Expanding access to ClinGen's evidence-based expert curation efforts

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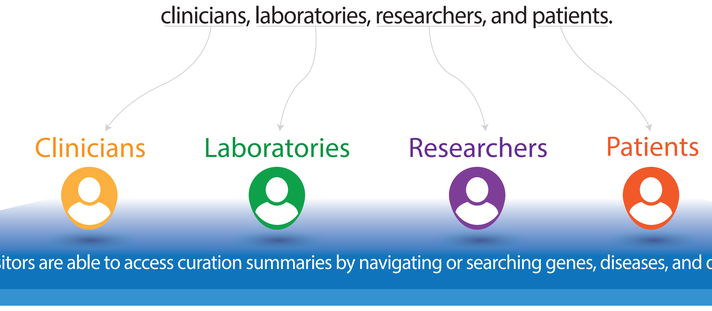
The NIH-funded Clinical Genome Resource (ClinGen) is dedicated to building an authoritative central resource that defines the clinical relevance of genes and variants for use in precision medicine and research.

ClinGen working groups have developed protocols to evaluate critical questions necessary to build a genomic knowledge base: Which genes, when altered, are implicated in human disease (gene-disease validity)? Which variants within these genes cause disease (variant pathogenicity)? How does this information affect medical management (clinical actionability)? Is haploinsufficiency or triplosensitivity a mechanism for disease (dosage sensitivity)?

The ClinGen website (www.clinicalgenome.org) has evolved into a hub through which ClinGen's curation activities and links to vetted external resources is available to clinicians, laboratories, researchers, and patients.



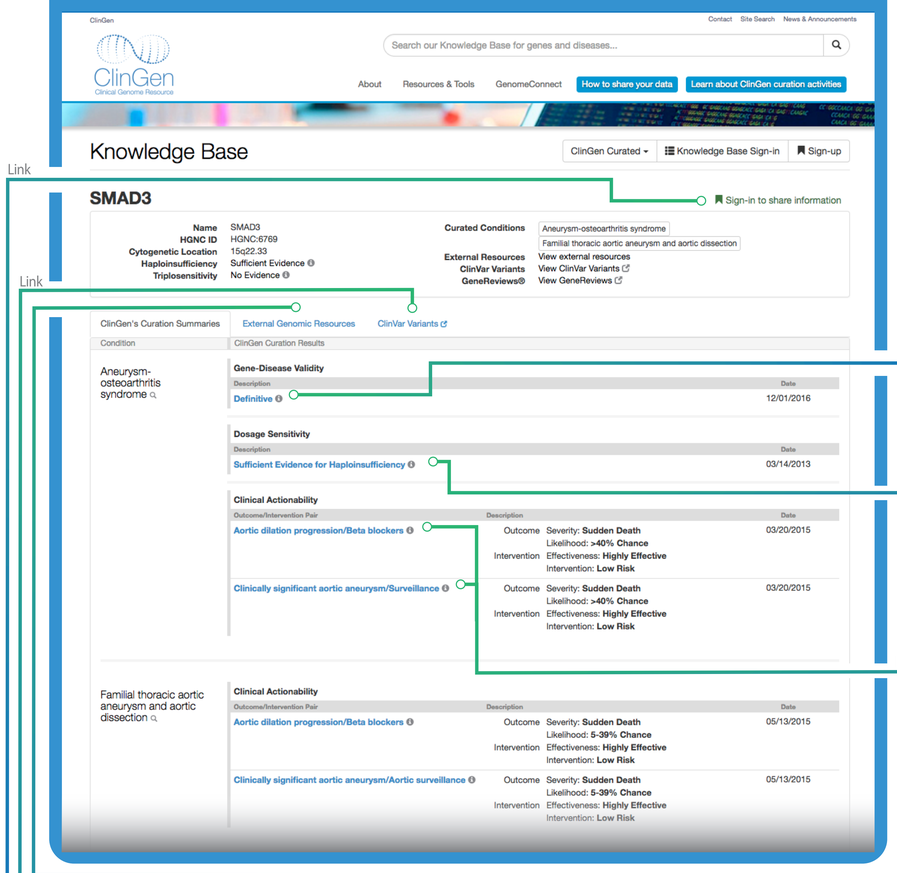
ClinGen began releasing curated information through the website in February of 2015. In March of 2016, a search interface was added to allow visitors to search for curated information by gene and condition. In March of 2017, gene-centric and disease-centric views were incorporated. Search capabilities were enhanced by using a graph database, a database designed to handle highly connected information. This enables the search interface to manage the complex relationships between ontologies and ClinGen's curation summaries to allow more precise matching and long-term flexibility.

