Germline Variant Calling, Annotation & Interpretation — Hands-on

Learn how to move from raw NGS data to clinically interpretable germline variants in rare disease and inherited disorders. This module focuses on validated germline pipelines, VCF QC, annotation strategy and evidence driven interpretation workflows that align with ACMG thinking and clinical reporting needs.

Germline Variant Calling, Annotation & Interpretation

Help Desk · WhatsApp

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Session 1

Fee: Rs 8800 Apply Now

Germline Pipelines & Input Structures

Germline use cases and study types

rare disease **carrier screening** **hereditary cancer**

Pipelines from BAM to VCF for germline calling

single sample **trio and family** **multi sample joint calling mindset**

VCF structure and key fields for interpretation

headers and metadata **INFO and FORMAT tags** **genotypes and phasing basics**

Session 2
Fee: Rs 11800 Apply Now

VCF QC, Filtering & Triage

QC checks at variant level

depth and allele balance **quality metrics** **callable regions**

Filtering strategies and prioritisation tiers

population frequency cutoffs **impact based filters** **mode of inheritance filters**

Shortlisting variants for manual review

candidate gene lists **phenotype driven filters** **export to spreadsheet and curation tools**

Session 3
Fee: Rs 14800 Apply Now

Annotation, Databases & ACMG Logic

Functional and clinical annotation layers

gene and transcript impact **in silico predictions** **constraint and intolerance scores**

Key databases for germline interpretation

gnomAD and population frequency **ClinVar submissions** **HGMD and locus specific resources**

ACMG style reasoning for germline variants

evidence codes overview **combining criteria** **P LP**

VUS LB B buckets mindset

Session 4

Fee: Rs 18800 Apply Now

Mini Capstone: Variant Curation & Report Draft

Curate a small set of candidate variants for a case

Theory + Practical

Assign provisional classifications with evidence table

evidence codes mapping | rationale notes | audit friendly layout

Draft a variant summary for clinical reports

HGVS representation | phenotype linkage | suggested follow up testing