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Macrocephaly and developmental delay caused by missense variants in RAB5C

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HNRNPC haploinsufficiency affects alternative splicing of intellectual disability-associated genes and causes a neurodevelopmental disorder

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Monoallelic variation in DHX9, the gene encoding the DExH-box helicase DHX9, underlies neurodevelopment disorders and Charcot-Marie-Tooth disease

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A de novo missense variant in EZH1 associated with developmental delay exhibits functional deficits in Drosophila melanogaster

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De Novo Variants in MRTFB have gain of function activity in Drosophila and are associated with a novel neurodevelopmental phenotype with dysmorphic features

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A novel, de novo intronic variant in POGZ causes White-Sutton syndrome

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The clinical and molecular spectrum of QRICH1 associated neurodevelopmental disorder

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Finding commonalities in rare diseases through the Undiagnosed Diseases Network

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Mutations in GET4 Disrupt the Transmembrane Domain Recognition Complex Pathway

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Defining the clinical phenotype of Saul-Wilson syndrome

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Using MARRVEL v1.2 for Bioinformatics Analysis of Human Genes and Variant Pathogenicity

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