DIHYDROLIPOAMIDE DEHYDROGENASE DEFICIENCY: ACUTE HEPATIC PRESENTATION WITH RECURRENT LIVER FAILURE

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Background:
The phenotypes of dihydrolipoamide dehydrogenase (DLD) deficiency are an overlapping continuum that ranges from early-onset neurologic manifestations to adult-onset liver involvement and, rarely, a myopathic presentation. Isolated liver involvement can present as early as the neonatal period and as late as the third decade. We report an Algerian girl with recurrent hepatic failure.

Case Report: A 2 years-old girl (Fig.1) from Algeria and consanguineous parents was first seen for hepatomegaly and recurrent hepatic failure since the age of 4 months. She was tested negative for Galactosemia, fructosemia, glycogen storage disease. She developed hepatic failure (ASAT: 4620 UI/l, ALAT: 3414 UI/l, GGT: 99, PT: 42%) preceded by nausea and emesis after intercurrent infections that were corrected each time by fresh frozen plasma and vitamin K supply. Amino acids by Ion exchange chromatography showed normal citrulline and slight elevation of isoleucine with normal leucine and valine, without allo isoleucine. Organic acids by GC-MS were normal and no hyperlactacidemia was noted. Between acute metabolic episodes, neurological development and hepatic assessment were within normal limits. Unfortunately, we didn’t explore the girl while she was on acute metabolic failure because she lives in Algeria. Genetic analysis finds a homozygous pathogenic variant in DLD gene c.685G>T p.(Gly229Cys) consistent with the diagnosis of autosomal recessive DLD.

Conclusions: The diagnosis of DLD is established in a proband with suggestive clinical and supportive laboratory findings and/or by identification of biallelic pathogenic variants in DLD. Targeted analysis for the c.685G>T pathogenic variant may be considered first in individuals of Algerian with probably a founder effect or Ashkenazi Jewish ancestry.