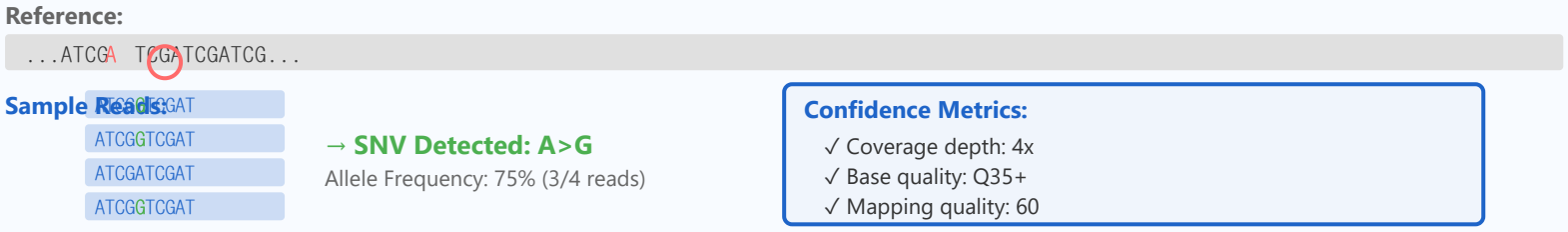
Variant Calling



Variant Calling Process

- Identify differences between sample and reference genome
- Distinguish true variants from sequencing errors
- Calculate confidence scores for each variant
- Filter low-quality and false positive calls

GATK

Gold Standard

Genome Analysis Toolkit

FreeBayes

Bayesian

Haplotype-based

DeepVariant

Deep Learning

Google AI method

Variant Types

SNVs/SNPs

Single nucleotide variants - most common (~50M per genome)

Indels

Small insertions/deletions - 1-50 bp

Structural Variants

Large deletions, duplications, inversions, translocations (>50 bp)

Copy Number Variants

Changes in gene copy number