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What's New in GeneReviews

Updated: 30 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

Updated

- Hereditary Transthyretin Amyloidosis 30 May 2024
- Pyruvate Carboxylase Deficiency 30 May 2024
- ALK-Related Neuroblastic Tumor Susceptibility 23 May 2024
- Caffey Disease 23 May 2024
- 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

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Revised

Alpha-Thalassemia 23 May 2024

Danon Disease 23 May 2024

Fibrodysplasia Ossificans Progressiva 23 May 2024

SHOX Deficiency Disorders 23 May 2024

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

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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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