GENEReviews®

Collection of journal-style, expert-authored and peer-reviewed content with descriptions for heritable diseases and conditions

https://go.usa.gov/xP5u4

Diagnosis, management & genetic counseling

Search by
Disorder, Text or Author, Gene Symbol or Protein Name

725 chapters with links to disease-specific and/or general consumer resources

Chapters focused on
- A single gene or phenotype
- Summaries of genetic causes of common conditions

Breast Cancer
- Myotonic Dystrophy
- Beta-Thalassemia
- Marfan Syndrome
- Williams Syndrome
- Prader-Willi Syndrome
- Noonan Syndrome

Hereditary Hearing Loss
- Cleidocranial Dysplasia Spectrum Disorder
- Urea Cycle Disorders
- Dystrophinopathies
- Angelman Syndrome
- Mitochondrial Disorders

Neurofibromatosis

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Hypermobile Ehlers-Danlos Syndrome
- Lynch Syndrome
- Huntington Disease

Beckwith-Wiedemann Syndrome
- Charcot-Marie-Tooth Hereditary Neuropathy

Ovarian Cancer
- Spinal Muscular Atrophy
- Hereditary Ataxia
- Tuberous Sclerosis Complex

Recent reviews
- Leber Congenital Amaurosis / Early-Onset Severe Retinal Dystrophy Overview
- Asparagine Synthetase Deficiency
- Hereditary Ataxia Overview
- Krabbe Disease

Genetic counseling and testing terms used in GeneReviews® are linked to:
- Definitions in the glossary
- Educational materials
- Resources for genetic professionals

Questions or feedback?
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