



Germline Diagnostics & Carrier Screening Pipelines — Hands-on

Build auditable, production-grade pipelines for germline diagnostics and carrier screening. From raw reads to interpretive reports, learn trio/singleton analysis, inheritance-based prioritization, CNV/SV detection in germline contexts, residual risk calculations, and ACMG/AMP classification with clear, clinician-ready outputs.

Germline Diagnostics & Carrier Screening Pipelines

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[Session 1](#)

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Germline Pipeline Foundations

Pre-processing & calling (germline)

[BWA MEM2 / GATK](#) [BQSR / dedup](#) [HaplotypeCaller \(GVCF\)](#)

Quality metrics & coverage

[%20x/%30x targets](#) [callability/gaps](#) [hard-to-sequence](#)

loci

Annotation & gene panels

VEP/snpEff **clinically curated lists** **transcripts/MANE**

Session 2

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Inheritance Models & Prioritization

Trio & singleton strategies

de novo / AR / AD / X-linked **compound het**
segregation

Phenotype-driven ranking

HPO mapping **OMIM/Orphanet** **gene-disease validity**

Population & artifact filters

gnomAD/MAF thresholds **low-complexity regions**
technical flags

Session 3

Fee: Rs 25520 Apply Now

CNV/SV in Germline & Carrier Screening

CNV calling (exon-level)

ExomeDepth/CNVkit **reference panels** **QC &**
thresholds

Germline SV detection

Manta/GRIDSS (overview) **breakpoint checks**
orthogonal MLPA/ddPCR

Carrier screening workflows

panel design & genes **detection rate & coverage**
partner testing

Session 4

Fee: Rs 31120 Apply Now

Classification, Residual Risk & Reporting

ACMG/AMP classification (germline)

PVS1/PS/PM/PP **BS/BP** **Bayesian adjustments**

Residual risk & counseling notes

detection rate × prevalence **ethnicity considerations**

VUS handling

Reports, reanalysis & QA

templates & disclaimers **reclassification cadence**

audit trails & SOPs