Curation tool

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Agenda

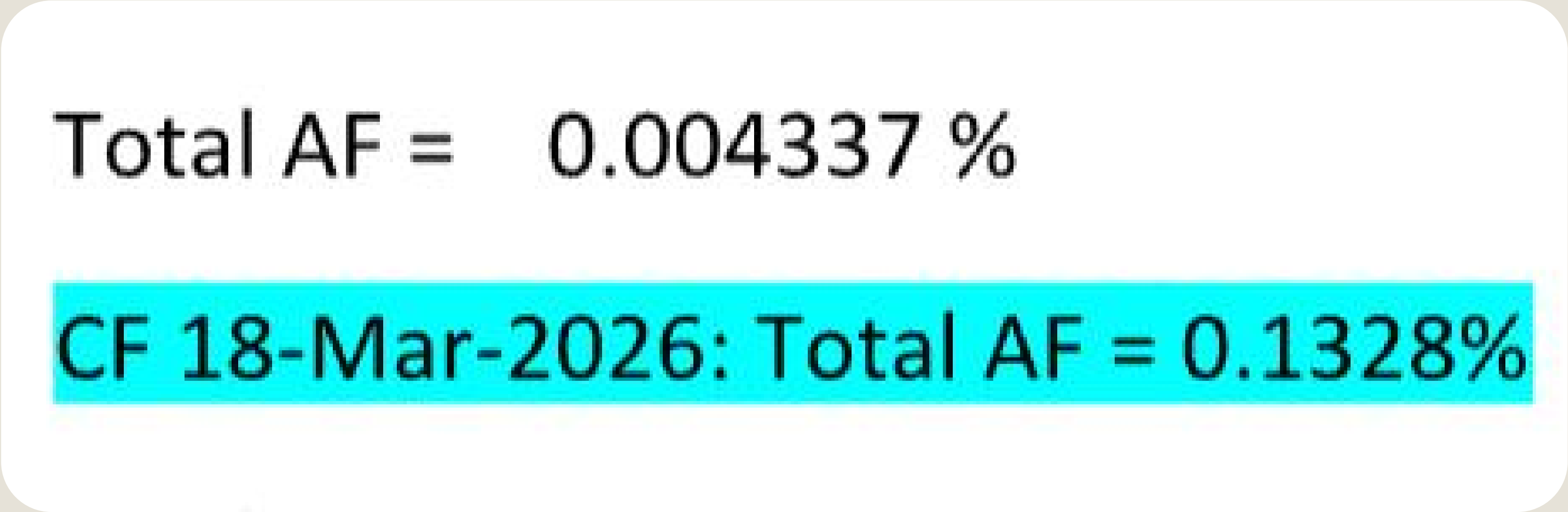
The problem

The solution

Future development

The problem

Transcription errors



Total AF = 0.004337 %

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

Lack of guidelines

Search: smad4

No matching records found

A million tabs

The solution

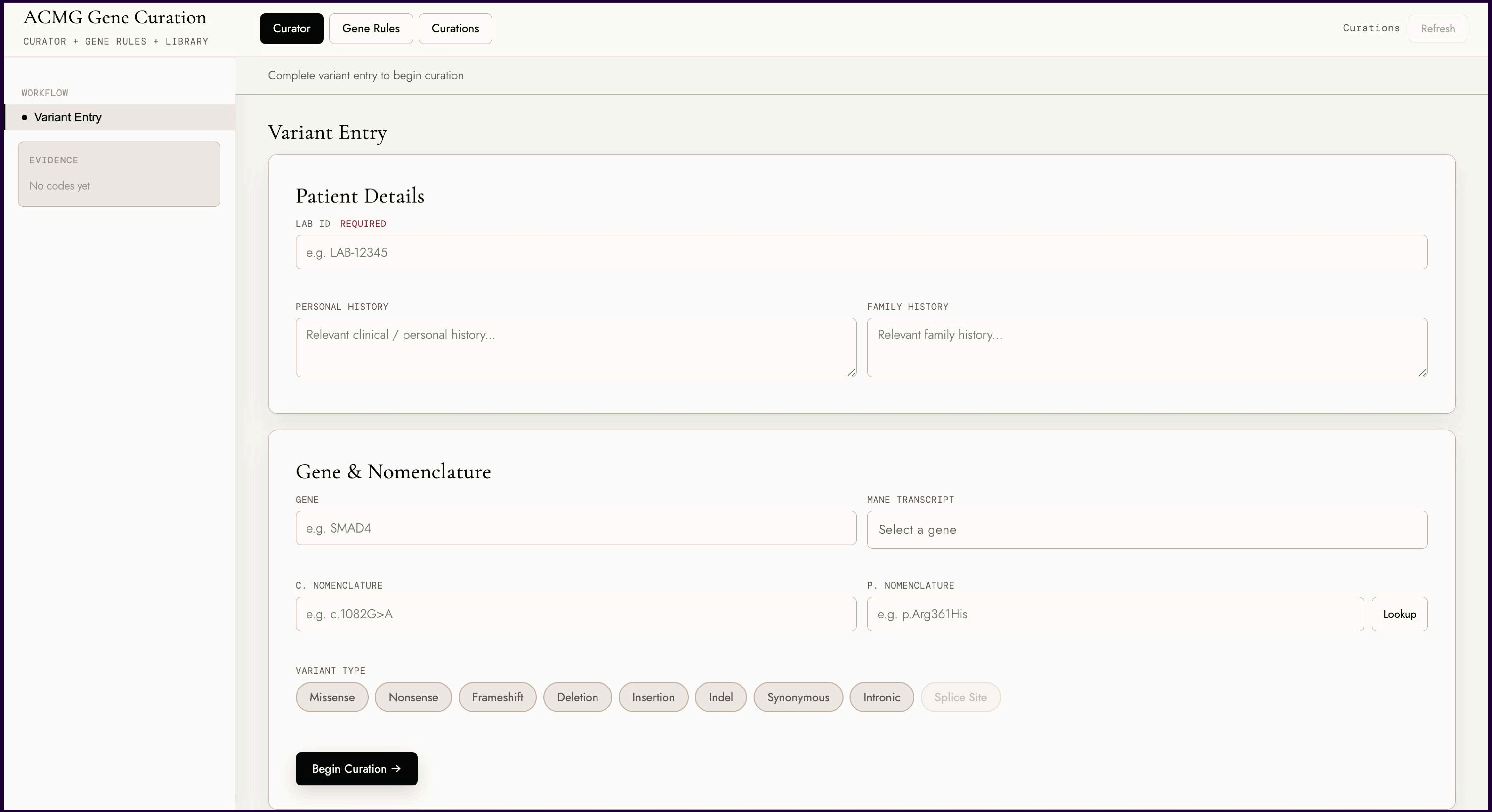
Just have an excel sheet to store and calculate

Pros: Easy

Cons: Too many excel sheets

So I thought, why not combine them into an app

CURATION TOOL



ACMG Gene Curation

CURATOR + GENE RULES + LIBRARY

WORKFLOW

- Variant Entry

EVIDENCE

No codes yet

Complete variant entry to begin curation

Variant Entry

Patient Details

LAB ID REQUIRED

e.g. LAB-12345

PERSONAL HISTORY

Relevant clinical / personal history...

FAMILY HISTORY

Relevant family history...

Gene & Nomenclature

GENE

e.g. SMAD4

MANE TRANSCRIPT

Select a gene

C. NOMENCLATURE

e.g. c.1082G>A

P. NOMENCLATURE

e.g. p.Arg361His

VARIANT TYPE

Missense Nonsense Frameshift Deletion Insertion Indel Synonymous Intronic Splice Site

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 classifcation 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

Future directions

1. Additonal 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 !