Genetics Uncoded:

Facts about Mucolipidosis IV

What Your Test Results Mean

Carriers typically show no symptoms of Mucolipidosis IV (ML IV); however, carriers are at an increased risk of having a child with ML IV. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

MSUD Explained

ML IV is an inherited metabolic disorder in which harmful amounts of fats and proteins accumulate within the lysosomes, or recycling compartments, of cells in the body. Individuals with ML IV do not produce enough of one of the proteins, mucolipin-1, needed to properly dispose of fats and proteins in the body. Over time, this excessive build-up of fats and lipids within the lysosomes causes developmental delay, visual impairment, and other signs of the disease.

There are two forms of ML IV. In the most common form, typical ML IV, developmental delays are noted in the first year of life. Maximal developmental level achieved is typically 18 months of age. Neurodegeneration is seen in approximately 15% of individuals with typical ML IV. By the teens, severe vision loss or blindness caused by corneal clouding or progressive retinal degeneration is present in most individuals. Additional characteristics of the disease include impaired stomach acid production causing elevated gastrin in the blood, as well as iron deficiency that can lead to anemia. Individuals with the more mild form, atypical ML IV, have milder delays and eye abnormalities than those with typical ML IV.

Treatment of individuals with ML IV typically includes supportive therapies and iron supplementation for iron deficiency anemia. Life expectancy may be shortened, but individuals with ML IV typically live into adulthood

How the Genetics Work

The clinical features of ML IV can be explained by pathogenic variants in the *MCOLN1* gene. In general, individuals have two copies of the *MCOLN1* gene. Carriers of ML IV have a single variant in one copy of the *MCOLN1* gene while individuals with ML IV have variants in both copies of their genes, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

Questions? Contact us at **1-855-776-9436** to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.

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Revised 12/15/16