World’s first patient-initiated, crowdfunded genome sequencing project uncovers young child’s unknown genetic disease

The Rare Genomics Institute (RGI), a non-profit organization that uses genome sequencing and other biotechnology to help children with rare genetic diseases, and an Ivy League medical research center have jointly identified a new gene variant in four-year-old Maya Neider that may indicate a brand-new disease. This finding marks the first time that a patient-initiated, crowdfunded genome project has uncovered the genetic basis of a rare disease.

Suffering from global developmental delays, Maya has undergone multiple operations, is unable to speak, and has difficulty hearing. Despite visiting countless physicians, her condition had remained unexplained for years. Doctors agreed that “something genetic” was responsible for her condition, yet six genetic tests – each screening for a myriad of known genetic defects – yielded no definitive explanation.

View the original article here: World’s First Crowdfunded Genome Sequencing Project Uncovers 4-Year-Old’s Unknown Genetic Disease