The Clinical Genome Resource (ClinGen): An Overview

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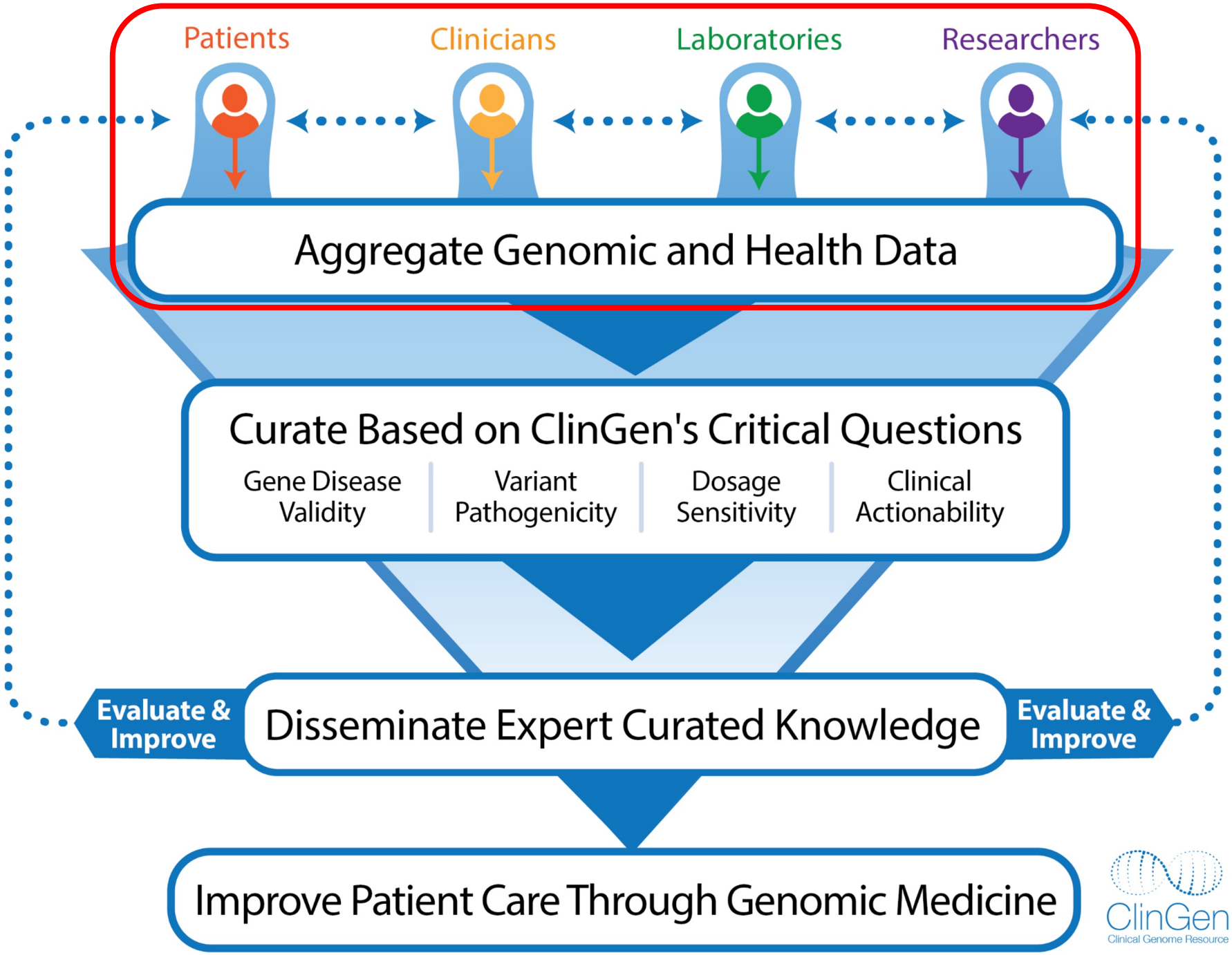
ClinGen
Clinical Genome Resource

The Clinical Genome Resource (ClinGen)

- Initially primarily funded by the National Human Genome Research Institute (NHGRI), part of the National Institutes of Health (NIH) in the United States in 2013
  - Current funding through 2026
- 3 separate grants:
  - Baylor/Stanford (with co-funding from the National Cancer Institute)
  - Broad/Geisinger
  - University of North Carolina at Chapel Hill
- Governed by a Steering Committee including all PIs, additional key representatives from each grant, and representatives from NHGRI and NCBI's ClinVar
- Designated as a Global Core Biodata Resource
- As of April 2026:
  - >45 working groups, >120 expert panels
  - >2700 individuals contribute to ClinGen from 76 different countries

ClinGen's Mission

- Build and support authoritative central resources that define the clinical relevance of genes and variants for use in precision medicine and research


Patients
Clinicians
Laboratories
Researchers
Aggregate Genomic and Health Data
Curate Based on ClinGen's Critical Questions
Gene Disease Validity
Variant Pathogenicity
Dosage Sensitivity
Clinical Actionability
Evaluate & Improve
Disseminate Expert Curated Knowledge
Evaluate & Improve
Improve Patient Care Through Genomic Medicine
ClinGen
Clinical Genome Resource

Aggregating Genomic and Health Data

- Important: ClinGen does NOT generate any new sequencing data
- ClinGen relies on previously generated data from multiple sources to power its curation work, including (but not limited to):
  - Classified variants and associated phenotype information submitted to NCBI's ClinVar by patients, clinicians, laboratories, and/or researchers →important for determining which variants may have been observed in affected individuals
  - Previously generated population data sets (ex: gnomAD) →important for determining which variants have been observed in the general population and at what frequencies
  - Gene and variant annotations from external sources → variant effect predictions, *in silico* predictions, functional data annotations, etc.
  - Published literature →can be a source of all of the above
  - All of this information can either be programmatically or manually annotated in our curation interfaces.

What is the difference between ClinVar and ClinGen?

ClinGen
Clinical Genome Resource

- NIH-funded project
- Encourages data submission to ClinVar
- Uses this information to answer critical curation questions about genes and variants
  - Curations adjudicated by experts following transparent evaluation procedures
- Provides feedback to ClinVar on usability, feature requests, etc.

