Can we identify the genes responsible for developmental delay in children?

Developmental delay affects 1-2% of children worldwide. Symptoms often associated with developmental delay include impaired cognition, failure to meet developmental milestones, craniofacial and skeletal abnormalities, autism, and seizures.

Specific diagnoses for children with developmental delay are in many cases elusive, and the lack of a diagnosis is a major hardship for patients and their families.

In an effort to end the diagnostic odyssey for children with developmental delay, we have employed large-scale DNA sequencing to identify specific genetic variants that are causally relevant to developmental disabilities. As part of the NHGRI-funded Clinical Sequencing Exploratory Research Consortium, we began enrolling affected children into our study in 2013. Thus far, we have sequenced 371 children who present with developmental delay, and we have found the genetic cause – and thus contributed to more precise and definitive clinical diagnoses – in 27%.

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Through our study, we observed that finding a pathogenic variant in an affected child is more challenging when close relatives have a neurological condition. This finding suggests that the underlying genetics in such “multiplex” families are more complex and harder to interpret than in “simplex” families, and that this distinction influences the success rate in terms of pathogenic variant discovery.

[Read the full study here]

The GLP aggregated and excerpted this blog/article to reflect the diversity of news, opinion, and analysis. Read full, original post: Finding the genetic causes of developmental delay