Polycystic Kidney Disease, Autosomal Recessive

NxGen MDx

What Your Results Mean

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

Polycystic Kidney Disease, Autosomal Recessive Explained

What is Polycystic Kidney Disease, Autosomal Recessive?

ARPKD is a severe condition that affects the kidneys and occasionally the liver. Affected individuals are often born with many cysts (fluid-filled sacs) in the kidneys, which impairs their ability to perform their normal functions. The cysts can also affect other organs, particularly the liver. In most affected individuals, symptoms are apparent soon after birth. Symptoms include difficulty breathing, enlarged cyst-filled kidneys, enlarged liver, high blood pressure, low blood cell counts, frequent urination, and pain in the back or sides. Rarely, affected individuals may not show symptoms until childhood or early adulthood. In these individuals, liver disease is more severe and kidney disease is often mild.

Prognosis

Prognosis is generally unfavorable, though survival rates have increased in recent years. Approximately 30% of affected infants die within the first year of life due to breathing difficulties. More than 50% of affected children develop kidney failure by age 10. With respiratory support and kidney replacement for newborns, the ten-year survival of those who live beyond the first year of life has improved to 82%. Fifteen-year survival is estimated to be 67-79% and may be improving.

Treatment

Treatment for newborns focuses on stabilization of respiratory function by mechanical ventilation. Sometimes dialysis or surgery on the kidneys may be necessary in infancy. Once kidneys begin to fail, regular dialysis is required. Kidney transplantation may also be appropriate. Medications are administered to control high blood pressure. Liver transplantation is increasingly considered as a viable option for those with severe high blood pressure.

Resources

PKD Foundation
https://pkdcure.org/

National Organization for Rare Disorders (NORD)
https://rarediseases.org/rare-diseases/autosomal-recessive-polycystic-kidney-disease/

National Society of Genetic Counselors
https://www.nsgc.org/

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