Two Siblings with Galactosemia Type-4: clinical differences

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Background

Galactose mutarotase (GALM) deficiency is an inherited metabolic disease caused by the deficiency of the first enzyme in the Leloir pathway. GALM deficiency was reported first in 2018. To date, eight cases have been reported. In addition, we present two siblings with GALM deficiency who have different clinics.

Case Report

• We evaluated the first case due to a congenital cataract at three months old. She had elevated galactose and galactose-1-phosphate and normal galactose1-phosphate uridylyltransferase (GALT) activity.

• We excluded Galactokinase 1 (GALK1) and UDP-galactose 4′-epimerase (GALE) deficiencies by genetic analysis and other laboratory and clinical findings.

• She had a homozygous mutation p.Gly277Arg (c.829G>A) in the GALM (NM_138801) gene.

• She is three years old at the last visit, and her physical examination is normal, except for cataracts.

• The same mutation was found to be homozygous in the patient's asymptomatic sibling during family screening.

Discussion

• The clinical spectrum of MCE deficiency ranges from asymptomatic to life-threatening metabolic decompensation attacks.

• This report of a patient diagnosed with MCE deficiency with acute metabolic ketoacidosis attacks and moderate MMA-uria persists in periods without decompensation.

• In addition, this report provides a new phenotype of the clinical and biochemical characterization of MCE deficiency.