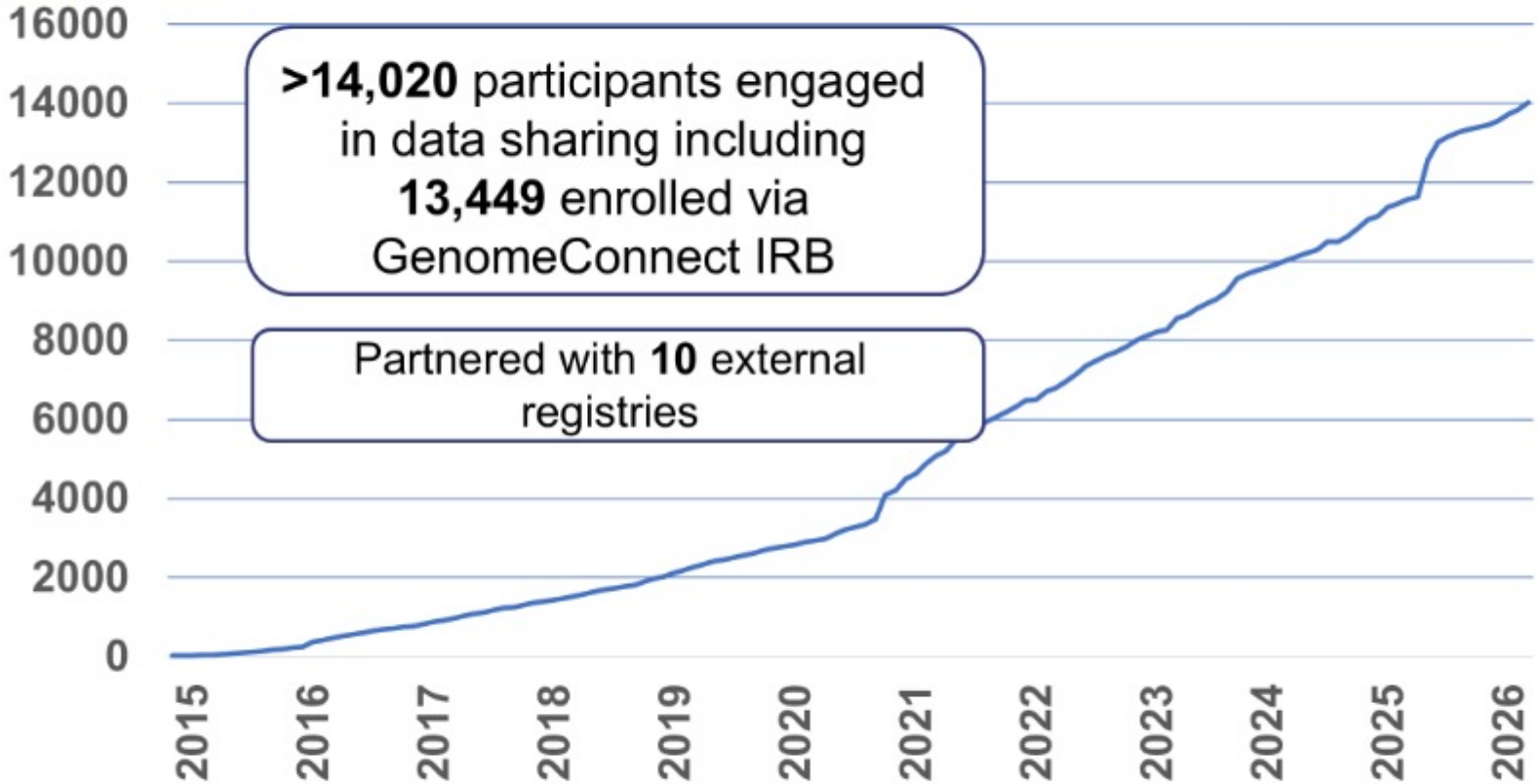
NCBI ClinVar

- Intramural NIH funding
- Created and maintained by the National Center for Biotechnology Information (NCBI)
- Public archive of reports of variants and their relationships to human disease
- Submitter-driven resource
  - Vast majority of submissions accepted (rare exceptions)
  - ClinVar does not classify variants themselves – only reports/aggregates what has been submitted.

A partnership to improve knowledge of genomic variation

GenomeConnect: The ClinGen Patient Registry

GenomeConnect
The ClinGen Patient Portal



>14,020 participants engaged in data sharing including **13,449** enrolled via GenomeConnect IRB

Partnered with **10** external registries

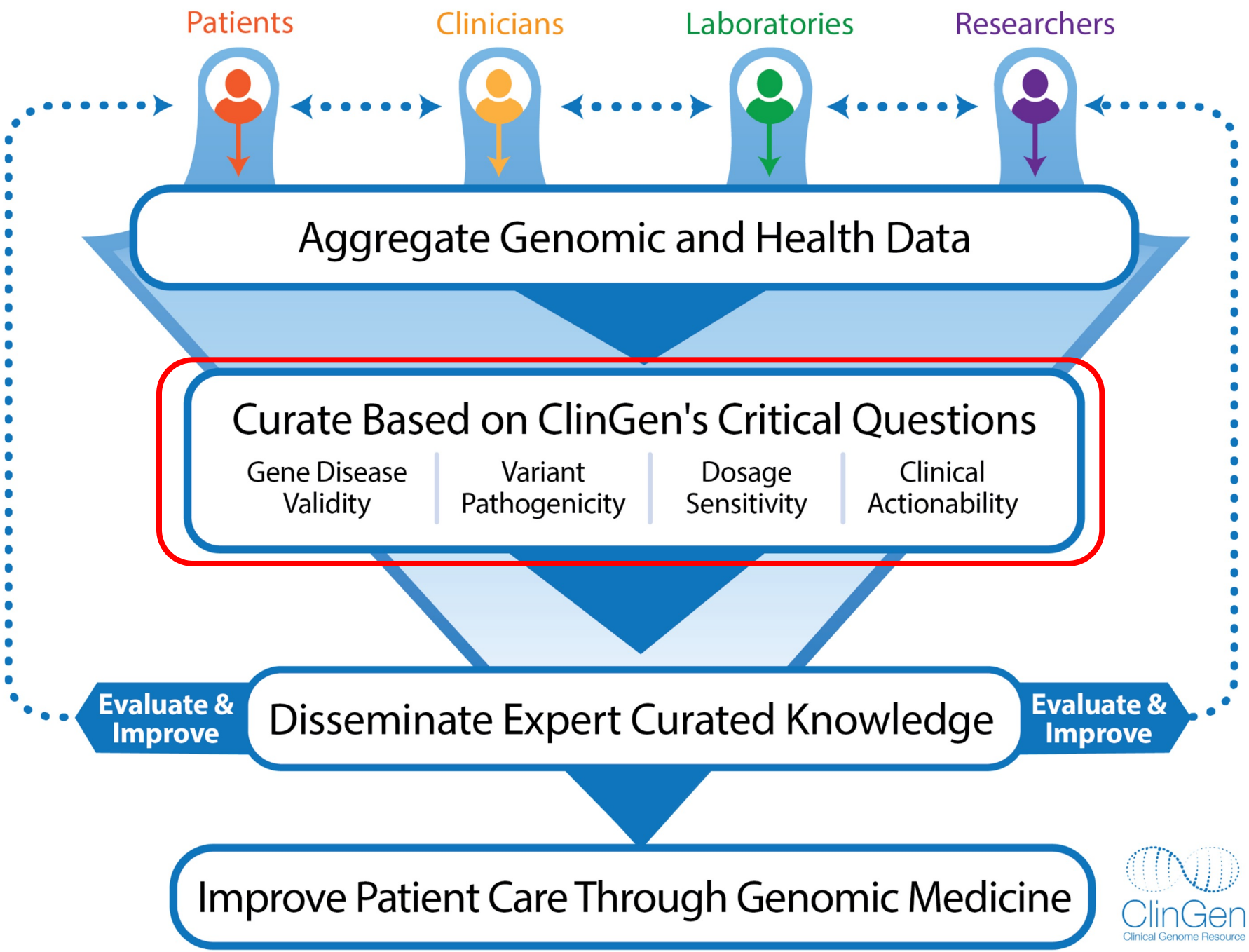
- Participants consent to data sharing and recontact
- Upload genetic testing report(s)
- Provide health history via survey(s)
- Deidentified data shared with ClinVar
- Connect with others; potentially receive genetic result updates

>6,260 variants shared with ClinVar

>470 variant updates returned

Available in **English** and **Spanish**

Data as of March 31, 2026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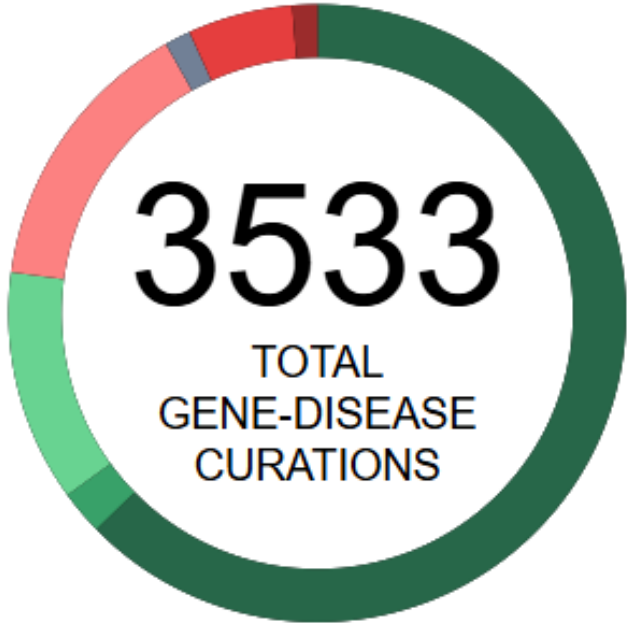
Gene-Disease Validity

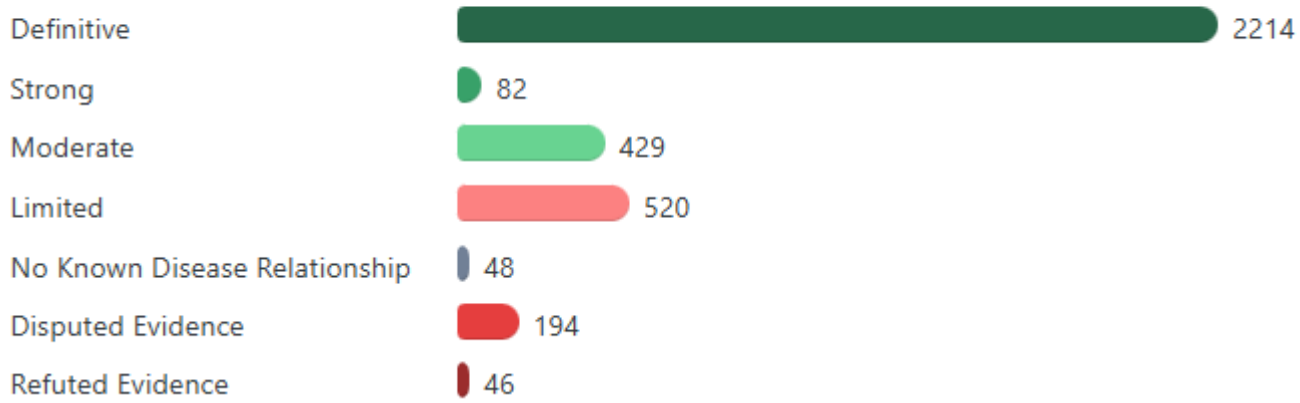
- Does pathogenic variation in a given gene cause disease?
- Semi-quantitative scoring metric describing the strength of evidence supporting or refuting claims of gene-disease relationship[1]
- Information can be used to guide clinical genomic test development or analysis pipelines[2,3]



45 active Gene Curation Expert Panels (GCEPs) over **17** different clinical domains

Classification Statistics

Gene-Disease Clinical Validity has **3533 curations** encompassing **2911 genes**.

