GENETICS AND NEUROPATHY

Basics of Genetics

Each cell in our body contains DNA, which provides instructions for our body to generate the proteins necessary for daily life. DNA is made up of tens of thousands of genes each of which codes for a specific protein. When there are changes in genes, referred to as mutations or variants, genetic diseases or predispositions can occur. Health conditions may also be multifactorial in nature, meaning that they arise due to a combination of environmental factors in addition to multiple genes. Current genetic testing technology is able to evaluate for known monogenic (caused by a change in a single gene) conditions, but typically cannot evaluate for multifactorial causes of disease.

Underlying Genetics of Neuropathy

The underlying causes of neuropathy can be varied including: prior infection, autoimmune conditions, idiopathic, and hereditary genetic conditions. It is estimated 0.04% of the population has a hereditary peripheral neuropathy (Eggermann et al., 2018). Comparatively 2.4% of the population is affected by peripheral neuropathy and this number increases to 8% in older populations (Hammi & Yeung, 2026). Genetic forms of neuropathy may be isolated or part of a broader syndrome. Specific features that may be seen in those with isolated hereditary neuropathies include: foot deformities, thin calf muscles, and scoliosis (Hereditary Neuropathies | National Institute of Neurological Disorders and Stroke). If an individual has other health conditions or congenital abnormalities in conjunction with neuropathy, that increases the suspicion of a syndromic cause. Other factors that may prompt a search for a suspected genetic cause include a young age of diagnosis or a strong family history of neuropathy.

There are more than 100 genes that have been associated with isolated hereditary neuropathy documented within the medical literature. Common isolated hereditary neuropathies include Charcot-Marie-Tooth disease (CMT), hereditary sensory and autonomic neuropathy, hereditary motor neuropathy, and small fiber neuropathy (SFN). It can be difficult to differentiate between types of hereditary neuropathies based solely on an individual's symptoms, thus genetic testing may be used to clarify a diagnosis. Current literature recommends that those with a suspected hereditary neuropathy should undergo a thorough diagnostic evaluation to rule out metabolic, nutritive-toxic, infectious, and inflammatory/autoimmune causes (Eggermann et al., 2018).

What Can Genetic Testing Tell Us?

Individuals may undergo genetic testing to identify a possible hereditary neuropathy because that may alter their medical management. Determining if there is a monogenic cause for a person's neuropathy can also inform the risk of disease in their family members. Having a positive result on testing may prompt additional testing for family members. Finally genetic testing can provide a catharsis to patients by ending the journey to identify the underlying trigger for their neuropathy.

How to Access Testing

For those interested in clinical genetic testing, a meeting with a genetic counselor is recommended. A referral can be placed by one's individual doctor or they can connect with a local genetic counselor via https://findageneticcounselor.nsgc.org.

References

Eggermann, K., Gess, B., Häusler, M., Weis, J., Hahn, A., & Kurth, I. (2018). Hereditary Neuropathies. Deutsches Ärzteblatt International, 115(6), 91–97. https://doi.org/10.3238/arztebl.2018.0091 Hammi, C., & Yeung, B. (2026). Neuropathy. In StatPearls. StatPearls Publishing. http://www.ncbi.nlm.nih.gov/books/NBK542220/

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