

Who submits to ClinVar?

- 480 Labs submitting results from clinical testing
- 494 Labs submitting results from research
- 85 Clinics
- 15 Consortia
- 12 Resources (including OMIM™, GeneReviews™, UniProt)
- 31 Locus-specific databases (LSDB)
- 6 Expert Panels
- 1 Practice Guidelines/Professional Societies

1062 Total Submitters*



More than **702,000** submitted records representing more than **443,000** unique variants from **1070** submitters

Expert Panels in ClinVar

Expert panel	total # of interpretations	# of interpretations of pathogenic/likely pathogenic
InSIGHT	2405	1376
CFTR2	291	291
ENIGMA	7258	4831
PharmGKB	431	none - classified for drug response
ClinGen Inherited Expert Panel	102	39
ClinGen RASopathy Expert Panel	233	58

How are data submitted to ClinVar used?

Data submitted to ClinVar are fully public and available for any use.

To view on the ClinVar web site

ncbi.nlm.nih.gov/clinvar/

To download on the ClinVar ftp site

[ftp.ncbi.nlm.nih.gov/pub/clinvar](ftp://ncbi.nlm.nih.gov/pub/clinvar)

Submitted data are linked to related information in

- ★ the NIH Genetic Testing Registry (GTR)
- ★ MedGen
- ★ dbSNP
- ★ external sites including locus-specific databases (LSDBs)
- ★ dbVar
- ★ PubMed

Interested in submitting to ClinVar?

Where do I start?

1. Go to **ClinVar Submission Portal**
2. Create a MyNCBI account and register your organization
3. Submit your data

ClinVar Submission Portal

ncbi.nlm.nih.gov/clinvar/docs/submission_portal/

ClinVar Submission Templates

ftp://ftp.ncbi.nlm.nih.gov/pub/clinvar/submission_templates/

ClinVar Submission Wizard – for one or a few variants

Lite Submission Template (Excel) – for many sequence variants with minimal data

Full Submission Template (Excel) – for many sequence variants with detailed case data, and for all structural variants.

TSV/CSV formatted files – for non-Excel users, based on tabs and columns in the Excel submission templates

XML – for submitters with detailed data and strong bioinformatics support

I uploaded my data. What happens next?

Your submission will be added to ClinVar's processing queue.

- ★ Wizard submissions are usually assigned for processing within a week
- ★ File submissions require longer for assignment and processing

Once assigned, a curator will review the submission and contact you with questions

You may check the status of your submission at any time in **Submission Portal**.

Want to learn more?

Learn more about submission to **ClinVar**

ncbi.nlm.nih.gov/clinvar/docs/submit/

Get a checklist for faster processing

ncbi.nlm.nih.gov/clinvar/docs/spreadsheet/#checklist

Check our our videos on  **YouTube**

bit.ly/clinvar_videos