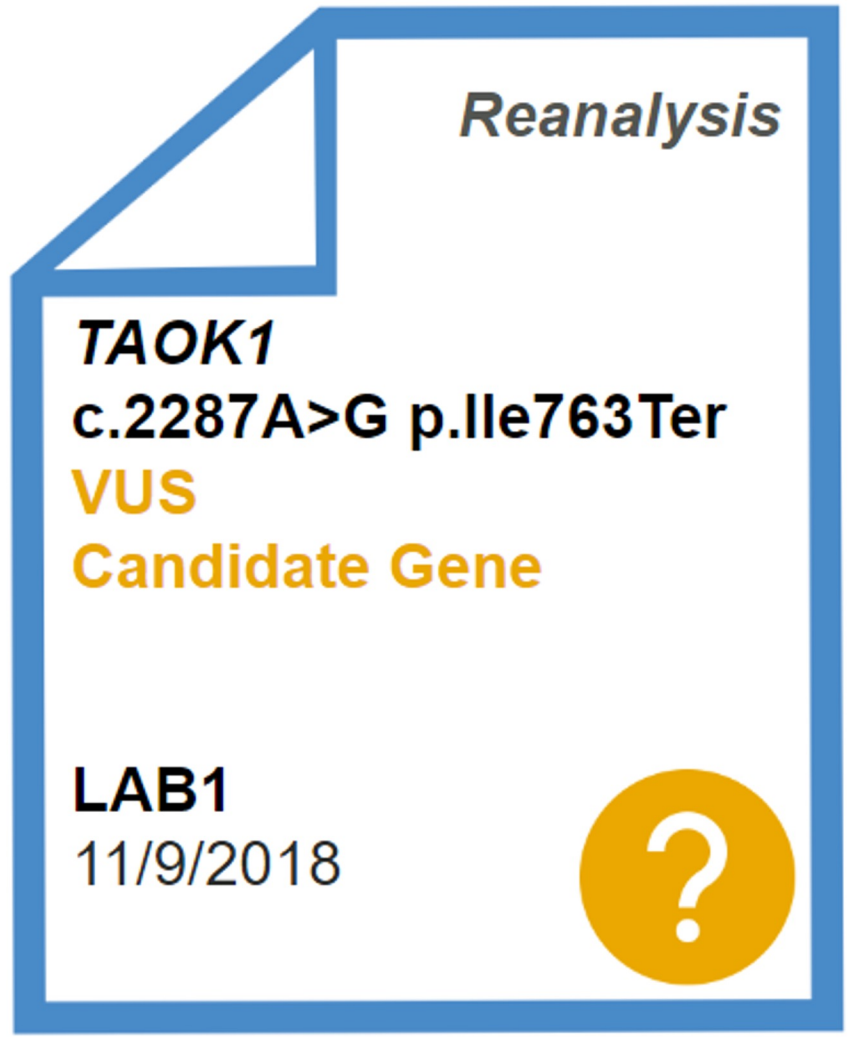


[1]Strande *et al.* 2017; PMID:28552198
[2]Bean *et al.* 2020; PMID:31732716
[3]Thaxton *et al.* 2021; PMID:34694049

As of April 14, 2026

Case Example

- 10-year-old male with autism and intellectual disability and his guardian present to your clinic after relocating to the area
- Previous negative Fragile X testing and microarray
- Exome sequencing identified a VUS in *TAOK1*. According to the report, *TAOK1* was a candidate gene at the time of report.



https://bit.ly/ClinGenGene

Case Example

Gene:	TAOK1 HGNC:29259		
Disease:	syndromic intellectual disability MONDO:0000508		**Definitive** Classification - 08/04/2021
Mode of Inheritance:	Autosomal dominant inheritance HP:0000006	**SOP:**	ClinGen Gene Validity Evaluation Criteria SOP8
Expert Panel:	Intellectual Disability and Autism	**Contributors:**	

Summary

TAOK1 - syndromic intellectual disability

Replication over time:	YES	Contradictory Evidence:	NO
Expert Panel:	Intellectual Disability and Autism		

Evidence Summary:

Although intragenic deletions in TAOK1 had been observed in large cohort studies of individuals with neurodevelopmental disorders in 2011 and 2018 (PMIDs: 21841781, 29674594), the first report focusing on variants in TAOK1 as a cause of autosomal dominant syndromic intellectual disability was published in 2019 (PMID: 31230721). TAOK1 (thousand and one amino acid kinase 1) encodes the serine/threonine-protein kinase TAO1. Individuals with pathogenic variants in this gene present with variable degrees of developmental delay and/or intellectual disability, behavior problems (including autism in some), hypotonia, macrocephaly, dysmorphic facial features, and joint hypermobility (PMID: 33565190). Truncating variants in this gene have been reported in 18 individuals in 2 publications that included detailed phenotype information and were the basis of this curation (PMIDs: 31230721, 33565190), although only 6 probands were scored in this curation because the maximum score for genetic evidence has been reached. Most variants occurred de novo; in two instances, they were inherited from affected parents. Additional de novo truncating variants have been identified by exome sequencing in affected individuals from large cohort studies of autism spectrum disorder and developmental disorders, but detailed phenotype information is not available in those studies (PMIDs: 31981491, 33057194). Furthermore, nine de novo TAOK1 missense variants were reported in the 2 publications focused on TAOK1 (PMIDs: 31230721, 33565190). Although the clinical interpretation of these variants is difficult in the context of nonspecific phenotypes, functional analysis showed that some missense variants resulted in loss of protein function, while others led to dominant-negative effects (PMID: 33565190). Thus, while the mechanism for disease is predicted to be haploinsufficiency based on protein truncating variants reported in affected individuals and functional studies in patient-derived fibroblasts (PMID: 31230721), further studies are needed to confirm the mechanisms of missense variants. This gene-disease relationship is also supported by experimental evidence, including non-human model organisms (PMIDs: 26996505, 31230721, 33565190) and functional in vitro studies (PMID: 14517247, 33565190). In summary, there is sufficient genetic evidence to support a definitive gene-disease relationship between TAOK1 and syndromic intellectual disability. This classification was approved by the ClinGen Intellectual Disability and Autism Gene Curation Expert Panel on 08/04/21 (SOP Version 8).

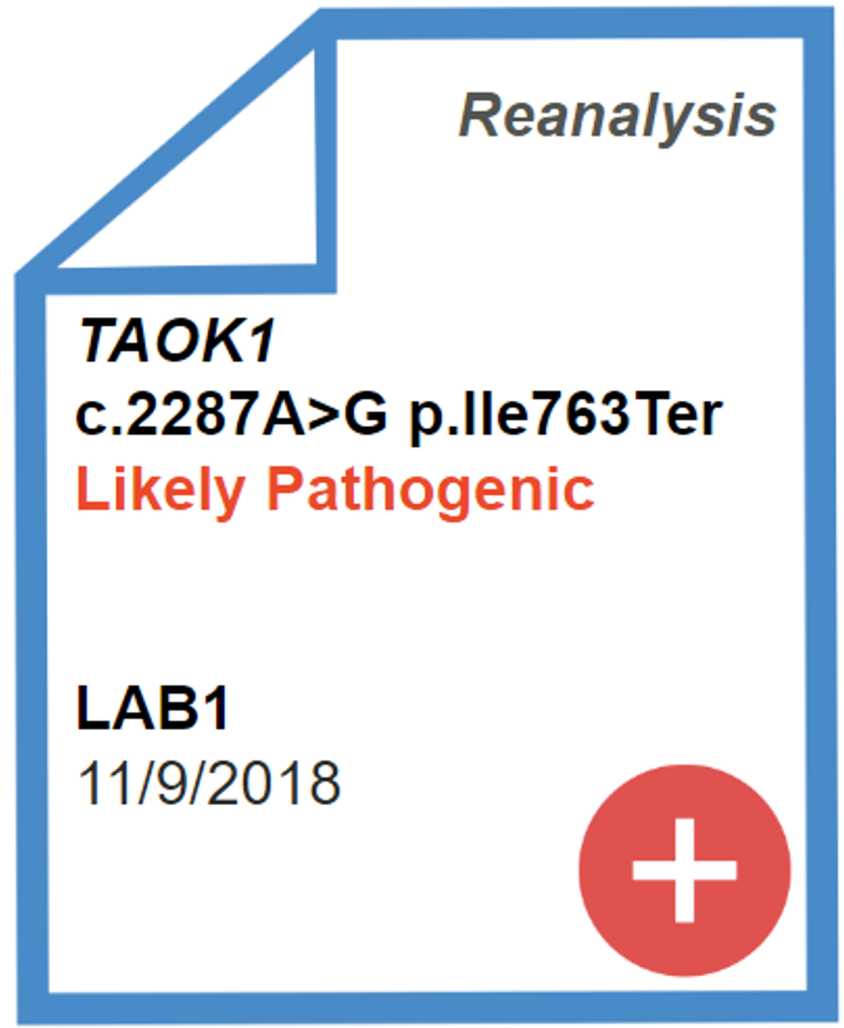
Gene Clinical Validity Standard Operating Procedures (SOP) - SOP8

