Family-Based Genetics — Trio Analysis & Mendelian — Hands-on

Understand how family based genetics and trio analysis strengthen rare disease and Mendelian diagnostics. This module focuses on pedigrees, inheritance patterns, trio logic and segregation style thinking so that participants can read and design basic family based analyses and summaries. Content is for training and awareness, not clinical, diagnostic or genetic counselling advice.

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Session Index

Session 1

Fee: Rs 8800 Apply Now

Family-Based Genetics Foundations

Why family based designs matter in rare disease genomics

increasing confidence in causal variants | **reducing candidate list burden** | **informing recurrence risk mindset**

Typical family structures in genomic testing (high level)

trio, quad and extended families | **affected and**

unaffected relatives snapshot | sample selection and priorities mindset

Mendelian language and categories (conceptual)

autosomal dominant and recessive idea | X linked and mitochondrial snapshot | de novo and compound heterozygous concepts

Session 2

Fee: Rs 11800 Apply Now

Pedigrees, Segregation & Inheritance Patterns

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Building and reading basic pedigrees

pedigree symbols and conventions snapshot | three generation family history mindset | affected status and key annotations

Recognising Mendelian inheritance patterns (conceptual)

dominant and recessive style patterns | X linked and mitochondrial hints | incomplete penetrance snapshot

Segregation thinking at a high level

who should carry a causal variant idea | fully, partially and non segregating patterns | sources of apparent mismatches mindset

Session 3

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Trio Analysis, De Novo & Segregation Evidence

Trio analysis logic at a conceptual level

child and parent genotype patterns | transmitted vs non transmitted alleles snapshot | using unaffected parents in filtering idea

De novo, recessive and X linked style variant patterns

de novo candidates and basic checks **homozygous and compound heterozygous idea** **simple X linked patterns snapshot**

Segregation style evidence in a family context (high level)

checking variant presence in additional relatives **supporting, conflicting and missing information** **link to variant interpretation frameworks (conceptual)**

Session 4

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Mini Capstone: Family Case Workup Snapshot

Start from a small anonymised family case description

Theory + Practical

Sketch inheritance model and trio style expectations

possible Mendelian patterns **who should carry a candidate variant** **simple segregation table snapshot**

Prepare a short genetics friendly summary paragraph

candidate pattern and rationale **data gaps and uncertainties** **notes for further clinical or laboratory follow up**