

MedGen, ClinVar, and GTR[®]

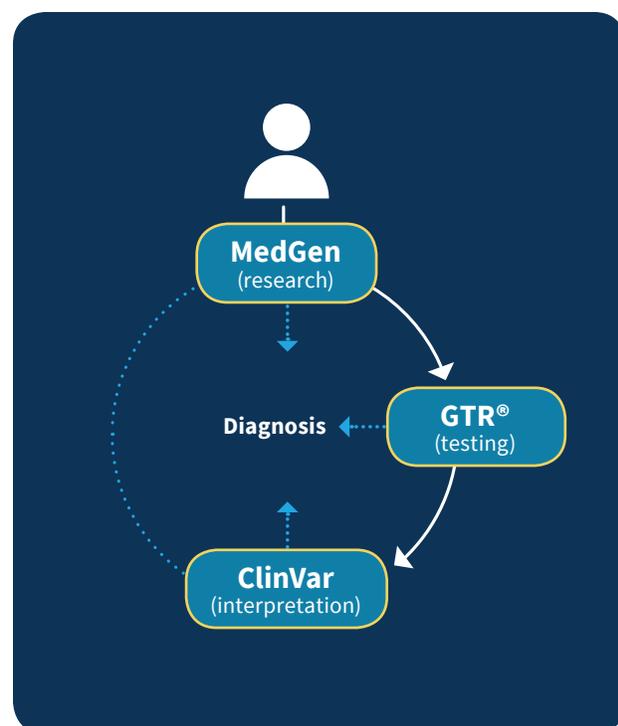


Using NCBI's medical genetics and human variation resources to research, diagnose and treat genetic conditions.

CASE STUDY: SUSPECTED MARFAN SYNDROME

A 9-year-old boy comes into the clinic for a medical release to play soccer. The child presents with myopia, arachnodactyly and has a family history of aortic dissection. These are flags that prompt you to investigate a possible genetic disease.

- 1** Search **MedGen** to help develop a differential diagnosis and learn about a condition, its diagnosis and etiology.
- 2** Search the NIH **Genetic Testing Registry (GTR[®])** to find the most appropriate genetic test for your patient – for example a panel that includes all the conditions in your differential diagnosis. Learn about the test's validity and utility, and find the ordering information.
- 3** Following testing, compare the test results to interpreted variants in **ClinVar** and determine pathogenicity
- 4** Return to **MedGen** for practice guidelines from medical and professional societies and the latest research to guide your treatment options. Access consumer resources to help your patient and his family understand his diagnosis, prognosis and available help. If the family is interested, check if there are available studies in [ClinicalTrials.gov](https://clinicaltrials.gov)



ABOUT OUR RESOURCES

ClinVar has more than 859,000 submitted records representing more than 542,000 unique variants from 1,314 submitters

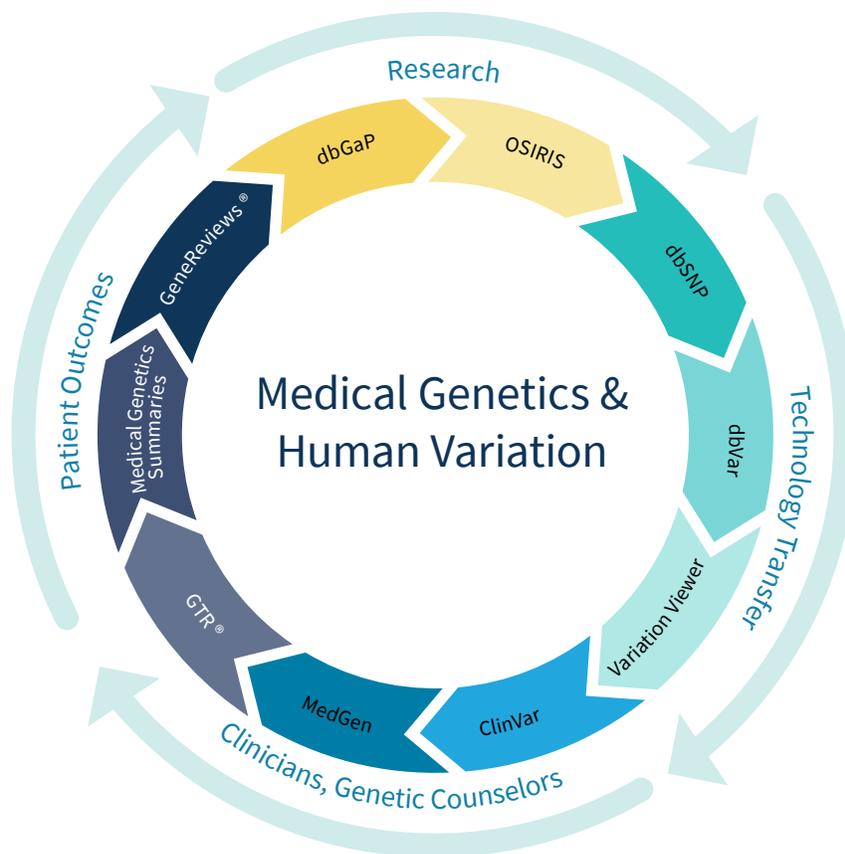
GTR[®] has 60,000+ tests, 12,000 conditions, 16,000+ genes, from 530 labs. It includes clinical and research molecular, cytogenetic and biochemical genetic tests.

MedGen helps research thousands of genetic phenotypes including Mendelian disorders, complex diseases, clinical features and drug responses. It aggregates information from authoritative resources so from one website you can access most available clinical, consumer and molecular resources.

SHARE YOUR DATA!

<https://go.usa.gov/xVPdr>

<https://go.usa.gov/xVPd2>



ClinVar

user-submitted database for information about genomic variation and its relationship to human health.



dbGaP

database for genotype and phenotype research studies.



dbSNP and dbVar

databases of small and large genomic variants including both common variations and clinical mutations.



GTR[®]

provider-submitted database of clinical and research molecular, cytogenetic and biochemical genetic tests and supporting information.



MedGen

aggregates information from and provides access to authoritative medical genetics resources.



Medical Genetics Summaries and GeneReviews[®]

up-to-date, peer-reviewed, medically actionable summaries for heritable diseases and pharmacogenetics.



OSIRIS

Open source short tandem repeat (STR) analysis tool for forensic, clinical and research use.



Variation Viewer

interactive browser for examination of nucleotide variants in a genomic context.