Role has been repeatedly demonstrated in research & clinical diagnostic settings

- Upheld over time (in general, at least 3 years)
- No convincing contradictory evidence

https://bit.ly/ClinGenGene

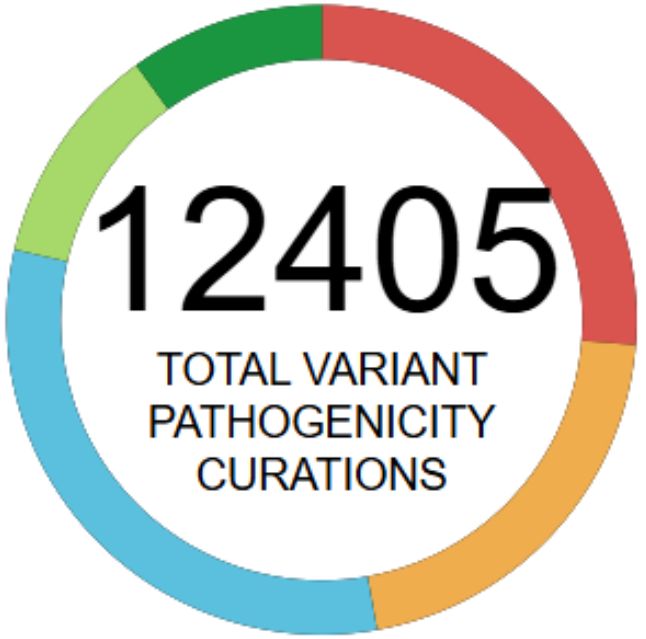
Case Example



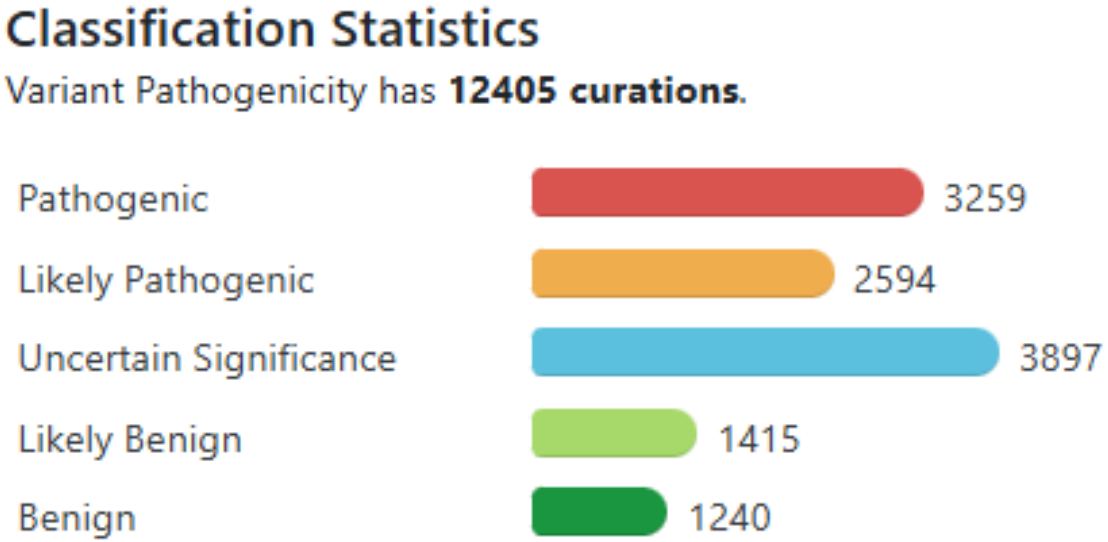
https://bit.ly/ClinGenGene

Variant Pathogenicity

- Which variants in a given gene cause disease?
- Variant Curation Expert Panels (VCEPs) provide gene/disease-level specifications to the 2015 ACMG/AMP sequence variant interpretation guidelines[1]
- Specifications available for public use[2]
- VCEPs use these to classify difficult variants and resolve discrepancies in ClinVar
- FDA-recognized process[3]



- **40** fully approved VCEPs
- **76** total VCEPs in various stages of development
- **14** different clinical domains



[1]Richards *et al.* 2015; PMID:25741868
[2]https://cspec.genome.network/cspec/ui/svi/
[3]https://clinicalgenome.org/about/fda-recognition/

As of April 14, 2026

Case Example

- 35-year-old female presents to cancer genetics clinic for family history of gastric cancer
- Patient shares her paternal grandfather and father have had genetic testing

Grandfather, gastric cancer age 43

CDH1
NM_004360.5
c.1057G>A (p.Glu353Lys)
Likely Pathogenic

LAB1
2/26/2021

Father, no personal history of gastric cancers

CDH1
NM_004360.5
c.1057G>A (p.Glu353Lys)
VUS

LAB2
11/9/2021

https://bit.ly/VariantCuration

NM_004360.5(CDH1):c.1057G>A (p.Glu353Lys)

Cite | Follow | Print | Download

Germline

Top reviewed classifications are shown here. Submission summary: 5 submissions 5 submitters 4 conditions

Reviewed by expert panel ★★★☆

Likely pathogenic
Aug 2023 by ClinGen CDH1 V...
FDA RECOGNIZED DATABASE

for CDH1-related diffuse gastric and lobular breast cancer syndrome

Somatic

No data submitted for somatic clinical impact

Somatic

No data submitted for oncogenicity

Classification (Last evaluated)	Review status (Assertion criteria)	Condition	Submitter	More information
Likely pathogenic (Aug 24, 2023)	★★★☆ (ClinGen CDH1 ACMG Specifications V3.1) Method: curation	CDH1-related diffuse gastric and lobular breast cancer syndrome (Autosomal dominant inheritance) Affected status: unknown Allele origin: germline	ClinGen CDH1 Variant Curation Expert Panel FDA RECOGNIZED DATABASE Accession: SCV001943339.2 First in ClinVar: Sep 29, 2021 Last updated: Sep 20, 2023	

Publications:
PubMed: 32133419

Other databases
https://erepo.clinicalgenome.org/evrepo/ui/interpretation/60a7410c-1a25-4d28-8b31-90b524be8a48

Comment:
The c.1057G>A (p.Glu353Lys) variant is absent in the gnomAD cohort (PM2_supporting; http://gnomad.broadinstitute.org). This variant has been reported in at least tw families meeting HDGC clinical criteria (PS4_moderate; SCV001378233.1, SCV000580696.4). There is an RNA assay demonstrating an abnormal out-of-frame transcript for this variant (PS3; PMID: 32133419). This variant is predicted to affect splicing by SpliceAI (Donor Gain: score = 0.54 at -3 bp) (PP3). In summary, this variant meets criteria to be classified as likely pathogenic based on the ACMG/AMP criteria applied as specified by the CDH1 Variant Curation Expert Panel (v3.1): PS3, PS4_moderate, PM2_supporting, PP3 (less)

