TREATMENT AND DETECTION OF INHERITED NEUROPATHIES AND ASSOCIATED DISORDERS

ABSTRACT

The present disclosure relates to methods of detecting and treating inherited neuropathy. In various aspects, the method comprises detecting the presence of a mutation in the sorbitol dehydrogenase (SORD) gene in a sample from a subject. In various embodiments, th SORD mutation is a DNA variant classified as pathogenic or likely pathogenic according to American College of Medical Genetics and Genomics (ACMG) criteria.

FIG. 1A