Zellweger Spectrum Disorder (PEX1-Related)

ZSD is a group of rare genetic disorders. The form of ZSD covered by this report is characterized by impaired hearing, vision, and organ function, as well as developmental disability and early death. A person must have two variants in the PEX1 gene in order to have this form of ZSD.

Overview Scientific Details

Jamie, you **have the variant** we tested.

You could pass this variant on to your children.

How To Use This Test

This test does not diagnose any health conditions.

Please talk to a healthcare professional if this condition runs in your family, you think you might have this condition, or you have any concerns about your results.

Review the Carrier Status tutorial
See Scientific Details

Intended Uses

- To test for the G843D variant in the PEX1 gene.
- To identify carrier status for ZSD.

Limitations

- Does **not test** for all possible variants for the condition.
- Does **not report** if someone has two copies of a tested variant.
- Does **not cover** ZSD caused by variants in other PEX genes.

Important Ethnicities

- This test does **not** include the majority of PEX1 variants that cause ZSD in any ethnicity.

You are a carrier.

You could pass this variant on to your children.

We detected one variant for ZSD.

People with only one variant are not expected to have ZSD.

Your results may be relevant for you if you're thinking about starting a family.

If you and your partner are both carriers, each child may have a **25% chance** of having this condition. Your relatives may also wish to consider testing if they plan to have children.

About Zellweger Spectrum Disorder (PEX1-Related)

ZSD is a continuum of rare genetic disorders that can present as severe, intermediate, or mild. It is a type of peroxisome biogenesis disorder.

When symptoms develop

Symptoms are typically present at birth or develop during infancy.

Typical signs and symptoms

- Decreased muscle tone
- Seizures
- Failure to gain weight
- Impaired vision and hearing
- Developmental disability
- Early death (severe form)

Ethnicities most affected

This condition affects people of all ethnicities.

How it's treated

There is currently no known cure. Treatment focuses on managing symptoms and preventing complications.

Read more at: MedlinePlus GeneReviews National Organization for Rare Disorders

Consider talking to a healthcare professional if you are thinking about having children.

If you're starting a family, a genetic counselor can help you and your partner understand if additional testing might be appropriate.

Connect with a GC

Share your results with your family.

Share your report

If you have other concerns about your results, consult with a healthcare professional.

Print report

Learn more about this condition and connect with support groups.

Learn more