

Commonly asked questions for CPT I deficiency

1. What is CPT I deficiency?

CPT I deficiency, also known as carnitine palmitoyltransferase I deficiency, is a fatty acid oxidation disorder (FAOD). It is a defect in one of the enzymes involved in the deployment of fats to fuel that can be used by the body. It becomes very important when the body is low on glucose or needs additional fuel such as when the child has not eaten for a period of time, during infections and other illnesses, during operations and when exercising vigorously.

2. How and when will we know if my baby has CPT I deficiency?

If your baby's newborn screening result showed a high free carnitine and an elevated carnitine ratio he or she probably has CPT I deficiency. If the ratio was between 100-175 your baby either could still have CPT I deficiency or the result may have been false positive. The newborn screening test will be repeated and additional tests will be undertaken to help determine if your baby has CPT I deficiency or not. Typically the results of these tests take up to 4 days to come back. Depending on the test results, additional testing can take a variable amount of time to confirm the diagnosis. In a very small minority of cases, it can be difficult to determine whether a child is affected or not.

3. How did my baby get this?

CPT I deficiency is an autosomal recessive disorder. This means that your baby has two mutated CPT1 genes, one from the mother and one from the father. Having only one mutated CPT I gene (a carrier) does not affect a person at all.

4. What does it mean for my child?

If your baby has CPT I deficiency, he or she will have to be fed regularly on a carbohydrate rich fat modified/decreased diet and can not be allowed to miss a meal. Medium chain triglyceride supplementation provides fat energy past the enzyme block. Carnitine should not be provided as, unlike all the other fatty acid oxidation defects, carnitine is elevated in CPT I deficiency. If he or she becomes ill, it may well be necessary early in the illness (i.e. when it might be considered mild) to provide extra energy in the form of glucose through addition to food or, if necessary, by intravenous drip.

5. What is the treatment? Does it work? Is the diet difficult to do/expensive?

CPT I deficiency is primarily treated by a high carbohydrate and fat modified/decreased diet that is given at regular defined intervals around the clock. As the diet is essentially normal it should not be a financial burden. However, ensuring that you and the baby wake up, initially every 4 hours, can be physically exhausting over time. If possible you should anticipate this and try and ensure that you have support from your spouse or other close contacts to assist you so that you may enjoy your time with your baby.

6. What about my other children/future children?

As CPT I deficiency is an inherited condition it is essential to have your other children tested. Children from the same father and mother as the affected infant have a 1 in 4 (25%) chance of having CPT I deficiency. Your other children can appear healthy and still have CPT I deficiency. If they have CPT I deficiency, successfully having weathered illnesses in the past is no guarantee that an illness in the future will not have serious consequences.

Since there is a risk for having a future child with CPT I deficiency it is important to let your obstetrician and pediatrician know that you have a child with CPT I deficiency if you are planning future pregnancies so that they may discuss the options with you and prepare accordingly. The obstetrician should furthermore be alerted to the association between CPT I deficiency and the HELLP and AFLP syndromes to the mother during pregnancy (see "History and Examination section" above).