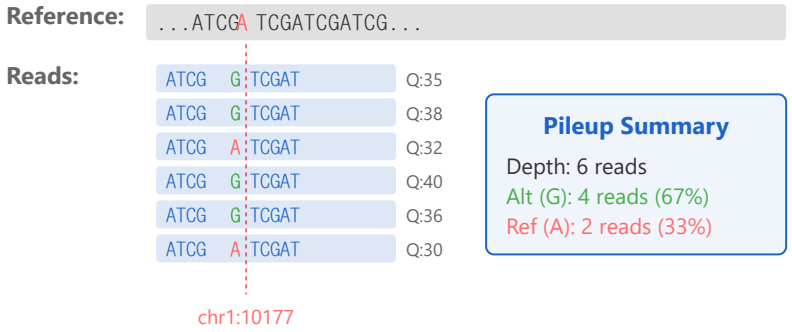
Variant Calling

Variant Calling Process & Algorithm

Step 1: Aligned Reads at Variant Position



Pileup Summary

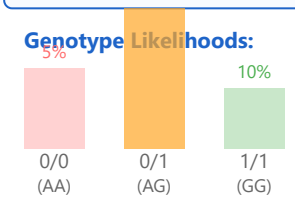
Depth: 6 reads
Alt (G): 4 reads (67%)
Ref (A): 2 reads (33%)

Step 2: Statistical Analysis

Bayesian Genotype Likelihood

$$P(G|D) = P(D|G) \times P(G) / P(D)$$

G=genotype, D=observed data



Quality Metrics

QUAL: 85
GQ (Genotype Quality): 42
DP (Depth): 6

Step 3: Variant Calling Decision Pipeline

Common Filtering Criteria

Quality Score	Read Depth	Allele Balance	Strand Bias
QUAL ≥ 30	**DP ≥ 10**	**0.25 ≤ AF ≤ 0.75**	**FS < 60**

GATK

FreeBayes

DeepVariant

Gold Standard

Genome Analysis Toolkit

Bayesian

Haplotype-based

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