

Genetic counselling and genomic testing for neurological conditions

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ABSTRACT

Genetic diseases and congenital malformations are one of the main causes of NCDs and main cause of mortality and morbidity. Recent advances in technology and easier access to information have resulted in greater understanding of genomic science and its application in neurological conditions. Issues related to the clinical utility, cost-effectiveness and ethical concerns of these diagnostic tools had been raised. All processes within a cell are regulated by an interacting network of multiple genes. Each healthy individual carries several faulty genes in the cells. A disorder due to a variation in a gene or copy number change in the chromosome is often suspected in a patient with learning difficulties and dysmorphism. Others are due to mitochondrial variations, polygenic or multifactorial inheritance where the environment plays a role. A genetic study scrutinizes the functioning and composition of the single gene while a genomic study addresses all or a large number of genes and their inter-relationships (the "network") in order to identify their combined influence on the growth and development of the organism. Variants are passed on to the offspring and become 'familial'. A variant is considered as 'pathological' when the change leads to disrupted information causing diseases. Some of the ethical concerns regarding genetic or genomic testing are tests involving asymptomatic children, genetic testing without genetic counselling, the lack of equitable access to genetic services and support and high costs of the tests. Other issues such as lack of knowledge by healthcare providers to interpret the pathogenicity of the variants, incidental findings, variants of uncertain significance, cascade screening and the lack of appreciation of the methodology of mutation detection techniques are also major concerns. Hence, understanding the challenges and opportunities presented by genomic testing must be understood as genetic diseases and genetic susceptibility have become the main cause of NCDs.

Keywords: Genetic Diseases are NCDs, Genomic Testing, Genetic Counselling, Ethics