Genome-wide diagnostic testing is now widely implemented in clinical practice to identify the underlying cause of neurodevelopmental disorders. Diagnostic yields are increasing, meaning that the number of individuals and families receiving a genetic diagnosis will climb over the next few years. The challenge now is to make genetic diagnosis more useful for affected individuals and their families. To achieve this, we need a much better understanding of the links between genetic cause and an individual's symptoms and impairments. One approach is to group genetic diagnoses into functional networks converging on molecular and cellular processes such as chromatin regulation, ion channels / excitability and synaptic signalling. Functional networks have distinct cellular, spatial and temporal expression patterns, suggesting that they may impact on different neurodevelopmental mechanisms. We want to know whether, how and why functional networks differentially influence cognitive development and mental health. Ultimately, the goal is to provide evidence-based prognostic information for each person after genetic diagnosis, and translate this knowledge into personalised mechanism-informed therapies which can improve specific symptom dimensions of value to each person. This project will involve big data (for example, the 100,000 Genomes Project, IMAGINE-ID, Simons Searchlight and SPARK) and computational analyses integrating genomic, cellular and phenotypic datasets.

References and URL(s)

URL
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URL
https://doi.org/10.1111/jnc.15135