CATR
Centre for Advanced Training and Research

Online Workshop On
Clinical Exome Sequencing, Variant Calling, ACMG Interpretation & Pharmacogenomics

EmpowerYour Journeyin ClinicalGenomicsandPrecisionMedicine
Learn • Analyze • Interpret • Innovate

Date & Time: 03-08 July 2026 | 7:15 PM IST

About the Workshop

Step into theworld of clinical genomics and precision medicine with this comprehensive 5-day online workshop focused on Clinical Exome Sequencing (WES), Variant Calling, ACMG Interpretation, and Pharmacogenomics.

Participants will learn the complete workflow beginning from sequencing fundamentals and FASTQ data handling to alignment, variant calling, annotation, ACMG interpretation, and pharmacogenomic analysis.

Workshop Highlights

- Introduction toNext-Generation Sequencing (NGS)
- Whole Exome Sequencing (WES) workflow
- Linux and command-line basics
- Alignment using BWA-MEM
- GATK Best Practices workflow
- Variant annotation using ANNOVAR
- ACMG guideline-based interpretation
- Pharmacogenomics and precision medicine
- Hands-onpractical sessions

Target Audience

Biotechnology andLife science field students | Researchers | Clinicians Bioinformatics Beginners| Healthcare Professional | Anyone interested in Genomics | Precision Medicine.

REGISTER NOW

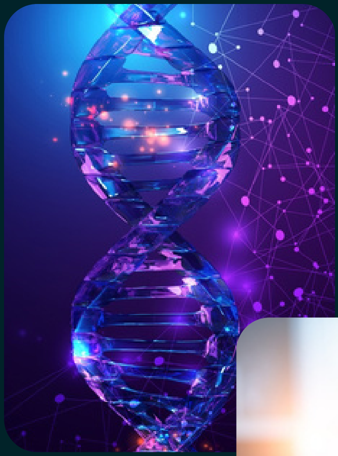
director@catredu.com www.catredu.com +91 8887565470

Course Module

Day 1— Foundationsof NGS and Sequencing Technologies

Theme: How sequencing data is generated

Session 1 : Introduction to Genomics



- DNA, genes, chromosomes
- Exons vs introns
- Central dogma
- Genetic variants
- SNP
- SNV
- Indels
- Structural variants
- Germline vsSomaticmutations

Session 2 : Next Generation Sequencing

- Sanger vsNGS
- Whole Genome Sequencing (WGS)
- Whole Exome Sequencing (WES)
- RNA-seq
- Single-cell sequencing
- Illumina Sequencing
- Flow cell
- Cluster generation
- Sequencing by synthesis (SBS)
- Fluorescent chemistry
- Paired-end sequencing
- Read depth and coverage

Session 3: Library Preparation

- DNA fragmentation
- Adapter ligation
- PCR amplification
- Capture probes
- Exome enrichment

Session 4 : FASTQ Files & Quality Scores

- FASTQ structure
- Read headers
- Phred scores
- Sequencing quality

Practical Session

- Linux Basics
- Viewing FASTQ Files
- Demonstration
- Sequence lines
- Quality lines Paired-
- end reads

Day 2 — Linux, Alignment and BAM Processing

Theme: How reads align to the reference genome

Session 1 : Linux for Bioinformatics
Session 2 : Conda and Environment Management
Session 3 : Alignment Concepts
Session 4 : BWA MEM and SAM/BAM Files

CIGAR Strings

Session 5 : Duplicate Reads and QC

- PCR duplicates
- Optical duplicates
- Why duplicate marking matters

Practical Session

- Alignment Pipeline
- Duplicate Marking
- BAM Inspection

Day 3 — GATK Best Practices and Variant Calling

Session 1 : GATK Best Practices Workflow

Pipeline overview:
FASTQ → BAM → BQSR → Variant Calling → Filtering

Session 2 : Base Quality Score Recalibration
(BQSR)

Session 3 : Germline vs Somatic Variant Calling

HaplotypeCaller
Mutect2

Session 4 : GATK Filtering Concepts

- Panel of Normals(PoN)
- Germline resource
- Strand bias
- Contamination
- False positives

Tool

- FilterMutectCalls

Session 5 : VCF Files and Normalization

- VCF structure
- PASS filtering
- INFO fields
- FORMAT fields
- Multiallelic variants

bcftools normalization
bcftools norm

Practical Session

- Base Recalibration
- Filter PASS variants
- Normalize VCF

Day 4 — Variant Annotation and ACMG Interpretation

Theme: How variants gain biological and clinical meaning

Session1 : VariantAnnotationConcepts

What annotation adds:

- gene names
- functional effects
- frequencies
- clinical significance

Session 2 : Annotation Databases

Databases

- RefGene
- dbSNP
- gnomAD
- ClinVar
- COSMIC
- InterVar

Session 3 : ANNOVAR Workflow

Tools

convert2annovar.pl

table_annovar.pl

Session 4 : ACMG Guidelines

ACMG Classes

- Pathogenic
- Likely pathogenic
- VUS
- Likely benign
- Benign

Evidence Categories

- PVS1
- PM
- PP
- BS
- BP

ClinVar vs ACMG

- Curated evidence
- Conflicting interpretations

Practical Session

Load ANNOVAR output in R

Generate plots

Interpret:

- Pathogenic variants
- VUS burden
- ClinVar conflicts

Session 5 : Somatic Interpretation

Topics

- Driver mutations
- Passenger mutations
- Actionable variants
- COSMIC database

Day 5 & 6 — Clinical Interpretation, Pharmacogenomics and Reporting

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Theme: From variants to precision medicine

Session1 : ClinicalInterpretationofVariants

Topics

- Actionable mutations
- Clinical significance
- Tumor suppressors
- Oncogenes
- Diagnostic relevance

Session2:CancerandClinical Databases

Topics

- COSMIC
- ClinVar
- gnomAD
- dbSNP Interpretation
- Recurrent mutations
- Rare variants
- Cancer relevance

Session 3 : Introduction to Pharmacogenomics Topics

- Drug metabolism
- CYP450 enzymes
- TPMT
- NUDT15
- UGT1A1
- Clinical Importance
- Adverse drug reactions
- Toxicity prediction
- Personalized medicine

Session 4 — Stargazer and Star Alleles

Topics

- Star allele nomenclature
- Diplotypes
- Metabolizer phenotypes

Session 5 — Clinical Reporting and Future Directions

- What goes into a clinical report
- Reporting limitations Validation requirements
- AI in genomics
- Precision medicine

Practical Session

Running StargazerLearning Outcomes

Participants will understand NGS workflows, variant calling, ACMG interpretation, pharmacogenomics, and clinical genomics reporting workflows.

Registration Fee

Indian Participants: **₹1,800**
Previous CATR Participants: **₹1,500**
International Participants: **USD 125**
Previous International Participants: **USD 100**

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