Newborn Screening, Inborn Errors of Metabolism

AB080. MATI/III deficiency with demyelination of central tegmental tract during neonatal period

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Background: Methionine adenosyltransferase I/III (MATI/III) (EC 2.5.1.6), encoded by *MAT1A* gene, is the enzyme that converts methionine to S-adenosylmethionine (SAM). MATI/III deficiency (OMIM 250850) is an inherited metabolic disease resulting in hypermethioninemia, which is detectable by newborn screening (NBS). There are two clinical phenotypes: a benign phenotype with autosomal dominant inheritance and the other resulting in severe manifestations. The later one can lead to brain demyelination and neurological decompensation and is inherited in an autosomal recessive fashion.

Methods: Clinical, biochemical, radiological, and genetic investigation of a patient with suspected MATI/III deficiency were reviewed and/or performed.

Results: Our patient was a 5-day-old girl with hypermethioninemia (2 mg/dL, cut off level is 1 mg/dL) detected by NBS. Plasma concentration of methionine increased up to 20 mg/dL and she was treated by methionine-restricted diet. She developed irritability. Brain MRI showed central demyelination. Gene analysis identified compound heterozygous mutations in the *MAT1A* gene: c.812A>G (Y271C) and c.1066C>T (R356W).

Conclusions: The findings in our case indicate that myelination may be disturbed during fetal stage in patient with MATI/III deficiency.

Keywords: MATI/III; inborn error of metabolism (IEM)

doi: 10.21037/atm.2017.s080

Cite this abstract as: Matsumoto S, Kido J, Sakamoto R, Nakamura K, Mitsubuchi H, Endo F. MATI/III deficiency with demyelination of central tegmental tract during neonatal period. Ann Transl Med 2017;5(Suppl 2):AB080. doi: 10.21037/atm.2017.s080

