Metachromatic Leukodystrophy, *ARSA*-Related

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of metachromatic leukodystrophy, *ARSA*-related. Carriers are not expected to show symptoms. You and your partner would both have to be carriers of metachromatic leukodystrophy, *ARSA*-related for there to be an increased chance to have a child with symptoms; this is known as autosomal recessive inheritance. Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a 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.

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 are carriers for metachromatic leukodystrophy, *ARSA*-related, each of your children has a 1 in 4 (25%) chance to have the condition.

Metachromatic Leukodystrophy, *ARSA*-Related Explained

What is Metachromatic Leukodystrophy, *ARSA*-Related?

Metachromatic leukodystrophy, *ARSA*-related is an inherited condition caused by lack of the enzyme arylsulfatase A. Without this enzyme, harmful substances build up in parts of the body, including the brain. The disorder is characterized by progressive deterioration of intellectual functions and motor skills, such as the ability to walk. Affected individuals also develop psychiatric symptoms such as delusions or hallucinations, loss of sensation in the extremities, incontinence, an inability to speak, blindness, hearing loss, seizures, and paralysis, eventually leading to death.

There are three types of metachromatic leukodystrophy, *ARSA*-related based on the age of onset: the infantile form, which appears at one to two years of age and is fatal by age ten; the juvenile form, which appears after age three but before adolescence and is fatal 10-20 years after onset; and the adult form, which can appear any time after puberty.

Prognosis

Prognosis is generally poor as all individuals eventually experience the complete loss of their motor and intellectual functions.

Treatment

Researchers are currently searching for an effective treatment for metachromatic leukodystrophy, *ARSA*-related, and experimental treatments include cord blood transplantation and gene therapy. Until a more effective treatment is found, management is supportive.

Resources

United Leukodystrophy Foundation
https://ulf.org/metachromatic-leukodystrophy-mld/
National Organization for Rare Disorders
https://rarediseases.org/rare-diseases/metachromatic-leukodystrophy/
National Society of Genetic Counselors
https://www.nsgc.org/

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