Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases

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Machine Learning of Patient Characteristics to Predict Admission Outcomes in the Undiagnosed Diseases Network

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De novo TRIM8 variants impair its protein localization to nuclear bodies and cause developmental delay, epilepsy, and focal segmental glomerulosclerosis

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DYRK1A pathogenic variants in two patients with syndromic intellectual disability and a review of the literature

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Novel NUDT2 variant causes intellectual disability and polyneuropathy

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

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Mutations in GET4 Disrupt the Transmembrane Domain Recognition Complex Pathway

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Homozygous splice-variants in human ARV1 cause GPI-anchor synthesis deficiency
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The Genome Empowerment Scale (GEmS): An Assessment of Parental Empowerment in Families with Undiagnosed Disease

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An integrated clinical program and crowdsourcing strategy for genomic sequencing and Mendelian disease gene discovery

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