Genetics Uncoded:

Facts about Alkaptonuria

What Your Test Results Mean

Carriers show no symptoms of this disorder and are not at risk to develop symptoms of the disorder. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Alkaptonuria Explained

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 to darken. In individuals with alkaptonuria, homogentisic acid is also excreted in urine, which turns the urine black upon immediate exposure to air. This condition can also cause arthritis, heart problems, kidney stones, and prostate stones. Management is symptomatic and there are no treatments available; however, alkaptonuria is not known to decrease the lifespan of affected individuals.

How the Genetics Work

Alkaptonuria is an autosomal recessive disorder caused by pathogenic variants in the *HGD* gene. In general, individuals have two copies of the *HGD* gene. Carriers of alkaptonuria have a single pathogenic variant in one copy of the *HGD* gene while individuals with alkaptonuria 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 **1-855-776-9436** to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.

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