CNV, SV & Structural Variants in Clinical Context — Hands-on

Gain a working understanding of copy number variants (CNVs), structural variants (SVs) and complex rearrangements in clinical genomics. This module connects wet lab assays to bioinformatics pipelines, QC and interpretation workflows, showing how CNV and SV findings are integrated into real clinical reports for rare disease and oncology cases.

CNV, SV & Structural Variants in Clinical Context

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Session Index

Session 1

Fee: Rs 8800 Apply Now

CNV/SV Biology & Assay Landscape

Biology of CNVs, SVs and structural variation

deletions and duplications **inversions and translocations** **complex rearrangements overview**

Assay options for CNV/SV detection

karyotype and FISH snapshot **microarray and MLPA** **NGS based approaches**

Clinical scenarios where CNVs/SVs matter

developmental delay and syndromes **hereditary cancer genes** **oncology fusions and rearrangements**

Session 2
Fee: Rs 11800 Apply Now

CNV/SV Pipelines & QC from NGS

CNV/SV calling from exome and genome data

read depth and coverage based methods **split reads and discordant pairs** **panel vs WES vs WGS nuances**

QC metrics for structural variant pipelines

probe or exon level coverage **noise and false positives** **confirmatory assay triggers**

File formats and visualization

CNV tables and BED like outputs **IGV snapshots coverage plots for review**

Session 3
Fee: Rs 14800 Apply Now

Interpretation, Classification & Reporting

Annotation sources for structural variants

ClinGen dosage sensitivity **DECIPHER and DGV snapshot** **curated gene panels**

Guideline frameworks for CNV/SV classification

ACMG/ClinGen CNV guidelines **evidence categories overview** **LP P VUS LB B buckets**

Reporting structural variants in context

HGVS for CNVs and fusions (high level) **phenotype**

linkage and penetrance notes follow up and family studies

Session 4

Fee: Rs 18800 Apply Now

Mini Capstone: CNV/SV Case Workup

Work through a structural variant case

Theory + Practical

From pipeline output to curated CNV/SV table

coordinates and genes affected classification and evidence validation status and comments

Draft a report friendly structural variant summary

narrative summary risk communication points recommendations for clinicians