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Methylation of the 4q35 D4Z4 repeat defines disease status in facioscapulohumeral muscular dystrophy
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Persistent hypokalaemia and intermittent muscle weakness
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O'Donnell-Luria-Rodan syndrome: description of a second multinational cohort and refinement of the phenotypic spectrum
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Identification of a novel homozygous synthesis of cytochrome c oxidase 2 variant in siblings with early-onset axonal Charcot-Marie-Tooth disease
Andrea Gangfuß, Andreas Hentschel, Nina Rademacher, Albert Sickmann, Burkhard Stüve, Rita Horvath, Claudia Gross, Nicolai Kohlschmidt, Fabian Förster, Angela Abicht, Anne Schänzer, Ulrike Schara-Schmidt, Andreas Roos, Adela Della Marina
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Sensory neuropathy due to RFC1 in a patient with ALS: more than a coincidence?
Florian Schoeberl, Angela Abicht, Clemens Kuepper , Stefanie Voelk , Stefan Sonnenfeld , Matthias Tonon , Annalisa Schaub, Veronika Scholz , Stephanie Kleinle , Hannes Erdmann , Dieter A Wolf , Peter Reilich
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Congenital myopathy and epidermolysis bullosa due to PLEC variant
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Cutaneous T-cell lymphoma mimicking myopathy with lipoatrophy
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Initial Clinical Experience with NIPT for Rare Autosomal Aneuploidies and Large Copy Number Variations
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Effect of Discontinuation of Nusinersen Treatment in Long-Standing SMA_3
Miriam Hiebeler, Angela Abicht, Peter Reilich, Maggie C Walter
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Sensory neuropathy due to RFC_1 in a patient with ALS: more than a coincidence?
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Location matters – Genotype – phenotype correlation in $LRSAM_1$ mutations associated with rare Charcot-Marie-Tooth neuropathy CMT_2P
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Next Generation Sequencing in Pediatric Epilepsy Using Customized Panels: Size Matters
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Congenital myopathy and epidermolysis bullosa due to PLEC variant
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