

Commonly asked questions

1. What is VLCADD?

VLCADD, also known as Very Long Chain Acyl-CoA Dehydrogenase Deficiency, is a fatty acid oxidation disorder (FAOD). It is a defect in one of the enzymes responsible for converting 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, or when exercising vigorously.

2. How and when will we know if my baby has VLCADD?

If your baby's newborn screening result showed a markedly elevated C14 & C14:1 levels, he or she probably has VLCADD. If the result was less marked your baby either could still have VLCADD or it may have been a false positive result. The newborn screening test will be repeated and additional tests will be undertaken to help determine whether or not your baby has VLCADD. 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 or not a child is affected.

3. How did my baby get this?

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

4. What does it mean for my child?

If your baby has VLCADD, he or she will have to be fed regularly on a fat modified diet and cannot be allowed to miss a meal. Some children also take carnitine, a mild supplemental medicine, but your metabolic physician will be able to let you know if this is appropriate for your child. 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?

VLCADD is primarily treated by a high carbohydrate and fat modified diet that is given at regular defined intervals around the clock. As the diet is essentially normal it should not be a major additional 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 VLCADD 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 VLCADD. Your other children can appear healthy and still have VLCADD. If they have VLCADD, 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 VLCADD it is important to let your obstetrician and pediatrician know that you have a child with VLCADD if you are planning future pregnancies so that they may discuss the options with you and prepare accordingly.