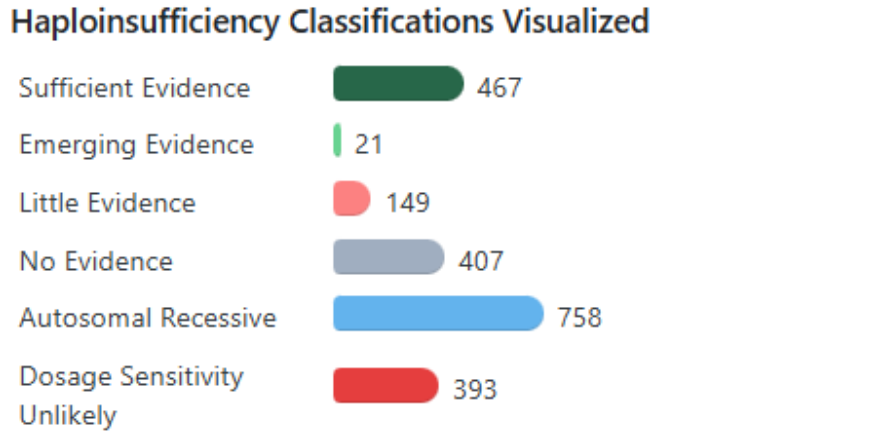
Dosage Sensitivity

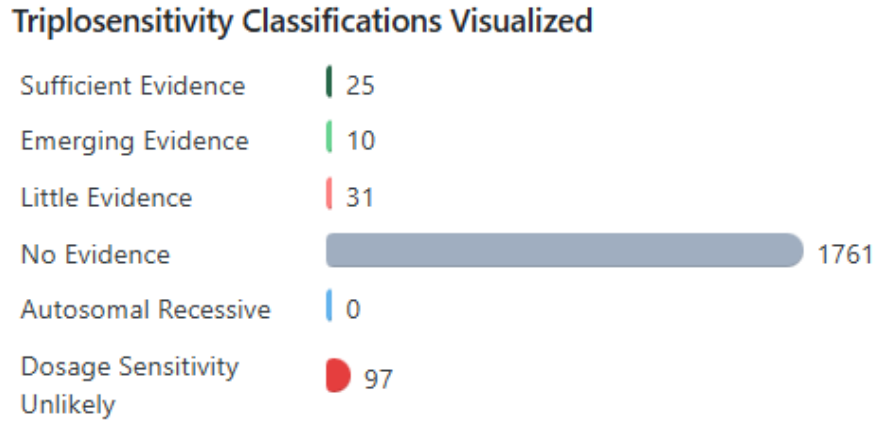
- Does loss or gain of an entire copy of this gene or genomic region cause disease?
  - Haploinsufficiency (HI)
  - Triplosensitivity (TS)
- Scoring metric based on observations in human cases[1,2]
- Information can be used to aid in the classification of copy number variants (CNVs)
  - Part of ACMG/ClinGen technical standards for constitutional CNV evaluation[3]
  - Can guide discrepancy resolution efforts between laboratories[4]
- Can also be used to identify genes for which HI and/or TS are valid mechanisms for disease[5]
  - E.g. Is PVS1 applicable to loss of function variants in this gene?

4119
Total
Dosage Sensitivity Curations

1680
Single Genes
Evaluated

516
Genomic Regions
Evaluated





As of April 14, 2026

[1]Riggs *et al.* 2012; PMID:22097934
[2]https://clinicalgenome.org/docs/dosage-standard-operating-procedure-scoring-guide/
[3]Riggs *et al.* 2020; PMID:31690835
[4]Riggs *et al.* 2018; PMID:30095202
[5]Thaxton *et al.* 2021; PMID:34694049

Case Example

- You are preparing to see a child with intellectual disability and dysmorphic features, including micrognathia
- History of microarray in 2015, no further testing since
  - Deletion of 5p15.2 was identified and classified as VUS at the time
- Should you order additional testing?

arr(GRCh37)chr5p15.2 (13,701,224-14,903,954)x1

Variant of Uncertain Significance

Genes included:
DNAH5, TRIO, OTULINL, ANKH

Lab 1
12/20/2015

https://bit.ly/DosageCuration

TRIO

Gene Facts

3 Haplo Score

0 Triplo Score

Dosage Sensitivity Summary (Gene)

Dosage ID: **ISCA-25787**
View legacy report...

Curation Status: **Complete**

Issue Type: **Dosage Curation - Gene**

Haploinsufficiency: **Sufficient Evidence for Haploinsufficiency (3)**
Read full report...

Triplosensitivity: **No Evidence for Triplosensitivity (0)**
Read full report...

Last Evaluated: **01/24/2018**

https://search.clinicalgenome.org/kb/gene-dosage/HGNC:12303

https://bit.ly/DosageCuration

Haploinsufficiency (HI) Score Details

HI Score: 3

HI Evidence Strength: **Sufficient Evidence for Haploinsufficiency** (Disclaimer)

HI Disease: micrognathia-recurrent infections-behavioral abnormalities-mild intellectual disability syndrome Monarch

HI Evidence:

PUBMED: 25533962

The 'Deciphering Developmental Disorders' (DDD) study (Nature, 2015) reported finding 2 patients with de novo missense and a patient with a de novo 82Kb deletion of 16 exons in the TRIO gene.
The study used a combination of exome sequencing and array-based detection of chromosomal rearrangements and included 1,133 children with severe, undiagnosed developmental disorders and their parents.

PUBMED: 25363760

De Rubeis et al. (2014) used exome sequencing and statistical analyses to identify autism associated genes. TRIO was identified as one such candidate gene as a result of a de novo loss of function mutation in one case (out of 3,871 autism cases).

PUBMED: 26721934

Ba et al (2016) reported the detection of a de novo deletion of TRIO in a child with intellectual disability (ID).
Three additional cases with truncating mutations were found by targeted sequencing of the TRIO gene in over 2300 patients with ID.
The authors indicate that the data supports loss of TRIO function as causal for the clinical phenotype.

https://bit.ly/DosageCuration

https://search.clinicalgenome.org/kb/gene-dosage/HGNC:12303

Case Example

arr(GRCh37)chr5p15.2
(13,701,224-14,903,954)x1

Pathogenic

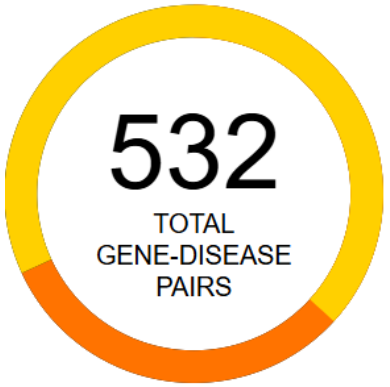
Genes included:
DNAH5, TRIO, OTULINL, ANKH

Lab 1
9/2/2022

https://bit.ly/DosageCuration

Clinical Actionability

- Which genes, when significantly altered, confer a high risk of serious disease that could be prevented or mitigated if the risk were known?
- Semi-quantitative scoring metric[1,2] assesses outcome-intervention pairs based on:
  - Severity of outcome
  - Likelihood of disease
  - Effectiveness of intervention
  - Burden/risk of intervention to the patient
- Information can be used to guide decisions on reporting secondary findings[3]

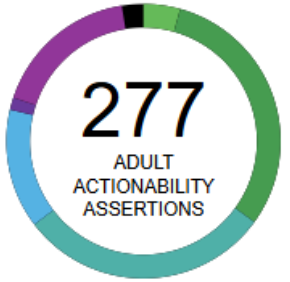


276 Total Clinical Actionability Topics

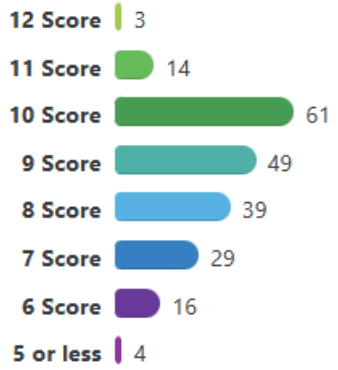
227 Total Actionability Reports	49 Total Topics Failed Early Rule-out
297 Total Genes Included in Actionability Reports	51 Total Actionability Updated Reports

Adult Context

119 Adult Actionability Reports
28 Total Topics Failed Early Rule-out
257 Unique Genes In Adult Actionability Topics

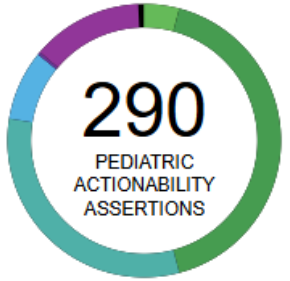


215 Total Adult Outcome-Intervention Scored Pairs

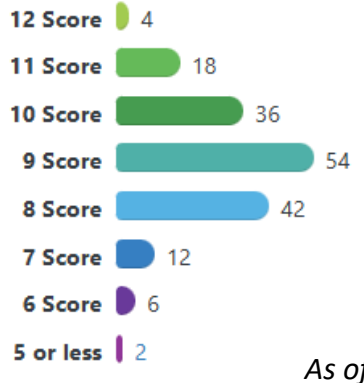


Pediatric Context

108 Pediatric Actionability Reports
21 Total Pediatric Topics Failed Early Rule-out
256 Unique Genes In Pediatric Actionability Topics



174 Total Pediatric Outcome-Intervention Scored Pairs



As of April 14, 2026

[1]Hunter *et al.* 2016; PMID:27124788
[2]Webber *et al.* 2018; PMID:30311382
[3]Miller *et al.* 2021; PMID:34012068

Case Example

- Your team is developing a research protocol that will involve genome sequencing in children, and you've been asked to come up with the approach for secondary/incidental finding return.
- Which genes have actionability implications in the pediatric setting?

