MedGen
Free information related to human medical genetics
https://go.usa.gov/xP9Cz

Get all clinical features of a disorder
Find all disorders with particular features based on data from HPO and OMIM

Records for
- Mendelian disorders
- Pharmacological response phenotypes
- Complex disorders
- Clinical features or findings

Data available for bulk download

Content-specific links to
- PubMed
- ClinicalTrials.gov
- GARD
- Orphanet
- PubMedHealth
- MedlinePlus

List of professional practice guidelines

Access disease and phenotype information from multiple resources including
- GeneReviews
- Genetics Home Reference
- Medical Genetics Summaries
- OMIM
- PharmGKB

Links to
- ClinVar to a list of variants relevant for the condition
- GTR to a list of orderable tests

Need help? Have suggestions about how to make us better? Contact us at medgen_help@ncbi.nlm.nih.gov.