

PROTOCOL FOR NEWBORN SCREENING RESULT

Elevated acylcarnitine, associated with Carnitine Palmitoyltransferase I (CPT I) Deficiency

Repeat newborn screening result

Normal repeat newborn screening result.

If the first screen showed a markedly elevated carnitine profile, a normal second screen result is reassuring BUT this does not rule out CPT I deficiency. Therefore the metabolic physician may want to continue treating the baby as though he/she has CPT I deficiency while awaiting the results of more definitive tests.

If the first screen was only mildly elevated however, the newborn screening increase was probably transient (false positive).

Once the metabolic team has confirmed that the infant does not have CPT I deficiency, it is essential to reassure the family that their baby is well and that they should treat their baby as entirely normal. Many people can be traumatized by a false positive result and counseling may be appropriate. If the metabolic physician remains concerned, however, then he/she will discuss this further with you and may decide to continue with frequent feeds and early intervention if the baby becomes sick. It is important to remember, however, that this does not mean that the baby has CPT I deficiency but only that the metabolic doctor is taking an extra cautious approach until definitive results are available to keep the baby safe and well.

Abnormal repeat newborn screen result.

An elevated carnitine profile on the second sample is very suspicious of CPT I deficiency and further evaluation by the metabolic doctor is definitely required. The baby must be treated as though he/she has CPT I deficiency while definitive testing is carried out.

See **markedly elevated carnitine profile, probable CPT I deficiency** discussions in first newborn screening result section.