Gene-centric View

The gene-centric view displays general information about a gene and organizes gene-disease validity, dosage sensitivity, and clinical actionability curation summaries by disease. The view also provideslinks to curation details, associated diseases, and external resources such as ClinVar.

SMAD3

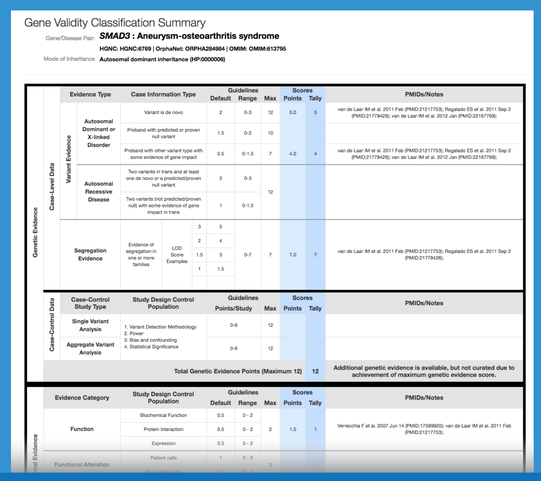
Gene-centric curation summaries



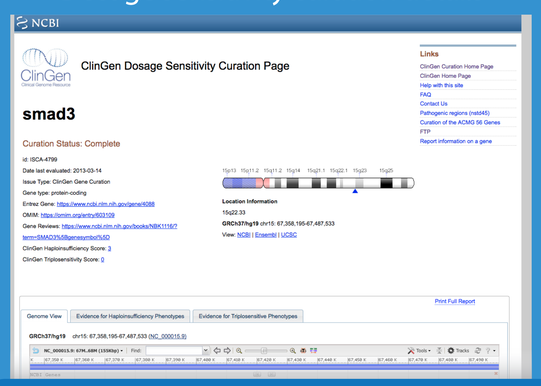
ClinGen Curation Information

ClinGen provides curation summaries in an effort to disseminate the collective knowledge and resources for unrestricted use in the community. This information may be found by performing a search for a gene or disease, or visitors can view a list of all ClinGen curation summaries.

