



# Facts about

## *PEX1*-Related Zellweger Syndrome Spectrum



### What Your Test Results Mean

**Carriers typically show no symptoms of *PEX1*-related Zellweger syndrome spectrum (ZSS); however, carriers are at an increased risk of having a child with ZSS.** Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

### ● ***PEX1*-Related Zellweger Syndrome Spectrum Explained**

ZSS is an inherited condition that prevents the body from properly breaking down toxins and fatty acids. As a result, these substances accumulate in the body's tissues and cause severe damage. The most severe form of ZSS is called Zellweger syndrome; infants with this disease have very poor muscle tone, skeletal abnormalities, mental retardation, feeding problems, hearing and vision loss, and seizures. These infants experience failure of multiple organs and typically do not survive for more than one year.

The milder forms of ZSS are neonatal adrenoleukodystrophy (NALD) and infantile Refsum disease. These forms are usually not evident until late infancy or early childhood and slowly develop many of the symptoms seen in individuals with Zellweger syndrome. Children with these less severe conditions often have hypotonia, vision problems, hearing loss, liver dysfunction, developmental delay, and some degree of intellectual disability. Individuals with NALD generally survive into childhood, while those with infantile Refsum disease may even reach adulthood. There is no cure for ZSS; treatment is symptomatic.

### ● **How the Genetics Work**

ZSS is an autosomal recessive disorder caused by variants in the *PEX1* gene. In general, individuals have two copies of the *PEX1* gene. Carriers of ZSS have a single variant in one copy of the *PEX1* gene while individuals with ZSS 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 **1-866-661-7966** to set up an appointment to discuss your results in more detail with a Clarity Genetics genetic counselor.