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Journal Information

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Classification History

Year	A+	A	B	C
2026		✓	✓	
2025		✓	✓	
2024		✓	✓	
2023		✓	✓	
2021/2022		✓	✓	
2020		✓	✓	
2019		✓	✓	
2018		✓		

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Recent Articles

Mucopolysaccharidoses in Morocco: a 19-year retrospective study reveals a founder mutation in mucopolysaccharidosis type I and delineates the clinical spectrum
2026-10-27
Authors: Azzeddine Laaraje, Abdelilah Radi, Abdelhakim Ourrai et al.
DOI: 10.1097/mcd.0000000000000584

Application of deep learning and explainable AI-supported medical decision-making for facial phenotyping in genetic syndromes
2026-10-8
Authors: Ömer Sümer, Tobias Huber, Jiayi Cheng et al.
DOI: 10.1097/mcd.0000000000000581

A prenatal-onset severe phenotype of SLC35A2-related congenital disorder of glycosylation associated with a mosaic hemizygous intronic variant
2026-10-6
Authors: Yukiko Kuroda, Tomoko Saito, Yasuhiro Kawai et al.
DOI: 10.1097/mcd.0000000000000583

Expanding the phenotype of GDI1-related neurodevelopmental disorder: report of the fifth family
2026-9-29
Authors: Bryson M. Lewis, Shruti Sinha, Lois J. Starr
DOI: 10.1097/mcd.0000000000000582

Second-trimester prenatal phenotype of complete trisomy 10 with combined encephalocele and myelomeningocele
2026-9-29
Authors: Cem Terece, Osman Aydin, Şans Odabasi Aydin et al.
DOI: 10.1097/mcd.0000000000000578

Marbach–Rustad progeroid syndrome caused by a hotspot LEMD2 variant: expanding the phenotypic spectrum with skeletal, cutaneous, and renal findings
2026-9-23
Authors: Hayriye Nermin Keçeci, Ayça Burcu Kahraman, Nurdan Çiftçi et al.
DOI: 10.1097/mcd.0000000000000580

Nonimmune hydrops fetalis with skeletal dysplasia due to a novel homozygous recessive TAPT1 splice-donor variant
2026-9-14
Authors: Naren Satya Srinivas M., Shivani Mishra, Himani Pandey et al.
DOI: 10.1097/mcd.0000000000000577

Developmental delay, white matter changes, and multiple endocrine hormone deficiencies in MADD-related disorders: a new case and phenotypic expansion
2026-9-2 Vol. 35 Issue 4 Pages 170-176
Authors: Vykuntaraju K. Gowda, Varunvenkat M. Srinivasan, Archana Varghese et al.
DOI: 10.1097/mcd.0000000000000574

Primrose syndrome with a novel ZBTB20 variant with congenital glaucoma and hydrocephalus
2026-9-2 Vol. 35 Issue 4 Pages 183-184
Authors: Abhinav Thakral, Robert Wallerstein
DOI: 10.1097/mcd.0000000000000566

A case of serpentine fibula-polycystic kidney syndrome presenting with foetal limb shortening: a disorder of foetal osteogenesis and part of the phenotypic spectrum of Hajdu–Cheney syndrome
2026-9-2 Vol. 35 Issue 4 Pages 180-182
Authors: Yumi Ito, Naoki Ito, Yukiko Kuroda et al.
DOI: 10.1097/mcd.0000000000000568