

Curation tool

Agenda

The problem

The solution

Future development

The problem

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Transcription errors

Total AF = 0.004337 %

CF 18-Mar-2026: Total AF = 0.1328%

Lack of guidelines

Search:

⚡ Disease

No matching records found

A million tabs



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CATTAACCAAAATTTCTCTCTCCATTCTCCACAAACTTTTCTTAATATTCACAGCTTCTTCTT

The solution

Just have an excel sheet to store and calculate

Pros: Easy

Cons: Too many excel sheets

So I thought, why not combine them into an app

ACMG Gene Curation

CURATOR + GENE RULES + LIBRARY

Curator Gene Rules Curations

Curations Refresh

Complete variant entry to begin curation

Variant Entry

WORKFLOW

- Variant Entry

EVIDENCE

No codes yet

Patient Details

LAB ID REQUIRED

PERSONAL HISTORY

FAMILY HISTORY

Gene & Nomenclature

GENE

MANE TRANSCRIPT

C. NOMENCLATURE

P. NOMENCLATURE

 Lookup

VARIANT TYPE

Missense Nonsense Frameshift Deletion Insertion Indel Synonymous Intronic Splice Site

Begin Curation →

Capabilities: Classifier

Retrieval

- Nomenclature
- GnomAD entry
- ClinVar entry with submission classification and notes
- Nearby variants in ClinVar
- In-silico predictor scores
- Links to literature searches

Capabilities: Classifier

Automated Classification

- Population data classification based on GnomAD frequencies
- In-silico classification based on variant type, location, and scores
- Automated scoring of de-novo, proband counting codes according to ClinGen SVI criteria

Real time addition of codes according to the Bayesian point scoring system.

*All classifications have a manual override

Capabilities: Gene Information

Gene information storage

- Exon nucleotide and protein boundaries
- Functional domain boundaries- literature specific
- Gene-specific code modification
- Literature information

The classifier updates in real time to the information stored and updated here

Capabilities: Curation record

Contains the curation information as a saved record according to the gene name and location

- Stores editable curations with the date and timestamp
- Allows for report generation as a shareable pdf

1. Additional variant types added: Splice-site, Deletions, Duplications
2. VCEP gene integration
3. MAVE study integration
4. Report writing assistance

I would love to get into touch regarding creating a personalised version of this tool for your curation team!

Please email me at ladmudra@gmail.com for a live demonstration :))

Thank you !