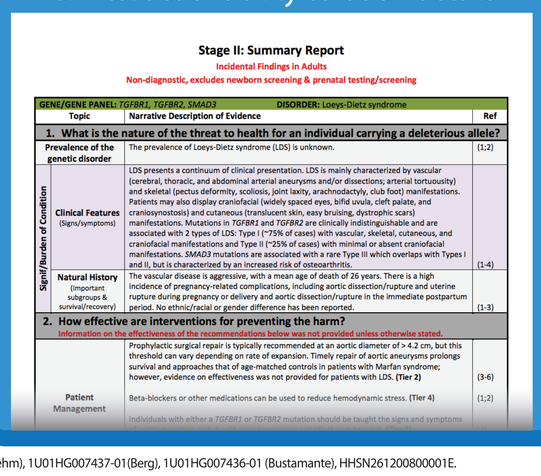
Gene-disease validity curation details



Dosage sensitivity curation details



Clinical actionability curation details

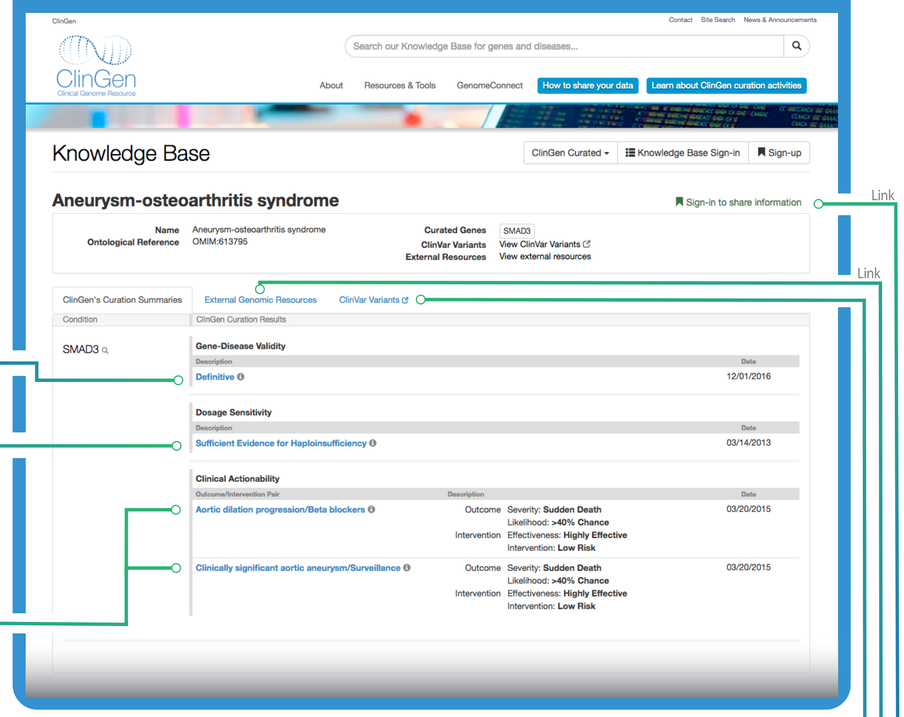


Disease-centric View

The disease-centric view displays general information about a disease and organizes gene-disease validity, dosage sensitivity, and clinical actionability curation summaries by gene. The view also provides links to curation details, associated genes, and external resources such as ClinVar.

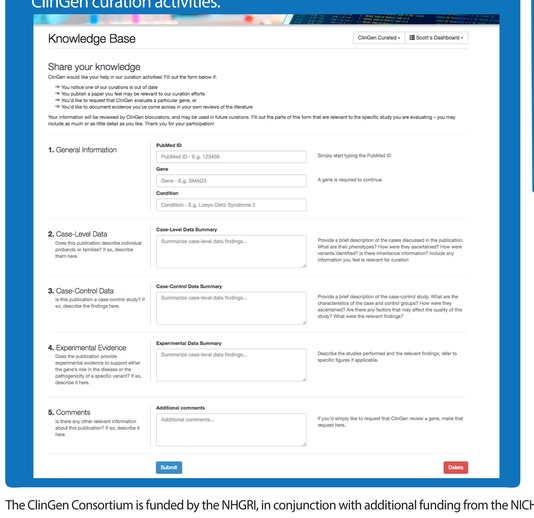
Aneurysm-osteoarthritis syndrome

Disease-centric curation summaries



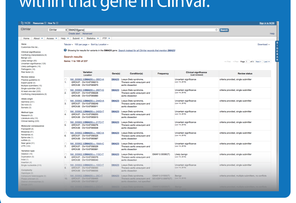
Share Gene Information

Visitors can share information from the literature to supplement ClinGen curation activities.



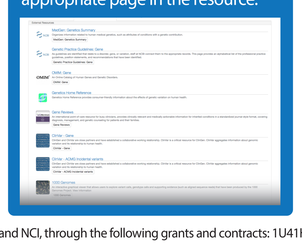
ClinVar Variants

Links directly to a search for variants within that gene in ClinVar.



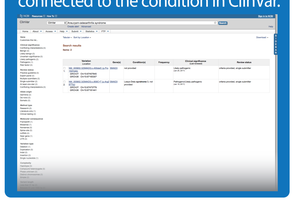
External Resources

Links take visitors to the most appropriate page in the resource.



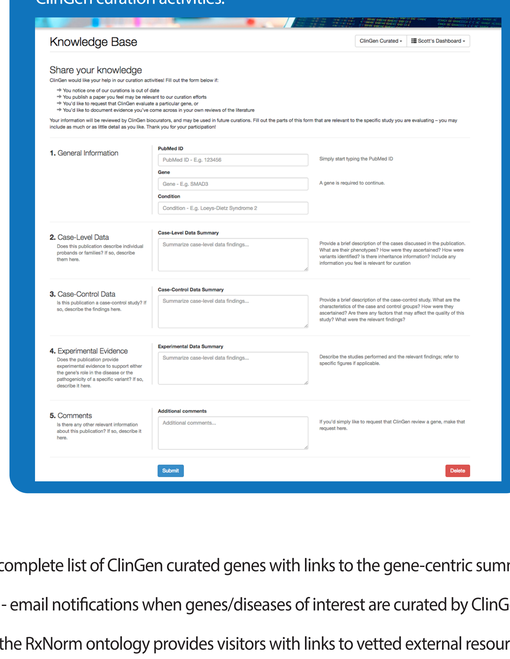
ClinVar Variants

Links directly to a search for variants connected to the condition in ClinVar.



Share Disease Information

Visitors can share information from the literature to supplement ClinGen curation activities.



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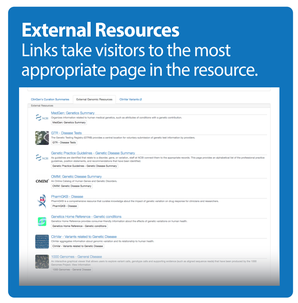

Additional Features

- **All ClinGen Curation Summaries List:** A complete list of ClinGen curated genes with links to the gene-centric summary.
- **Follow ClinGen Curations:** Coming Soon - email notifications when genes/diseases of interest are curated by ClinGen.
- **Drug Search:** Searching for a drug within the RxNorm ontology provides visitors with links to vetted external resources.

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