Information for Parents and Carers

CARNITINE TRANSPORTER DEFECT

Why are babies screened for carnitine transporter defect?
There are a number of disorders that can have good outcomes if treated early. Further investigations are needed to determine if there is a problem that needs treatment. Because the newborn screening sample is collected very early in life, it is possible for the baby’s blood to reflect a problem in the mother.

What is carnitine transporter defect?
This is a very rare disorder. Carnitine is partly made in the body, and partly provided by the food we eat, such as red meat and dairy products. It is needed for the transport of fats into the cells of the body. We need carnitine to help us get energy from the stores of fat we have in the body. Babies and children with this defect may get low blood sugar if they fast and later may get heart muscle problems (cardiomyopathy). Adults with the disorder can also develop cardiomyopathy.

Why is this a problem?
Energy is stored in the body either as a sugar substance called glycogen or as fat. When we need energy we use up the glycogen stores first, after which we need to access the fat stores. Children with a defect in the carnitine transporter cannot get dietary fats into the cells where they can be converted to energy. This is why they may suffer from low blood sugar when fasting, or during an illness. Low blood sugar may be serious and can result in coma. Giving glucose will immediately switch off the need for energy from fats. The heart depends more on energy from fat (fatty acids) than other organs, which is why, without treatment, a heart muscle disorder (cardiomyopathy) may occur.

What is the treatment for carnitine transporter defect?
The disorder can be easily treated by taking extra carnitine by mouth every day, as a medication. Extra carnitine should be taken for life. This appears to prevent all complications. There are very few recorded side effects from carnitine, and these are mild. If the dose is too high, there may be mild diarrhoea and perhaps a fishy smell, but lowering the dose abolishes these effects.

How do you get carnitine transporter defect?
The carnitine transporter defect is inherited from your parents. Genes are the inheritance particles we get from our parents that determine how each part of the body works. They come in pairs, one from each parent. To inherit carnitine transporter defect, both parents must carry one good and one faulty gene. The baby has by chance inherited a faulty gene from each parent. There is a one in four, or 25% chance that any further baby in the family will also have a defect in the carnitine transporter. Being a carrier of one faulty gene does not seem to cause any problems in most cases. Some mothers who are known to be carriers have found the latter part of pregnancy very tiring, and have found that taking some carnitine by mouth is very helpful indeed. Other carrier mothers have not noticed a problem.

What is the outlook?
The few children we have treated in New South Wales have all done well on carnitine treatment. The small number of reports from around the world also echoes this. As this disorder was only first described less than 30 years ago, the very long-term outlook is not known, but seems likely to be good.