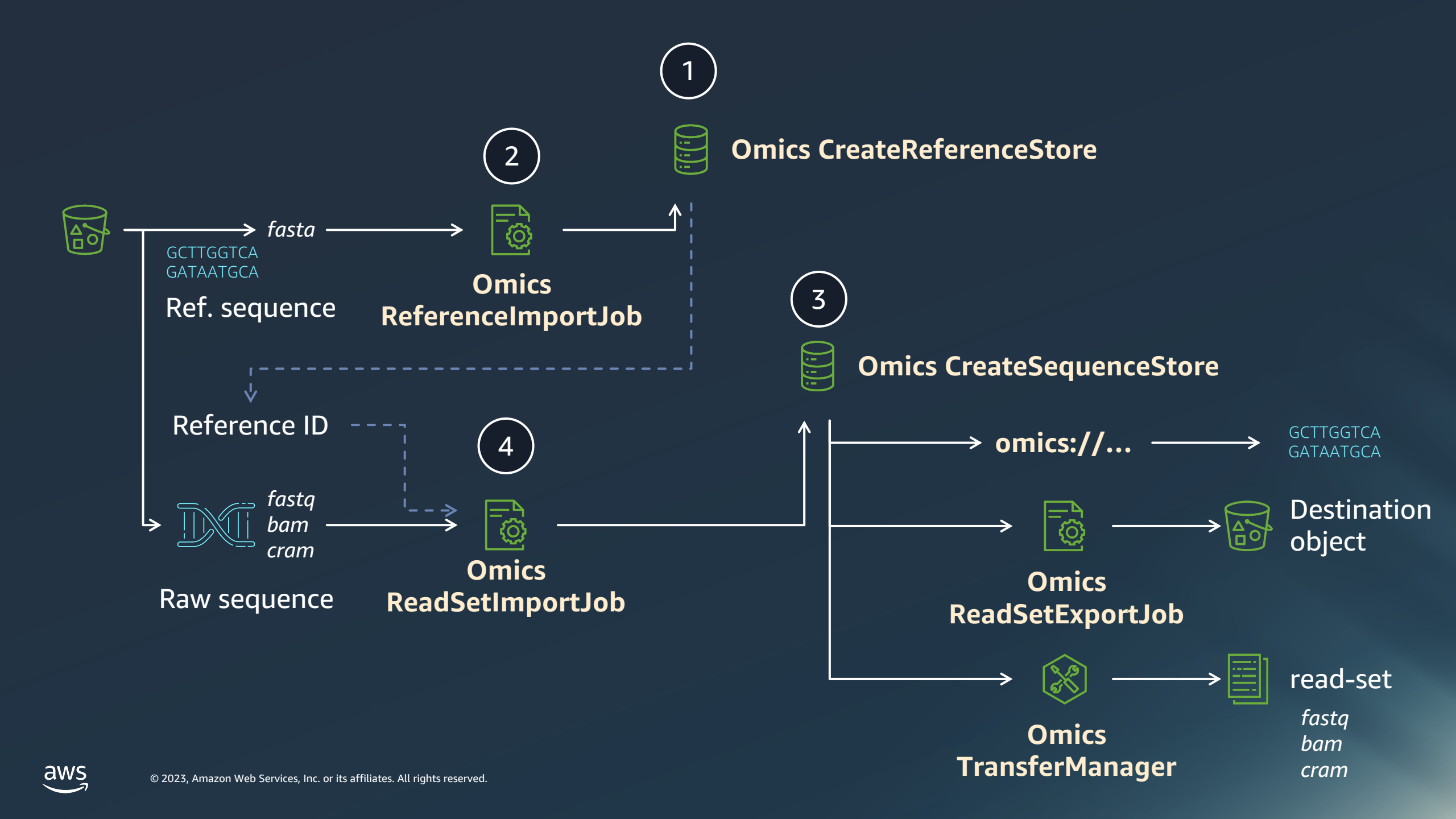

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Workflows

Science not infrastructure

