Alkaptonuria

What Your Results Mean

Test results indicate that you are a carrier of alkaptonuria. Carriers are not expected to show symptoms. You and your partner or donor would both have to be carriers of alkaptonuria 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 or donor are carriers for alkaptonuria, each of your children has a 1 in 4 (25%) chance to have the condition.

Alkaptonuria Explained

What is Alkaptonuria?

Alkaptonuria is an inherited disorder caused by an enzyme deficiency that results in a build-up of homogentisic acid in the body. Over time, this build-up causes the connective tissues, such as the skin and cartilage, to darken. In individuals with alkaptonuria, homogentisic acid is also excreted into the urine, which then turns black upon immediate exposure to air. Arthritis occurring in the larger joints and the spine is also seen with alkaptonuria. Additional signs and symptoms include heart problems, kidney stones, and prostate stones.

Prognosis

Alkaptonuria is not known to decrease the lifespan of affected individuals, nor does it cause any developmental delays. However, quality of life can be affected as issues with mobility and joint pain can develop.

Treatment

Treatment of alkaptonuria is symptomatic and tailored to the individual. Medications such as anti-inflammatories can be beneficial in treating joint pain. Individuals may additionally benefit from physical and occupational therapy. Some individuals may also require surgical intervention to replace certain joints by 50-60 years of age.

Resources

Alkaptonuria Society
https://www.akusociety.org/
National Organization for Rare Disorders (NORD)
https://rarediseases.org/rare-diseases/alkaptonuria/
National Society of Genetic Counselors
https://www.nsgc.org/

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Revised: 4/2022