Rare Disease Genomics & Variant Prioritization — Hands-on

Diagnose rare diseases with a rigorous, phenotype-driven workflow. This module takes you from deep clinical phenotyping and pedigree modeling through trio-based WES/WGS analysis, multi-class variant discovery (SNV/INDEL/CNV/SV), prioritization with in-silico scores and population constraint, and final ACMG/AMP interpretation with audit-ready reports and reanalysis strategies.

Rare Disease Genomics & Variant Prioritization

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Session Index **Category 5 · Clinical & Translational Bioinformatics**

Session 1 — Phenotyping, Pedigrees & Inheritance | Session 2 — Trio WES/WGS: QC, Calling & Filtering | Session 3 — Interpretation & Prioritization | Session 4 — Reporting, Reanalysis & Data Sharing

Session 1

Fee: Rs 24920 Apply Now

Phenotyping, Pedigrees & Inheritance

Deep phenotyping

HPO terming & granularity **OMIM/Orphanet crosswalks** **phenotype-driven ranking (overview)**

Pedigrees & models

AR/AD/X-linked/mitochondrial | compound het & de novo | segregation checklists

Sample/metadata readiness

trio/quad strategies | consent & recontact | phenopackets (overview)

Session 2

Fee: Rs 29120 Apply Now

Trio WES/WGS: QC, Calling & Filtering

Quality control & coverage

depth/uniformity/dup rates | target capture pitfalls | sex/relatedness checks

Variant classes

SNV/INDEL (joint calling) | CNV/SV (exome & genome) | mitochondrial & repeat expansions (overview)

Filtering & inheritance models

de novo/compound het/recessive | population AF <= thresholds (gnomAD) | artifact & blacklist pruning

Session 3

Fee: Rs 33320 Apply Now

Interpretation & Prioritization

Evidence assembly

ClinVar/ClinGen/OMIM | gene–disease validity | constraint & intolerance

In-silico & splicing

CADD/REVEL/M-CAP | SpliceAI/MaxEnt (overview) | conservation/PP3 aggregation

ACMG/AMP for rare disease

criteria application & conflicts | segregation/PS2/PP1 strength | VUS management & upgrades

Session 4

Fee: Rs 38920 Apply Now

Reporting, Reanalysis & Data Sharing

Clinical reporting & counseling support

clear narratives & caveats | secondary/incidental findings (overview) | confirmatory testing guidance

Reanalysis & matchmaking

knowledge updates & SLAs | GeneMatcher/Matchmaker (overview) | phenotype drift handling

Data sharing & registries

ClinVar/DECIPHER submissions | consent & de-ID | FAIR principles