Pay-as-you-go pricing

Predictable cost per workflow

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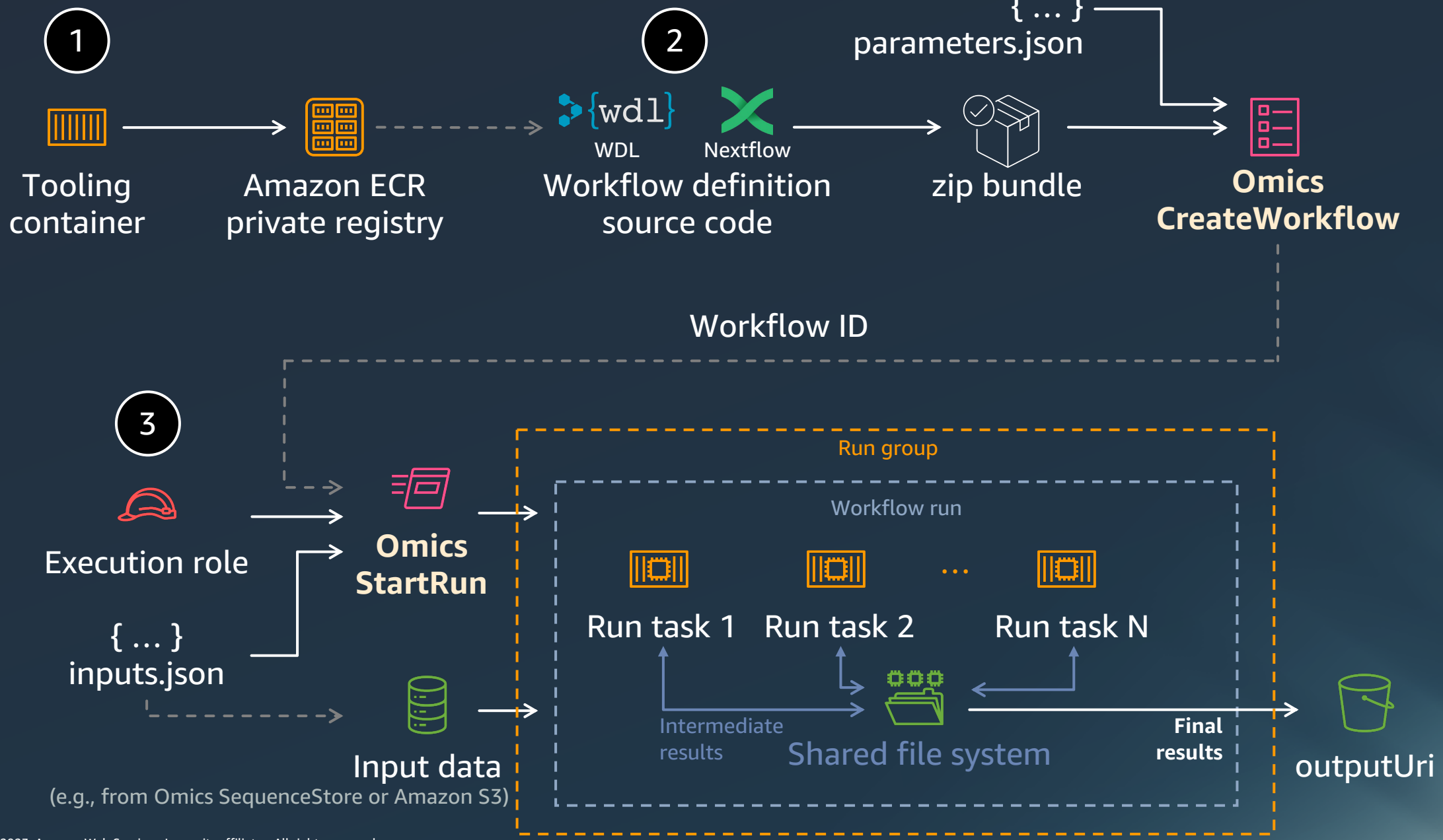

