A Neurological Disease Mimicking Central Hypothyroidism: MCT8 Deficiency

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İNTRODUCTİON:

Monocarboxylate transporter 8 (MCT8) is necessary for the transport of T3 to neurons. The case presented here is a male infant with neuromotor retardation initially treated for central hypothyroidism who showed no benefit from treatment and a final diagnosis of MCT8 deficiency was made.

CASE REPORT:

A male infant at 13 months of age was brought to the clinic because he was unable to sit without support. The perinatal history revealed that he was born after uneventful delivery with birthweight of 3650 gr as the first child of non-consanguinous parents. On physical examination, body weight was 9.5 kg (-0.74 SDS), length was 75cm (-0.92 SDS), and head circumference was 45 cm (-1.59 SDS). Neorological examination showed hypotonicity in the trunk and spasticity in the lower extremities. He had head control but he was unable to sit without support.

In the thyroid function tests, free T4 (fT4) was low (0.68 ng/dl; N: 0.8–1.9 ng/dl) and TSH was normal (2.8 mIU/L; N: 0.4–5.0 µU/ml). The other anterior pituitary hormones as well as the brain MRI and EEG were normal.

With the diagnosis of central hypothyroidism, L-thyroxin replacement treatment was started at 3 mcg/kg/day. The dose was increased to 8 mcg/kg/d during monthly follow-up, because no increase was observed in the fT4 level. At the end of 3 months, fT4 was low (0.74 ng/dl), TSH was normal (2.1 mIU/L) and free T3 (fT3) was high (5.3 pg/ml).

Despite the increase in fT3, low fT4 level and retarded neuromotor development led us to consider MCT8 deficiency (Allan-Herndon-Dudley syndrome). The L-thyroxin replacement treatment was terminated and further diagnostic workup was planned. The genetic analysis showed c.670 G>A (A224T) hemizygote mutation on the SCL16A2 gene. The family was given genetic counselling and the patient was followed up with supportive physical therapy.

CONCLUSİON:

Mutations in the SLC16A2 gene, which encodes MCT8, cause Allan–Herndon–Dudley syndrome that is characterised by abnormal thyroid hormone levels and severe neuromotor retardation. The syndrome is defined in male patients because of X-linked transmission. The characteristic thyroid hormone abnormalities in MCT8 defect are high fT3, low fT4, and normal/high TSH. In male children with retarded development and neurological findings, this syndrome should be considered when evaluating thyroid function tests.

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