Genetics Uncoded:

Facts about

Autosomal Recessive Alport Syndrome

What Your Test Results Mean

Carriers typically show no symptoms of Alport syndrome. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Autosomal Recessive Alport Syndrome Explained

Autosomal recessive Alport syndrome is an inherited disorder caused by abnormal type IV collagen production. It is characterized by kidney disease, sensorineural hearing loss, and sometimes eye abnormalities. Blood in the urine (hematuria) and high levels of protein in the urine are typically the first signs of the disorder followed by hearing loss. Kidney failure may occur in the teenage years, but may not develop until as late as 40-50 years of age. Kidney dialysis or transplantation is available for those with end-stage renal disease.

How the Genetics Work

Alport syndrome is an autosomal recessive disorder caused by variants in the *COL4A4* gene. In general, individuals have two copies of *COL4A4*. Carriers of *COL4A4*-associated Alport syndrome have a variant in one copy *COL4A4* while individuals with the disorder have variants in both copies the *COL4A4* gene, 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.

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