Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases

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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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BICRA, a SWI/SNF Complex Member, Is Associated with BAF-Disorder Related Phenotypes in Humans and Model Organisms

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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
Myopathy associated with homozygous PYROXD1 pathogenic variants detected by genome sequencing

Defining the clinical phenotype of Saul-Wilson syndrome

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

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The Undiagnosed Diseases Network as a tool for graduate medical education

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ClinPhen extracts and prioritizes patient phenotypes directly from medical records to expedite genetic disease diagnosis

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