GENE Seeker

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

Medium chain acyl-coenzyme A dehydrogenase deficiency

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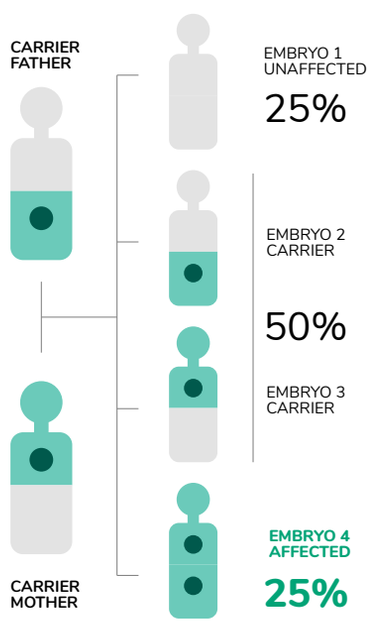
What is Medium chain acyl-coenzyme A dehydrogenase deficiency?

Medium chain acyl-CoA dehydrogenase (MCAD) deficiency (MCADD) is an inborn error of mitochondrial fatty acid oxidation characterized by a rapidly progressive metabolic crisis, often presenting as hypoketotic hypoglycemia, lethargy, vomiting, seizures and coma, which can be fatal in the absence of emergency medical intervention.

MCADD usually presents 3 to 24 months after birth in previously healthy infants. However, neonatal presentations are well described as are those in adults, given sufficient metabolic stress (such as significant alcohol ingestion). Nevertheless, many affected individuals remain asymptomatic throughout life. Typically hypoketotic hypoglycemia, lethargy and vomiting are triggered by an infection, fasting or surgery. Some patients, however, can present with a progressive metabolic crisis despite ketosis and normal blood glucose. Rarely patients may present in crisis with "paradoxically" gross ketosis. During a crisis, a patient may manifest with lethargy, emesis, respiratory arrest, seizures, hepatomegaly and rapid progression to cardiac arrest unless emergency treatment is implemented. Potential brain injury occurring during these episodes can lead to an increased risk of long term neurological damage. Sudden unexplained death can sometimes be the first manifestation of this disease. Historically, about 25% of undiagnosed patients die during their first presentation of a crisis.



What is the next step if I'm a carrier of Medium chain acyl-coenzyme A dehydrogenase deficiency?

If you are found to be a carrier of Medium chain acyl-coenzyme A dehydrogenase deficiency, 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 Medium chain acyl-coenzyme A dehydrogenase deficiency, 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 Phenylketonuria?

When both parents are carriers ofMedium chain acyl-coenzyme A dehydrogenase deficiency, the probability of having a child with Medium chain acyl-coenzyme A dehydrogenase deficiency 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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