Structural Variants, Missense Impact & Disease Mechanisms — Hands-on

Learn how to move from a list of variants to structural and mechanistic hypotheses. This module focuses on mapping missense changes onto protein structures, reasoning about local and global impact, and building interpretable disease mechanism narratives that complement genomic annotations.

Structural Variants, Missense Impact & Disease Mechanisms

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Session Index

Session 1

Fee: Rs 28800 Apply Now

Variant Types, Annotation & Structural Mapping

Variant classes and basic annotation concepts

missense vs truncating vs inframe **germline vs somatic context** **population and clinical databases ideas**

From sequence-level variants to protein coordinates

transcript and isoform awareness **mapping onto**

UniProt sequences | aligning to PDB/AlphaFold models

Structural context categories for variants

core vs surface vs interface residues | motifs, domains and active sites | disordered and unresolved regions

Session 2

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Local Structural & Biophysical Impact

Residue environment and local packing

hydrophobic cores and cavities | H-bonds, salt bridges and networks | secondary structure and motif context

Biophysical impact reasoning for substitutions

size, charge and polarity changes | stability gain vs destabilization ideas | effects on dynamics and flexibility concepts

Interfaces, active sites and regulatory regions

protein–protein and protein–ligand contacts | catalytic residues and binding pockets | allosteric and PTM-associated positions

Session 3

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Networks, Pathways & Mechanistic Hypotheses

Structural networks and distant effects

interaction networks inside proteins | possible allosteric communication routes | clustered vs dispersed variant patterns

Linking structural impact to function and pathways

enzyme kinetics and binding changes concepts **complex assembly and stability** **mapping onto signalling and metabolic pathways**

Integrating structural evidence with variant scores

tolerance landscapes and constraint ideas **using generic impact predictors alongside structure** **consistency with population and disease data**

Session 4

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Variant Interpretation Report & Case Studies

Case studies across disease areas

enzymopathies and metabolic defects **cancer-associated missense clusters** **channelopathies and receptor variants**

Structuring a variant interpretation report

variant tables with structural fields **figures for local environments and clusters** **mechanism and risk summarization**

Deliverables: structural variant dossier & follow up ideas

ranked variants with structural rationale **hypotheses for experiments or screening** **project-ready summary for teams and collaborators**