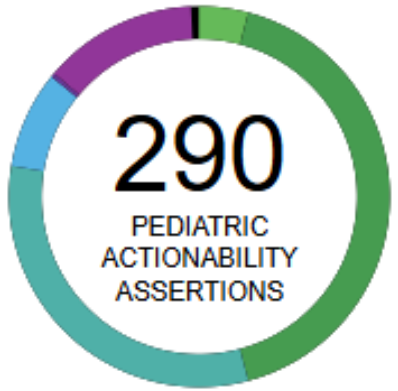
https://bit.ly/ClinGenActionability

Pediatric Context

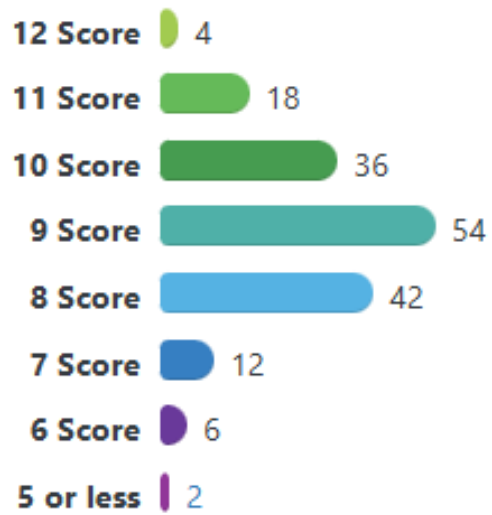
108 Pediatric Actionability Reports

21 Total Pediatric Topics Failed Early Rule-out

256 Unique Genes In Pediatric Actionability Topics



174 Total Pediatric Outcome-Intervention Scored Pairs



As of April 14, 2026

Case Example

Clinical Actionability

Gene	Disease	Report	Working Group	Assertions	Report & Date
FBN1	Marfan syndrome MONDO:0007947	Marfan Syndrome	Adult Actionability WG	Adult Definitive Actionability	03/01/2019
			Pediatric Actionability WG	Pediatric Definitive Actionability	03/01/2019
FBN1	familial thoracic aortic aneurysm and aortic dissection MONDO:0019625	Familial thoracic aortic aneurysms and dissections (FTAAD)	Adult Actionability WG	Adult Strong Actionability	03/01/2019
			Pediatric Actionability WG	Pediatric Strong Actionability	03/01/2019

2. How effective are interventions for preventing harm?

Information on the effectiveness of the recommendations below was not provided unless otherwise stated.

Patient Management	Management of thoracic aortic aneurysm and/or dissection requires coordinated input from a multidisciplinary team of specialists familiar with FTAAD, including a clinical geneticist, cardiologist, and cardiothoracic and vascular surgeons. **(Tier 4)**	(2)
	Prophylactic surgical repair of the aorta is recommended at 4.5-5.0 cm for patients with pathogenic variants in MYH11, SMAD3, and ACTA2 and at 4.0-4.5 cm for patients with pathogenic variants in TGFBR1 or TGFBR2. Earlier repair can be considered in patients with a family history of aortic dissection, growth of the aorta approaches 1 cm/year, or aortic regurgitation. In patients with Marfan syndrome (MFS), timely repair of aortic aneurysms prolongs survival and approaches that of age-matched controls; however, evidence on effectiveness was not provided for patients with FTAAD. **(Tier 2)**	(5, 6, 7, 8, 9)
	Beta adrenergic-blocking agents are recommended to reduce aortic dilation. **(Tier 2)**	(7)
	Though no evidence for effectiveness of these medications is available for FTAAD, a meta-analysis of five cohort studies among children and adolescents with MFS indicated that beta-blocker treatment decreased the rate of aortic dilation compared to no treatment (standardized mean difference: -1.30; 95% CI: -2.11 to -0.49; p=0.002). A randomized trial of 70 patients with MFS aged 12-50 years showed that beta-blocker vs. no treatment slowed the rate of aortic dissection as measured by the slope of the aortic ratio, calculated by dividing the measured aortic diameter by the diameter predicted by the participant's height, weight, and age (mean slope of the aortic ratio plotted against time: 0.084 vs. 0.023, respectively). However, none of the studies demonstrated an impact on mortality, occurrence of aortic dissection, or the need for elective repair of the aorta and/or aortic valve, though these studies were likely underpowered. **(Tier 1)**	(10, 11)
	Losartan was added as an alternative to beta adrenergic-blocking agents in FTAAD after studies showed its efficacy in children and young adults with MFS who were randomly assigned to losartan or atenolol. **(Tier 3)**	(2)
	A meta-analysis of six randomized clinical trials among children and adults with MFS indicated that losartan, an angiotensin II receptor antagonist, significantly decreased the rate of aortic dilation compared to no losartan treatment (standardized mean difference: -0.13; 95% CI: -0.25 to 0.00; p=0.04). However, improvements in mortality, cardiovascular surgery, or aortic dissection or rupture were assessed but not observed. Follow-up time in these studies ranged from 35 months to 3.5 years, which may have limited the ability to assess these outcomes. **(Tier 1)**	(12)

https://bit.ly/ClinGenActionability

https://actionability.clinicalgenome.org/ac/Pediatric/ui/stg2SummaryRpt?doc=AC134

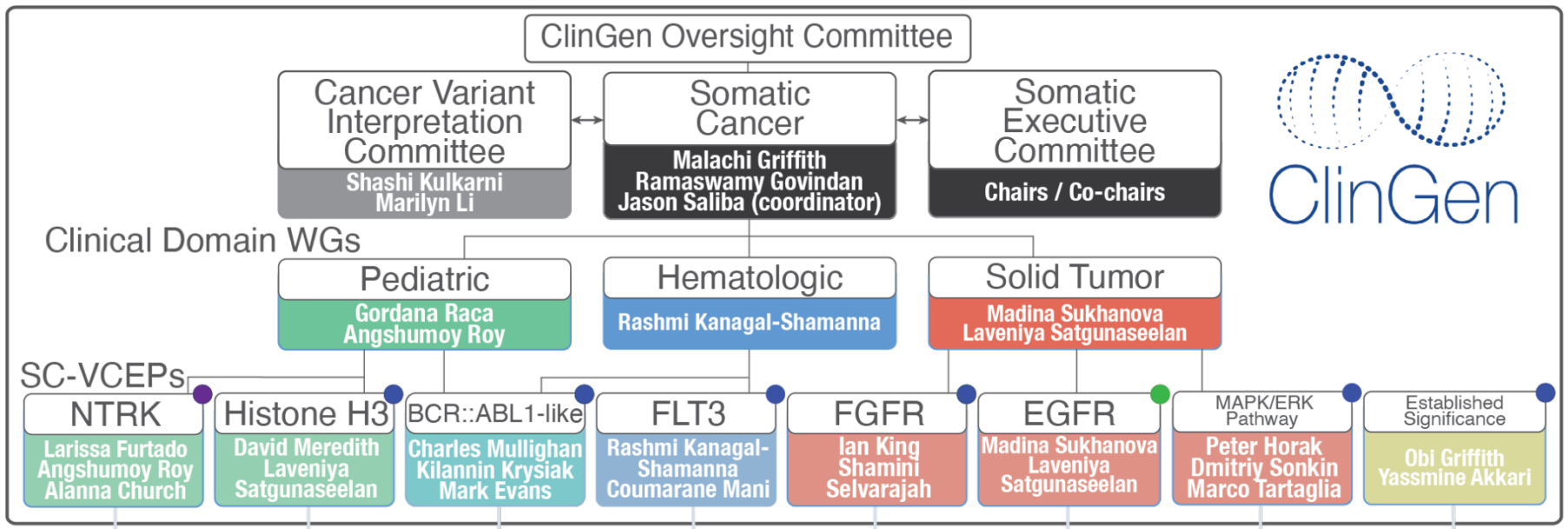
Somatic Cancer

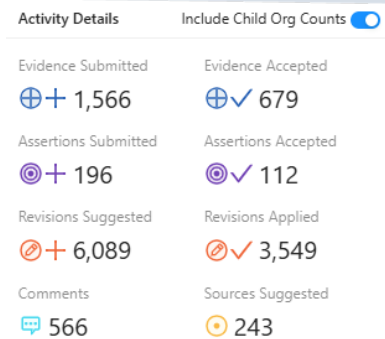
ClinGen Somatic Biomedical Knowledgebase - Received 5 year award (9/23-9/28) - FOA Number: PAR-20-097

Aim 1. Support creation of the ClinGen Somatic Knowledgebase

Aim 2. Promote FAIR (Findable Accessible Interoperable Reusable) sharing through genomic knowledge standards