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Analytics

Ingests variant and annotation data

Allows fine-grained access control

Scales with you

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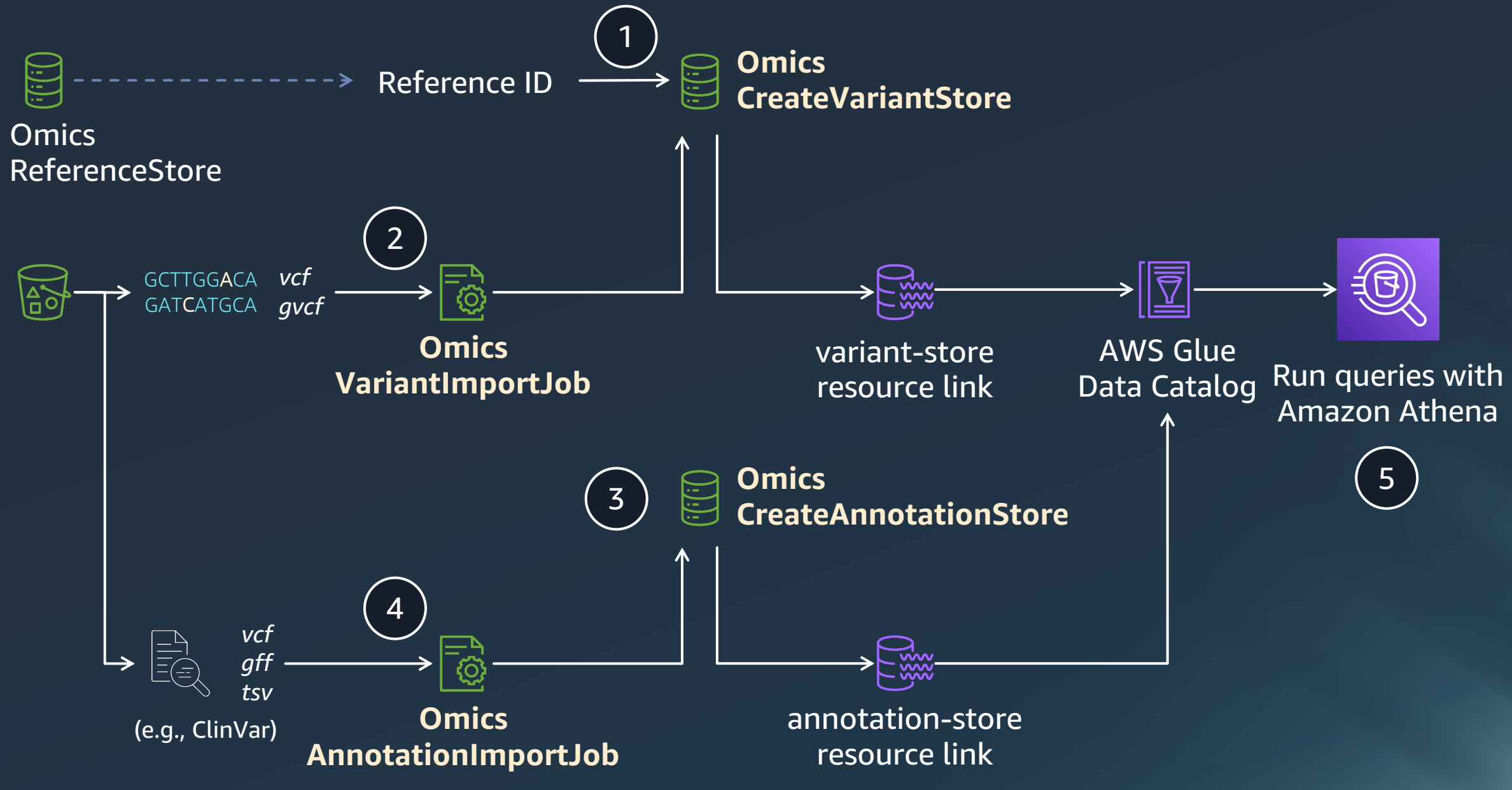

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Top use cases

Scale population sequencing

Understand how genomic variation maps to phenotypes across a population

Improve clinical genomics workflows

Build reproducible and traceable clinical genomics workflows

Accelerate clinical trials

Integrate genome analysis into clinical trials to test new drug candidates' efficacy

Enhance research and innovation

Streamline and control storage, access, and analysis of anonymized genomic data

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Demo!

