

Video: Researchers discover rare genetic brain disorder

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An international team of researchers, led by MCRI, have identified a rare genetic brain disorder that causes severe neurological damage in children after a mild episode of fever or illness.

The [children](#) died soon after. The research has been published in the January edition of the journal, *Brain*. Only six cases have been found worldwide. Whole-exome or whole-genome sequencing identified recessive NAXD variants in each case. Lead researcher Nicole Van Bergen said that given the underlying genetic basis of the disorder is now understood, [future research](#) will investigate whether targeted treatments are possible.

More information: Nicole J Van Bergen et al. NAD(P)HX dehydratase (NAXD) deficiency: a novel neurodegenerative disorder exacerbated by febrile illnesses, *Brain* (2018). [DOI: 10.1093/brain/awy310](#)

Provided by Murdoch Children's Research Institute

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