Aim 3. Improve the scalability, efficiency, and sustainability of somatic variant curation (using NLP approaches)



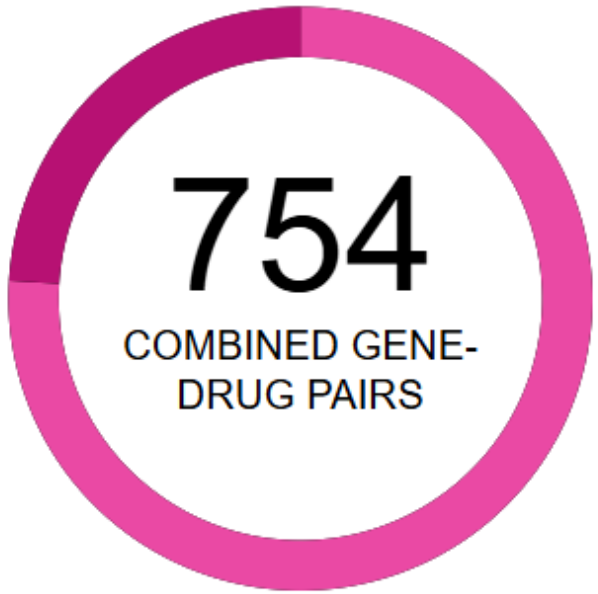


Pharmacogenomics

The overarching goal of the Pharmacogenomics is to study the variances in genes and their effects on drug response.

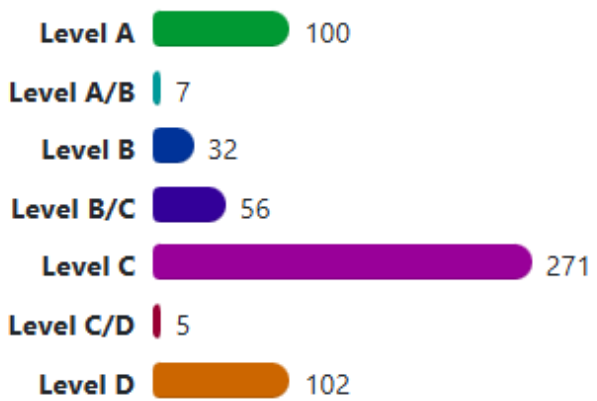
754 Combined Pharmacogenomics Gene-Drug Pairs

573	181
CPIC Gene-Drug Pairs	PharmGKB Gene-Drug Pairs
121	**47**
Genes Included in CPIC Gene-Drug Pairs	Genes Included in PharmGKB Gene-Drug Pairs



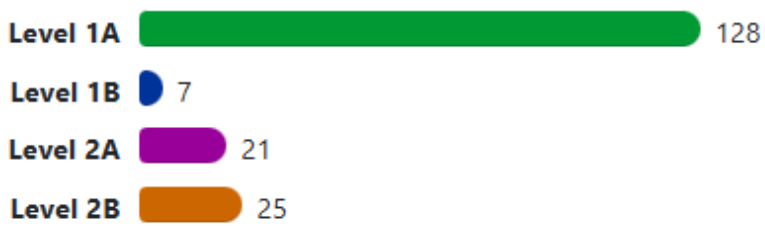
CPIC Gene-Drug Pairs by Highest Level of Actionability

Highest Levels Visualized



PharmGKB Gene-Drug Pairs by Highest Level of Evidence

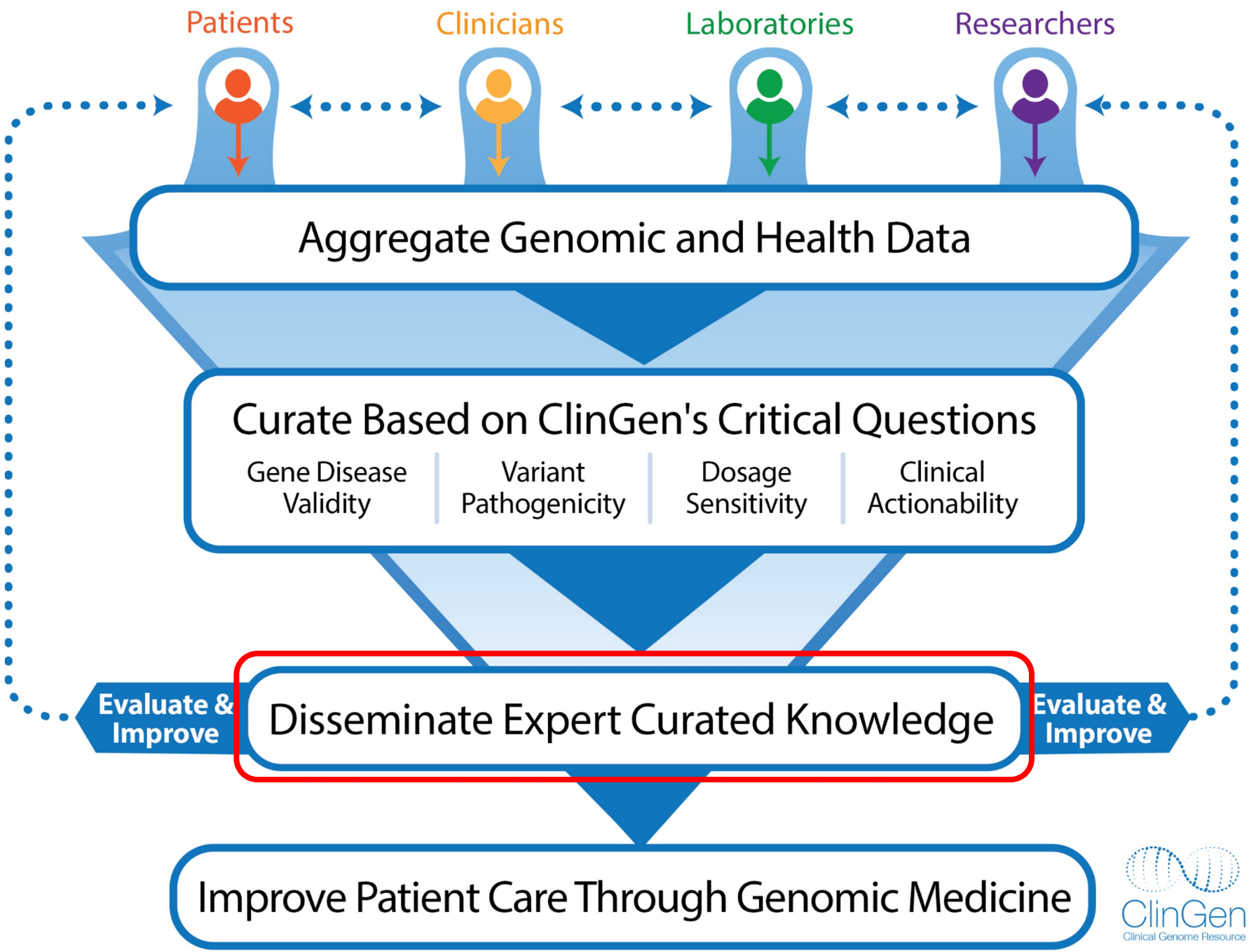
Highest Levels Visualized



As of April 14, 2026

ClinGen Pharmacogenomics (PGx) Working Group

- Validating gene-drug clinical validity framework to assess evidence to determine if a gene is associated with a particular drug response phenotype, aligned with ClinGen Gene-Disease Clinical Validity framework
- Beginning development of analogous gene-drug clinical actionability framework
- Continuing to expand PGx representation across clinical domains and expert panels
- Synergizing ClinPGx development with ClinGen in close collaboration with NHGRI and ClinGen SC


Patients
Clinicians
Laboratories
Researchers
Aggregate Genomic and Health Data
Curate Based on ClinGen's Critical Questions
Gene Disease Validity
Variant Pathogenicity
Dosage Sensitivity
Clinical Actionability
Evaluate & Improve
Disseminate Expert Curated Knowledge
Evaluate & Improve
Improve Patient Care Through Genomic Medicine
ClinGen
Clinical Genome Resource

Dissemination: Curation Results

- Curation results are made freely and publicly available shortly after completion - not held for publication, no restrictions on use
- https://clinicalgenome.org → Primary portal for browsing curation data
- Curation data utilized and displayed by numerous downstream users:
  - Clinical and research variant repositories (ClinVar, DECIPHER)
  - Resources describing gene-disease relationships (OMIM, GenCC)
  - Genome browsers (UCSC, Ensembl)
  - Variant interpretation tools/platforms (MutSpliceDB, other commercial tools)
  - Somatic cancer variant resources (CIViC)
  - Clinical decision-making resources (ACMG ACT sheets)
  - In addition, resources also utilize the unique canonical variant IDs provided by the Allele Registry, including ClinVar, Ensembl, gnomAD, CIViC, LitVar, and others

ClinGen
Clinical Genome Resource

Get Started · About Us · Curation Activities · Working Groups · Expert Panels · Documents & Announcements · Tools

Explore the clinical relevance of genes & variants

ClinGen is a National Institutes of Health (NIH)-funded resource dedicated to building a central resource that defines the clinical relevance of genes and variants for use in precision medicine and research.

