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Transcriptome-directed analysis for Mendelian disease diagnosis overcomes limitations of conventional genomic testing

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A relatively common homozygous TRAPPC4 splicing variant is associated with an early-infantile neurodegenerative syndrome

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De novo mutations in TOMM70, a receptor of the mitochondrial import translocase, cause neurological impairment

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Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7

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Limitations of Exome Sequencing in Detecting Rare and Undiagnosed Diseases

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Myopathy associated with homozygous PYROXD1 pathogenic variants detected by genome sequencing

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