GENE Seeker

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

Wilson's disease

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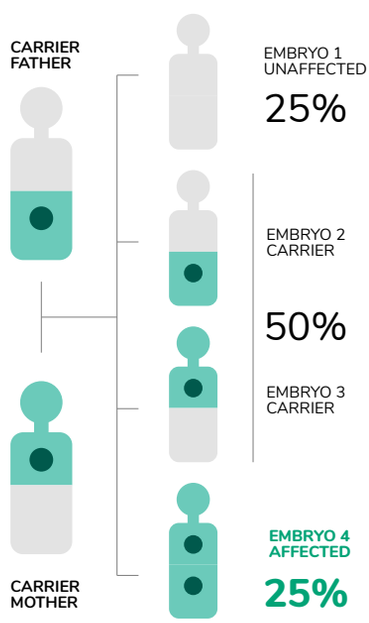
What is Wilson's disease?

A rare genetic disorder of copper metabolism presenting with non-specific hepatic, neurologic, psychiatric or ophthalmologic manifestations due to impaired biliary copper excretion and consecutive excessive copper deposition in the body.

The clinical spectrum is very wide, even within affected families. Likewise, age of onset is highly variable. Some patients remain asymptomatic for decades while few present symptoms before age 3 to 5. The disease can be observed in children after 3 years and most cases develop by 40 years of age. Late-onset cases after the fifth decade of life have also been described. Clinical presentation depends on gender and age. In children, at an average age of 10 years, hepatic manifestations typically prevail, most often commencing with liver damage. In general, hepatic manifestations (hepatomegaly, subacute or chronic hepatitis, acute liver failure or cirrhosis with portal hypertension) commonly precedes neurologic symptoms. Neurological manifestations (dystonia, intention tremor, dysarthria, coordination difficulties, chorea, choreoathetosis, and gait disorders) can be found in conjunction with hepatic symptoms or might also be the first clinical symptoms. Isolated psychiatric disorders (depression, phobias, compulsive behavior, personality changes, aggressiveness, or emotional instability) are rare and more commonly observed in conjunction with hepatic or neurologic disease. A wide range of other manifestations may also be present in affected patients: acute hemolytic episodes, delayed puberty, amenorrhea, repetitive miscarriage, Kayser-Fleischer rings due to copper deposits in the Descemet membrane, bone pain, arthralgia and osteoporosis, arrythmia, myocardiopathy, hematuria, nephrotic syndrome and renal lithiasis. Hepatocellular carcinoma has been reported in rare cases.



What is the next step if I'm a carrier of Wilson's disease?

If you are found to be a carrier of Wilson's 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 Wilson's disease 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 Wilson's disease?

When both parents are carriers of Wilson's disease, the probability of having a child with Wilson's 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.

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