Reykjavík May 11th 2013

To: Whom it may concern.

Re Carnitine palmitoyltransferase 1 deficiency.

Carnitine palmitoyltransferase 1 (CPT I) deficiency is a metabolic condition, which affects the energy metabolism of the body. This is a very rare genetic condition – approximately 50 cases have been reported in the medical literature. Because of lack of a certain enzyme (carnitine palmitoyltransferase 1) these individuals are unable to convert certain types of fat in the food (long chain fatty acids) into energy, which may result in energy failure in the body under certain conditions. This occurs most commonly during prolonged periods without food and drink, or during illness with fever or vomiting and diarrhea. When this happens the blood sugar (glucose) becomes low and the ketones (products of fat breakdown that are used for energy) in the blood are also low. This results in energy failure in the body, including the brain, with decreased level of consciousness, coma and convulsions. This may result in permanent brain damage and even death. These individuals are also at risk of developing abnormal liver function and even liver failure. In addition, they frequently have increased amount of acids in their blood due to abnormal kidney function (renal tubular acidosis).

Because individuals with CPT 1 deficiency are at risk of developing serious and even life-threatening condition due to low blood sugar it is important that they feed on a regular basis, especially infants and young children which should be fed a 3 – 4 hours intervals. As most types of fat (except coconut oil) contain long chain fatty acids individuals with CPT 1 deficiency should preferably consume diet which is high in carbohydrates and low in fat. They are also frequently given a special type of fat (MCT, medium chain fatty acids) which is more easily metabolized into energy than most other types of fat. Also, it is important to prevent as much as possible that they get infectious diseases which cause fever, vomiting or diarrhea. When this happens they frequently need to be admitted urgently to a hospital for evaluation and for administration of glucose containing solution into the blood to prevent or to correct low blood glucose, and to decrease the breakdown of fat in the body. To treat the increased amount of acids in the blood bicarbonate is given a few times every day. Regular follow-up by a doctor is important. During follow-up visits blood tests are done to evaluate the condition of the liver and the amount of acids in the blood.

In summary, CPT 1 is a serious, rare metabolic condition which may result in severe illness and even death if these individuals do not receive correct dietary and medical interventions as needed, especially infants and children.

Sincerely,

[redacted], MD
Director Neonatal Intensive Care Unint
Children's Hospital Ice and
101 Reykjavik
Iceland
Pnone: [redacted]
E-mail: [redacted]*@landspitali.is*