Genetics Uncoded:

Facts about

Autosomal Recessive Polycystic Kidney Disease

What Your Test Results Mean

Carriers typically show no symptoms of autosomal recessive polycystic kidney disease (ARPKD). Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Autosomal Recessive Polycystic Kidney Disease Explained

ARPKD is an inherited condition of early-onset liver and kidney problems. Symptoms include polycystic kidneys, high blood pressure, abdominal pain, recurrent urinary tract infections, and liver disease. Approximately 10% of children surviving the neonatal period will require liver transplantation. Over 50% will develop end stage renal disease in the first decade of life. Dual liver and kidney transplantation has proven successful.

How the Genetics Work

ARPKD is an autosomal recessive disorder that can be explained by variants in the *PKHD1* gene. In general, individuals have two copies of the *PKHD1* gene. Carriers of ARPKD have a single variant in one copy of the *PKHD1* gene while individuals with ARPKD 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 **1-866-661-7966** to set up an appointment to discuss your results in more detail with a Clarity Genetics genetic counselor.

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