GENE Seeker

You know the risk of your child being a carrier of a genetic disease.

Autosomal recessive polycystic kidney disease

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What is Autosomal recessive polycystic kidney disease?

A rare, genetic hepatorenal fibrocystic syndrome characterized by cystic dilatation and ectasia of renal collecting tubules, and a ductal plate malformation of the liver resulting in congenital hepatic fibrosis. Clinical presentation, whilst typically in utero or at birth, is variable and in the most severe cases includes Potter-sequence, oligohydramnios, pulmonary hypoplasia, and massively enlarged echogenic kidneys.

The clinical spectrum is broad and may include variable degrees of renal insufficiency, mild to severe life-threating neonatal respiratory distress/failure due to pulmonary hypoplasia, hyponatremia, hypertension and predisposition to urinary tract infections. Patients can progress to end-stage renal disease (ESRD) at varying ages. Congenital hepatic fibrosis (CHF) is invariably presents at birth, although may be clinically undetectable. The progressive manifestations of CHF typically include portal hypertension (pHTN), gastrointestinal varices and associated bleeding, bile duct disease (Caroli syndrome and cholangitis) and hepatosplenomegaly.

What is the next step if I'm a carrier of Autosomal recessive polycystic kidney disease?

If you are found to be a carrier of Autosomal recessive polycystic kidney disease, it is important that your partner be tested for the same genetic disorder.

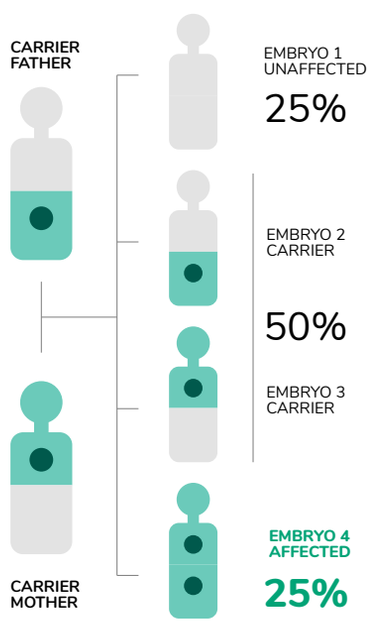
What if my partner is not a carrier?

If your partner's test for Autosomal recessive polycystic kidney disease is negative, the chance to have an affected child is low. However there is currently no test able to detect all existing mutations, so there is always a residual risk that the person who has done the test is a carrier of other less frequent mutations.

What if both me and my partner are carriers of Autosomal recessive polycystic kidney disease?

When both parents are carriers of Autosomal recessive polycystic kidney disease the probability of having a child with Autosomal recessive polycystic kidney disease is 25%.

We recommend that you discuss your results with your doctor or genetic counselor in order to know more about reproductive options.



If both you and your partner are carriers, speak with your doctor or genetic counselor about reproductive options.

1 frequency

70 people

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