CPT1 Deficiency

Carnitine Palmitoyl Transferase Deficiency

CPT (Carnitine palmitoyl transferase) is an enzyme in the body that determines how a person can convert fat to energy. "CPT1 deficiency" happens when one enzyme used to break down fat is missing or not working properly.

In Alaska, every baby is tested for CPT1 as part of the newborn screening test. "Classic CPT1" is a rare, serious disorder; babies with this deficiency cannot make the CPT enzyme at all. A mild form of CPT1 is common among Alaska Natives and Canadian Native peoples. Most Alaska Native babies with CPT1 can make 10-25% of the normal amount of the CPT enzyme.

Treatment:

Ten to 25% of enzyme activity is enough for a baby to maintain good health, if the baby eats frequently. Babies with mild CPT1 are as healthy and normal as other babies. However, if a baby with mild CPT1 does not eat for more than 8 hours, he/she can get sick very quickly and develop seizures and coma.

Guidelines for Feeding Your Baby with mild CPT1

Newborn to 6 months old:

Feed your baby every 3-5 hours. The amount of breast milk or formula your baby needs is the same as other babies. It is important to feed your baby regularly every 3 to 5 hours.

- If your baby won't eat for two feedings, take your baby to the health aide to be sure that your baby does not have a fever or other illness that can be treated.
- Don't wait until your baby gets sick; talk with the health aide and be sure that your baby is safe.
- If your baby hasn't eaten for 8 hours, the health aide will arrange for the baby to go to the hospital. Your baby cannot go longer than 12 hours without food or IV fluids without becoming very ill.

6 to 12 months old:

As he/she grows, your baby will change how often and how much he/she wants to eat. Space meals 4 to 6 hours apart. Let your baby sleep for 6 to 8 hours at night.

- Be sure that your baby is breast fed or has formula or a snack before he/she goes to bed and be sure that your baby has breakfast as soon as he/she gets up each morning.
- During the day, offer your baby cereal, fruits, and vegetables. Begin to use a cup.

- Illness: If your baby refuses to eat two meals, has a fever, or vomits, take your baby to the health aide.
- If your baby hasn't eaten for 8 hours, the health aide will arrange for the baby to go to the hospital. Your baby cannot go longer than 12 hours without food or IV fluids without becoming very ill.

Over 1 year old:

Your child can space his meals 6-8 hours apart. Offer your child three meals and three snacks each day at regular times. Be sure that your child has breakfast and a bedtime snack.

- Your child does not need more food than other children his age. Your child needs small amounts of food at regular times each day. Do not force your child to eat when he is well.
- If your child is ill and hasn't eaten for 12 hours, the health aide will arrange for him/her to go to the hospital. Meanwhile try to have the child sip some juice or drink something with sugar in it.

Medical Care and Medicines:

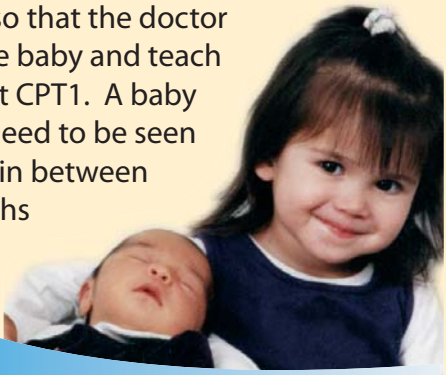
Your baby or child with CPT1 can have medications, medical care, or surgery as needed to stay healthy. If your baby or child needs surgery, IV fluids will be started before surgery and continue until the surgery is over and your baby or child is able to eat again.

Genetics of CPT1:

CPT1 is inherited. A baby with CPT1 has inherited two CPT genes -- one from the mother and one from the father. Brothers and sisters of a baby with CPT1 may have inherited this also. If other children in the family were born before October 2003, ask your doctor to test them for CPT1. This can be done by a cheek brush or dried blood spot at the pediatric clinic.

Metabolic Clinic:

CPT1 deficiency is a newly recognized condition. Doctors are still learning about why some children with CPT1 have mild problems and others have severe problems. Every baby with CPT1 needs to attend the metabolic genetics clinic before 6 months of age so that the doctor can examine the baby and teach the family about CPT1. A baby with CPT1 will need to be seen at the clinic again between 12 and 18 months of age.



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For more information about CPT1,
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