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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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Registry of Open Data on AWS

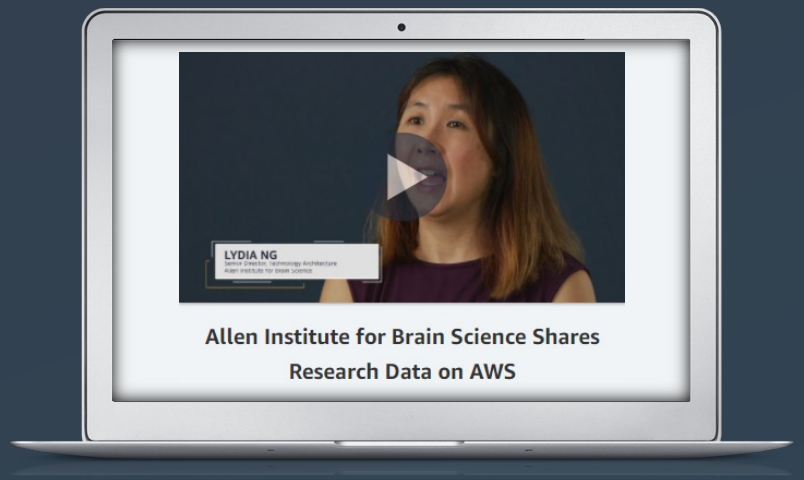
Amazon S3 buckets

Amazon SNS topics

Amazon RDS snapshots

Amazon EBS volumes

Direct data access from AWS native interfaces



More than 800 usage examples, tutorials, and publications

registry.opendata.aws

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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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Recap – What it cost

	Size	Cost dimension	Cost
ref. sequence	3 Gbase	Free	$ 0.00
raw sequence	1.184 Gbase	$ Gbase-month	$ 0.0068
workflow run *(GATK on NA12878)*	# vCPUs/task # GB RAM/task	$ omics instance/hr	$ 0.521
	1.2 TB storage 2 hrs total run	$ GB (Storage)-hr	$ 0.507
variants	0.009 GB	$ GB-month	$ 0.0023
annotations	0.050 GB		
			$ 1.04 total

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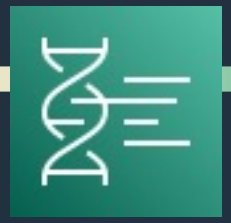
Multimodal analytics

Purpose-built services for healthcare and life sciences



Amazon Omics

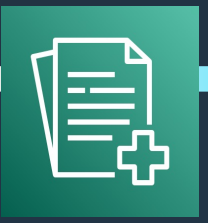
Transform genomic, transcriptomic, and other omics data into insights

