What Your Test Results Mean

Testing results indicate that you are a carrier of autosomal recessive polycystic kidney disease (ARPKD). Carriers typically show no symptoms of ARPKD; however, carriers are at an increased risk of having a child with ARPKD. Risk for the current or future pregnancies is dependent on your partner’s carrier status. Carrier testing of your partner is recommended in addition to consultation with a genetic counselor.

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 mutations in the PKHD1 gene. In general, individuals have two copies of the PKHD1 gene. Carriers of ARPKD have a single mutation in one copy of the PKHD1 gene while individuals with ARPKD have mutations in both copies of their genes, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

Recommended Next Steps

Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for more detailed risk assessment.

Questions?

Contact us at (855) 776-9436 to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.