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Yield of whole exome sequencing in undiagnosed patients facing insurance coverage barriers to genetic testing

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Pathogenic variants in USP7 cause a neurodevelopmental disorder with speech delays, altered behavior, and neurologic anomalies

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Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay

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A Recurrent De Novo Heterozygous COG4 Substitution Leads to Saul-Wilson Syndrome, Disrupted Vesicular Trafficking, and Altered Proteoglycan Glycosylation

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Clinical heterogeneity of mitochondrial NAD kinase deficiency caused by a NADK2 start loss variant

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Expanding the phenotypic spectrum of GABRG2 variants: a recurrent GABRG2 missense variant associated with a severe phenotype

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A window into living with an undiagnosed disease: illness narratives from the Undiagnosed Diseases Network

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