GENETICS

01

Prevalence of Genetically Triggered Aortopathy in Acute Aortic Syndrome in Aotearoa New Zealand.

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Rare de novo gain-of-function missense variants in DOT1L are associated with developmental delay and congenital anomalies.

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Severe neonatal onset neuroregression with paroxysmal dystonia and apnoea: Expanding the phenotypic and genotypic spectrum of CARS2-related mitochondrial disease.

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Germline mutations in WNK2 could be associated with serrated polyposis syndrome.

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GENETICS

06

A novel 11 base pair deletion in KMT2C resulting in Kleefstra syndrome 2.

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07

Biallelic ATP2B1 variants as a likely cause of a novel neurodevelopmental malformation syndrome with primary hypoparathyroidism.

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