Annotation Tools

Variant Annotation Purpose

- Predict functional effect of variants
- Add gene names and transcript information
- Include population frequency data
- Clinical significance and disease associations
- Conservation scores and pathogenicity predictions

VEP	ANNOVAR	SnpEff
Ensembl	**Comprehensive**	**Fast**
Variant Effect Predictor	Multiple databases	Genomic annotations

Annotation Databases

Population Databases

- gnomAD (global frequencies)
- 1000 Genomes
- ExAC, dbSNP

Clinical Databases

- ClinVar (pathogenicity)
- OMIM (disease-gene)
- COSMIC (cancer)

Prediction Tools

- SIFT (deleteriousness)
- PolyPhen-2
- CADD scores

Conservation

- PhyloP
- GERP++
- PhastCons