A NEW THERAPEUTIC APPROACH IN SCL25A42 MUTATION WITH KETOGENIC DIET

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Introduction: SLC25A42 gene encodes an inner mitochondrial membrane protein which is responsible for the exchange of coenzyme A (CoA) into the mitochondrial matrix. Because CoA has an essential role in major processes occurring in the organelles, deficiency of CoA in mitochondria results in energy depletion. Until now a sparse number of cases have been reported and common features comprise; elevated lactate levels, motor mental developmental delay, and proximal muscle weakness, and epilepsy.

Case Presentation: Here we presented a boy who consulted our clinic at the age of 10 months old, with severe motor developmental delay, ketoacidosis, and encephalopathy. He was born spontaneously, 3000gr, from consanguineous parents. His neuromotor mile stones were normal until his encephalopathic attack. Severe hypotonia, atypical facial features, dystonia were observed in his physical examination. At his encephalopathic attack, iv infusion of high glucose perfusion was started which was resulted in a ketolactic acidosis, and eventually glucose perfusion rate was reduced. Cranial MRI: Normal. His genetic study showed a homozygous variant of uncertain significance at SLC25A42 gene; c.287-289delCCA (p.Thr96del).
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Thereupon, we planned to give a ketogenic diet of which energy would be supplied from fats. Because he has been hypotonic and had been on continious enteral feeding with 130 kcal/kg energy so he was overweighted, we’ve planned adequate energy ketogenic diet for his age. He is now on a ketogenic diet for 9 months, his ketogenic diet contains; 70kcal/kg, %31 carbohydrates, %8 protein, %60 fat, %30 MCT. He has not had any encephalopathic attack since the treatment also his distonia had improved, his muscle power increased.

**Ketogenic Diet**
- 70kcal/kg energy
- %31 carbohydrates
- %8 protein
- %60 fat (%30 MCT)

**Conclusion:** SLC25A42 protein mutation causes CoA depletion in mitochondria. The severity of the clinical manifestations was highly variable even within affected individuals of the same family, ranging from asymptomatic lactic acidosis to severe intellectual disability, metabolic crisis, and severe muscular involvement. Ketogenic diet could be a supportive therapeutic option for mitochondrial membrane SLC25A42 protein mutation.