Zellweger Spectrum Disorder, *PEX1*-Related

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of Zellweger spectrum disorder, *PEX1*-related. Carriers typically show no symptoms. Risk for current or future pregnancies is dependent on your partner's carrier status. Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for more detailed risk assessment.

Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning and their own personal clinical management.

Recommended Next Steps

Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment. If both you and your partner or donor are carriers for Zellweger spectrum disorder, *PEX1*-related, each of your children has a 1 in 4 (25%) chance to have the condition.

Zellweger Spectrum Disorder, *PEX1*-Related Explained

What is Zellweger Spectrum Disorder, *PEX1*-Related?

Zellweger spectrum disorder, *PEX1*-related is an inherited condition that prevents the body from properly breaking down toxins and fatty acids. As indicated by the word "spectrum," people with a Zellweger spectrum disorder vary widely in the type and severity of their symptoms. The disease is generally grouped into three subtypes: Zellweger syndrome (the most severe), neonatal adrenoleukodystrophy (intermediate severity), and infantile Refsum disease (the mildest form). Infants with Zellweger syndrome present with poor muscle tone, feeding difficulties, developmental delay, mental retardation, seizures, distinctive facial features, and hearing and vision loss. They may also develop life-threatening complications related to liver and kidney disease. Individuals with neonatal adrenoleukodystrophy and infantile Refsum disease have similar but more slowly progressive features, including weak muscle tone, developmental delay, intellectual disability, vision problems, hearing loss, and liver and kidney disease.

Prognosis

Prognosis is generally unfavorable, though age of onset, severity, progression of symptoms, and life expectancy vary depending on the type of the Zellweger spectrum disorder. Children with Zellweger syndrome are diagnosed in infancy and have developmental delays, mental retardation, vision loss, hearing loss, seizures, heart disease, liver disease and kidney disease. Affected individuals typically do not survive beyond a year of life.

Children with neonatal adrenoleukodystrophy and infantile Refsum disease present in late infancy or early child and have a more variable and slowly progressive clinical course. However, they too can be affected with developmental delays, intellectual disability, hearing loss, visual impairment, liver disease, low muscle tone and episodes of hemorrhage or intracranial bleeding. Typically, individuals with neonatal adrenoleukodystrophy live into childhood and those with infantile Refsum disease may survive into adulthood with moderate to severe intellectual and physical impairments.

801 Broadway Ave NW, Suite 203, Grand Rapids, MI 49504 • Phone: (855) 776-9436 • Fax: (616) 710-4667 info@nxgenmdx.com
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Treatment

There is no cure for Zellweger spectrum disorders. Treatment focuses on management of symptoms and includes the use of a feeding tube to ensure adequate caloric intake, hearing aids to help with hearing loss, glasses and cataract removal in infancy to help with vision, medication to prevent seizures, and vitamin supplementation and primary bile acid therapy to improve liver function.

Resources

Global Foundation for Peroxisomal Disorders (GFPD)
https://www.thegfpd.org/
National Institute of Neurological Disorders and Stroke
https://www.ninds.nih.gov/Disorders/all-disorders
United Leukodystrophy Foundation
http://ulf.org/the-zellweger-spectrum/
National Society of Genetic Counselors
https://www.nsgc.org/

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