Amazon HealthLake Imaging and Analytics

Provide a complete view of individual or patient population health data

Amazon Comprehend Medical

Understand medical context using natural language processing



Amazon Transcribe Medical

Automatically convert medical speech to text

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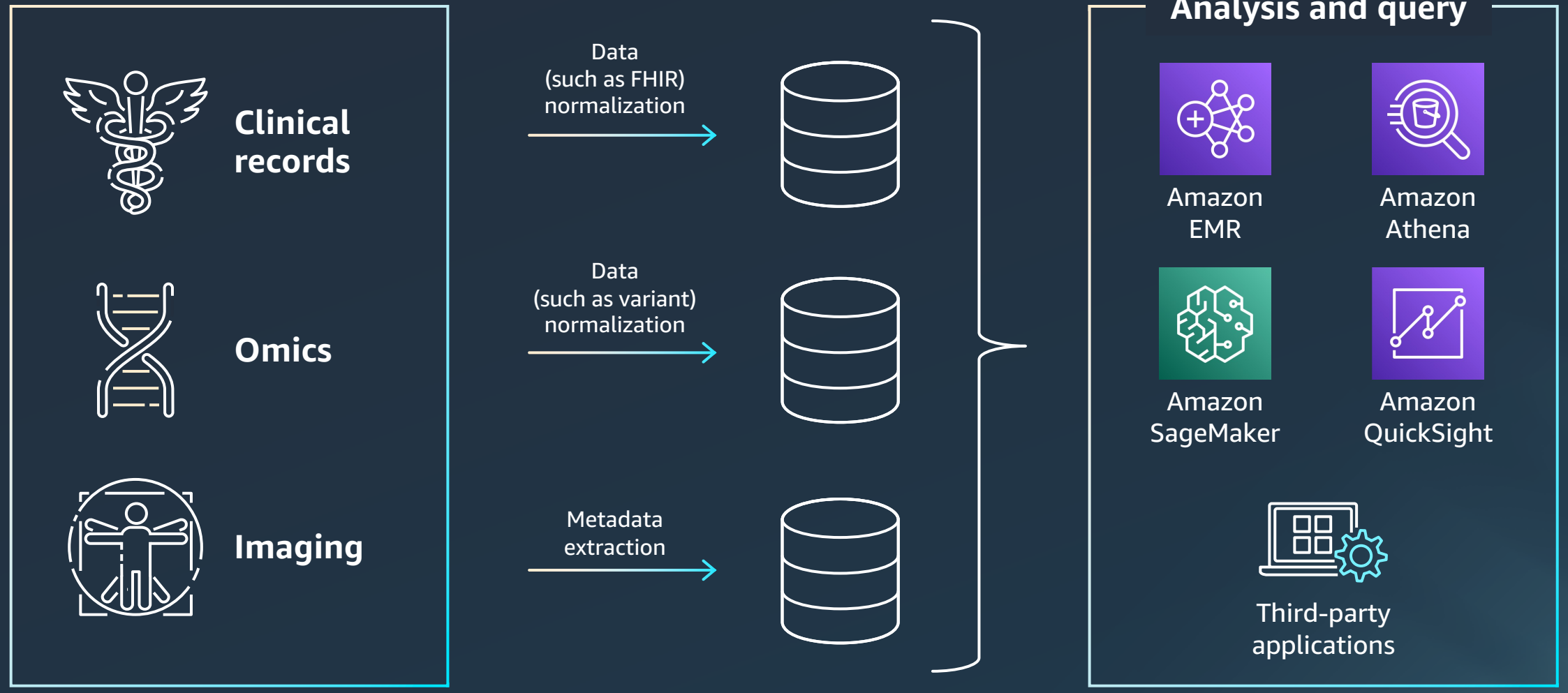
Multiomics and multimodal analyses



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Customers using Amazon Omics today …

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"We know that getting a comprehensive view of our patients is crucial to delivering the best possible care, based on the most innovative research. Combining multiple clinical modalities is foundational to achieving this. With Amazon Omics, we can expand our understanding of our patients' health, all the way down to their DNA."

Jeff Pennington, Associate VP, Chief Research Informatics Officer

Children's Hospital of Philadelphia

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> **Element Biosciences is opening the world of biology to new possibilities through Element's AVITI system. Amazon Omics provides a simple solution for running workflows in the cloud, and this will enable the scientific community to easily process their sequencing data without the need to set up any infrastructure, allowing them to focus on their research.**

Francisco Gracia, SVP Software and Informatics

Element Biosciences

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"AWS Omics allows researchers to use tools and languages from their own domain and considerably reduces the engineering maintenance effort while taking care of cost and resource allocation considerations, which in turn reduces time to market and NRE costs of new features and algorithmic improvements."

Ury Alon, VP Engineering

C2i Genomics

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How do I start?

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Reach out to us!

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Amazon Omics partners

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Get started with Amazon Omics

Developer guide

Webpage

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Additional resources to learn more

Genomics on AWS
aws.amazon.com/health/genomics

AWS for Health
aws.amazon.com/health

Open Data on AWS
aws.amazon.com/opendata

AWS Marketplace
aws.amazon.com/marketplace

AWS Partner Network
aws.amazon.com/partners/find

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Thank you!

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How did we do?

https://survey.immersionday.com/s_eYiF04R

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