Gene · Enter a gene symbol or HGNC ID (Examples: ADNP, HGNC:15766) Search

All Curated Genes · Gene-Disease Validity · Dosage Sensitivity · Clinical Actionability · Curated Variants · Statistics · More

ClinGen is defining the clinical relevance of genes and variants

Founded in 2013 by the National Human Genome Research Institute, ClinGen is a growing collaborative effort, involving three grants, nine principal investigators and over 2,500 contributors from more than 68 countries. Below are a series of recent updates that ClinGen has been working on.

Volunteer as an Expert

Interested in contributing your expert knowledge and/or data to a ClinGen GCEP or VCEP? Learn more and take our volunteer expert survey.

File Downloads and APIs

ClinGen Downloads and APIs

Visit our File Downloads and APIs page for a summary of available ClinGen curation files and API resources.

Volunteer to Curate

Please take a brief survey to tell us more about your

ClinGen
Clinical Genome Resource

www.clinicalgenome.org

Dissemination: Selected Recent Educational Events

Masterclass in Genomic Analysis (December 2025; Doha, Qatar)

- Hosted in collaboration with Sidra Medicine as part of the Precision Medicine and the Future of Genomics 2025 Summit
- 2-day event focused on gene and variant classification and publicly available resources (In-person)

ClinGen-PAPERI Virtual Training Workshop on Variant Interpretation and Clinical Genomics

- Hosted in collaboration with the Pan-African PGS Education and Research Initiative (PAPERI)
- Month-long virtual event; weekly lectures, live Q&A, weekly WhatsApp discussion forums

19th Annual International Biocuration Conference (April 2026, Cape Town, South Africa)

- Live 90-minute workshop focused on databases and resources

Would you like ClinGen to host a workshop for your audience?

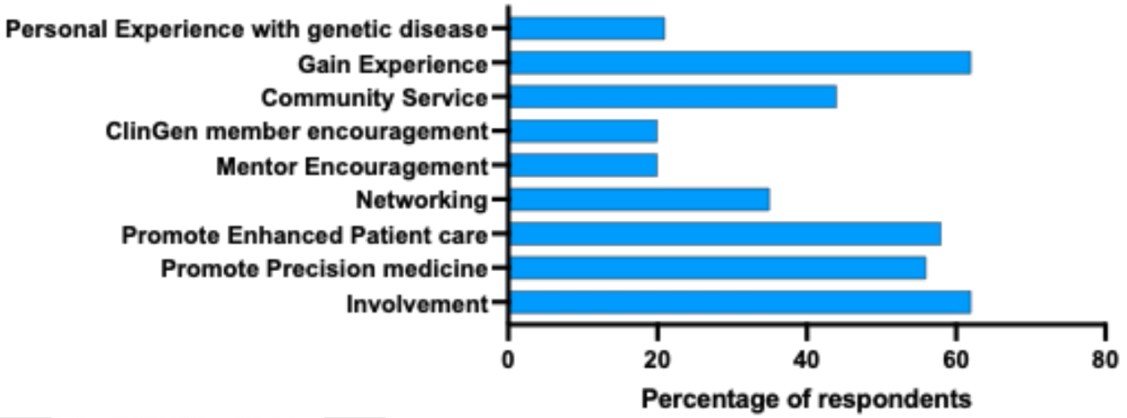
Scan the QR code for the workshop/training request form!

Dissemination: Community Curation

- ClinGen Community Curation (C3): Launched in 2018 as a way to engage interested volunteers in the ClinGen curation process
  - Crowd-sourcing curation effort → open to the broader community
  - Encouraging uptake of ClinGen curation standards
  - Learning/networking opportunity
  - Continuing education credits available
- Since launch:
  - Over 3700 people from 97 countries/territories have taken the volunteer interest survey
  - >2500 have completed training
  - Many remain active participants beyond requested 6 month commitment

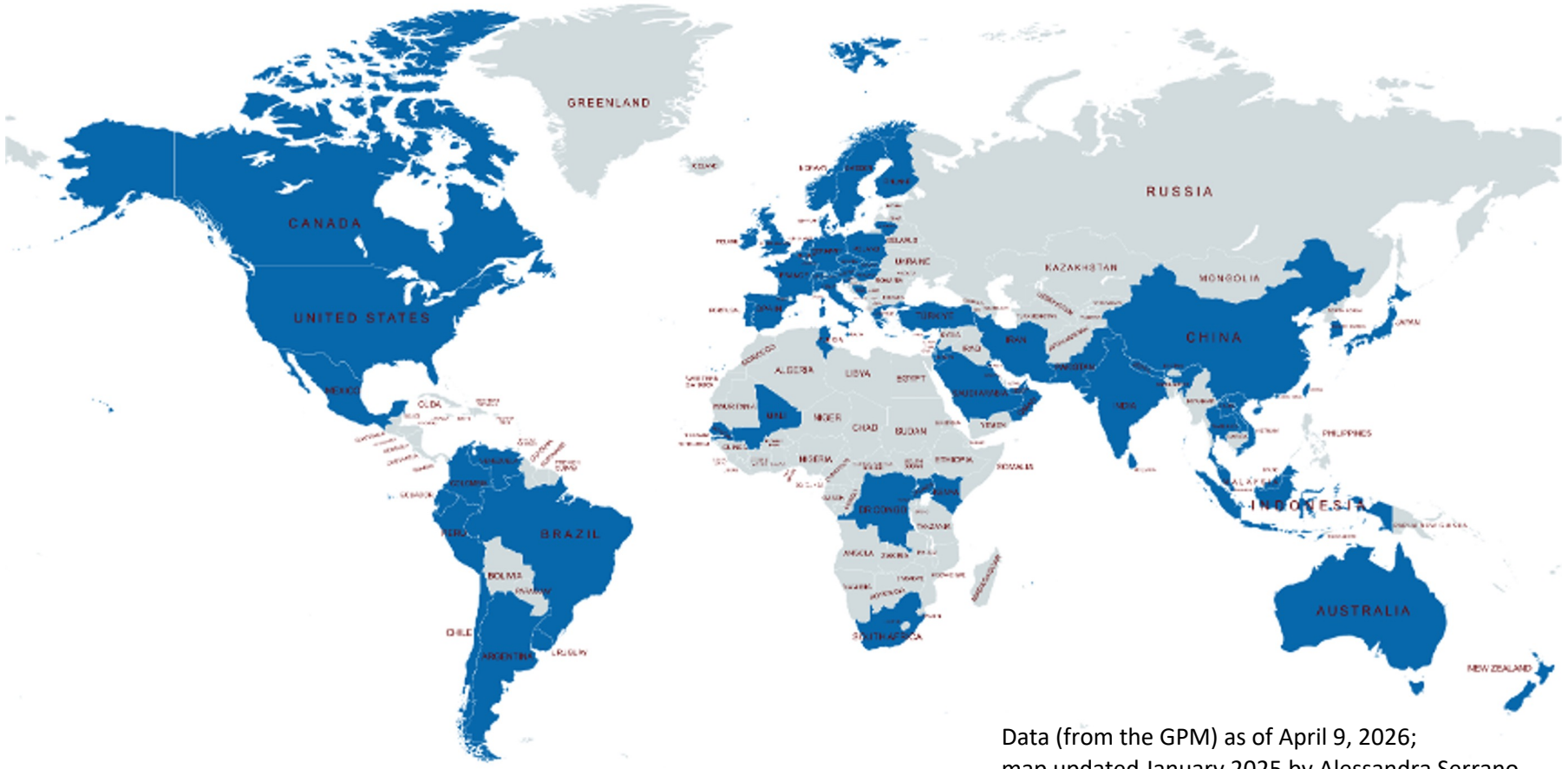
Why do people volunteer with ClinGen?

(multiple select)



Interested in volunteering? Scan the QR code to take the sign-up survey!

Acknowledgements



Data (from the GPM) as of April 9, 2026;
map updated January 2025 by Alessandra Serrano Marroquin

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Primary Funding Sources:
ClinGen is primarily funded by the National Human Genome Research Institute (NHGRI) through the following three grants:

- Baylor/Stanford: U24HG009649 (with co-funding from the National Cancer Institute)
- Broad/Geisinger: U24HG006834
- UNC & Co.: U24HG009650

ClinGen's work would not be possible without the valuable contributions of our **2,820** working group and expert panel members (from **76** countries).

Questions?
clingen@clinicalgenome.org