

# The ClinGen and ClinVar Partnership

Both provide resources to support genomic interpretation

## ClinGen - A Program

An NIH funded project

Building a central resource that defines the clinical relevance of genes and variants

ClinGen is addressing the following critical questions:

- Is the gene associated with disease?
- Is the variant pathogenic?
- Is the variant/gene information actionable?

Encouraging **data sharing**

- Promote lab submissions to ClinVar
- Facilitate patient data sharing through GenomeConnect



Assessing the clinical **validity** and **actionability** of genes and their relationship to diseases

Expertly **curating** and **interpreting** variants

- Provide curated knowledge to **ClinVar** and on **clinicalgenome.org**

Partnership to improve knowledge of genomic variation

**Expert Curation**

## ClinVar - A Database

- Funded by intramural NIH funding
- Freely accessible and downloadable public archive of reports of the relationship between variants and conditions
- Maintained by the National Center for Biotechnology Information (NCBI)

Supporting **sharing** of variants interpretations



Maintaining a publicly available **database** of:

- Interpretations of the clinical significance of variants
- Submitter information
- Supporting evidence and individual level data, when available

**ClinGen**

Find out more online...

**ClinVar**

-  <https://www.clinicalgenome.org/>
-  @clingenresource
-  ClinGen Resource
-  ClinGen Youtube Channel

<http://www.ncbi.nlm.nih.gov/clinvar/>

@NCBI\_Clinical

