



What's New in GeneReviews

Updated: 16 May 2024

Newly Posted

- [CTCF-Related Disorder](#) 25 Apr 2024
- [KMT2E-Related Neurodevelopmental Disorder](#) 18 Apr 2024
- [Osteoglophonic Dysplasia](#) 18 Apr 2024
- [CDKL5 Deficiency Disorder](#) 11 Apr 2024
- [DYNC1H1-Related Disorders](#) 21 Mar 2024

Updated

- [Factor V Leiden Thrombophilia](#) 16 May 2024
- [X-Linked Lymphoproliferative Disease](#) 16 May 2024
- [Autosomal Dominant Epilepsy with Auditory Features](#) 09 May 2024
- [Mitochondrial DNA-Associated Leigh Syndrome Spectrum](#) 09 May 2024
- [Schmid Metaphyseal Chondrodysplasia](#) 09 May 2024
- [Silver-Russell Syndrome](#) 09 May 2024
- [INSR-Related Severe Insulin Resistance Syndrome](#) 25 Apr 2024
- [PINK1 Type of Young-Onset Parkinson Disease](#) 25 Apr 2024
- [HFE-Related Hemochromatosis](#) 11 Apr 2024
- [Autosomal Recessive Polycystic Kidney Disease – PKHD1](#) 04 Apr 2024
- [CSF1R-Related Disorder](#) 04 Apr 2024
- [Geleophysic Dysplasia](#) 28 Mar 2024
- [EZH2-Related Overgrowth](#) 21 Mar 2024
- [Long QT Syndrome Overview](#) 21 Mar 2024

Trichorhinophalangeal Syndrome 21 Mar 2024

Revised

FMR1 Disorders 16 May 2024

22q11.2 Deletion Syndrome 09 May 2024

SETBP1 Haploinsufficiency Disorder 09 May 2024

Spinal Muscular Atrophy, X-Linked Infantile 09 May 2024

Arylsulfatase A Deficiency 25 Apr 2024

Charcot-Marie-Tooth Hereditary Neuropathy Overview 25 Apr 2024

GJB1 Disorders: Charcot-Marie-Tooth Neuropathy (CMT1X) and Central Nervous System Phenotypes 25 Apr 2024

Kabuki Syndrome 25 Apr 2024

LZTR1- and SMARCB1-Related Schwannomatosis 25 Apr 2024

Fabry Disease 11 Apr 2024

Mucopolysaccharidosis Type I 11 Apr 2024

Mucopolysaccharidosis Type II 11 Apr 2024

Myotonic Dystrophy Type 1 21 Mar 2024

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Revisions. Whenever a clinically relevant change in information occurs, revisions to a *GeneReview* entry are initiated by staff or the author(s).

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