

ClinVar, COSMIC, gnomAD & Knowledgebase Integration — Hands-on

Learn how to use ClinVar, COSMIC, gnomAD and related knowledgebases effectively in clinical genomics. This module focuses on efficient variant lookup, evidence aggregation, concordance checks and how to feed curated knowledge into ACMG/AMP style interpretation, oncology actionability and report-ready summaries.

ClinVar, COSMIC, gnomAD & Knowledgebase Integration

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Session 1

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Clinical Variant Databases & Roles

Landscape of clinical and research variant resources

[ClinVar](#) [COSMIC](#) [gnomAD & related population sets](#)

What each database is designed for (high level)

[clinical submissions and assertions](#) [somatic and tumour focussed resources](#) [population frequency baselines](#)

How variant databases support ACMG/AMP & oncology workflows

evidence lookups for criteria **actionability hints**
(high level) **data provenance mindset**

Session 2

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Working with ClinVar, COSMIC & gnomAD

Efficient variant lookup patterns in ClinVar

accessions vs coordinates **clinical significance**
fields **review status snapshot**

COSMIC for somatic and tumour context

tumour types and histology tags **recurrent vs rare**
events snapshot **linking to actionability frameworks**
(high level)

gnomAD for population frequency context

per population allele frequencies **coverage and**
filters snapshot **rare vs common signal mindset**

Session 3

Fee: Rs 14800 Apply Now

Knowledgebase Linking & Harmonisation

Pulling evidence together across databases

aligning variant identifiers **cross checking**
assertions **flagging discordant entries**

Connecting to clinical knowledgebases (high level)

disease focussed resources snapshot **oncology**
actionability resources snapshot **linking to**
guidelines and trials (conceptual)

From raw database hits to interpretable evidence blocks

summarising key fields **noting conflicts and caveats**
readiness for ACMG/AMP tables

Session 4

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Mini Capstone: Knowledgebase-Driven Case Review

Work through a small variant list using databases

Theory + Practical

Build a variant evidence summary table from
ClinVar/COSMIC/gnomAD

database hits and key fields **concordance and**
conflict notes **ready for ACMG/AMP use**

Draft a short narrative on database-driven evidence

plain language explanation **limitations and data gaps**
future re review triggers