DOPAMİNE TRANSPORTER DEFİCIENCY SYNDROME: A REPORT OF TWO CASES

Merve Yoldas Celik¹, Ebru Canda¹, Havva Yazıcı¹, Sanem Keskin Yılmaz², Muzaffer Polat³, Selcan Zeybek⁴, Esra Işık⁵, Sema Kalkan Uçar³ and Mahmut Çoker¹
1 Ege University Faculty of Medicine, Department of Pediatrics, Division of Pediatric Metabolism and Nutrition, Izmir
2 Ege University Faculty of Medicine, Department of Pediatrics, Division of Pediatric Neurology, Izmir
3 Celal Bayar University Faculty of Medicine, Department of Pediatrics, Division of Pediatric Neurology, Manisa
4 Tınaztepe University Faculty of Medicine, Department of Genetics, Izmir
5 Ege University Faculty of Medicine, Department of Pediatrics, Division of Pediatric Genetic, Izmir

Background:
Dopamine transporter deficiency syndrome (DTDS) is caused by SLC6A3 mutations. Here we presented 2 patients with DTDS for discuss their clinical and laboratory findings.

Case I Report:
Twenty-month-old girl.
At 3 months; hypotonia and feeding difficulties and neurodevelopment delay

On physical examination;
Weight: 7.8 kg (-1.7 SDS)
Height: 69 cm (-2.4 SDS)
Head Circumstance: 46 cm (-0.7 SDS)
Dysmorphic features,
Distinct rigidity findings,
Diffuse dystonia
Bilateral fisting

CSF analysis;
- Elevated homovanillic acid (HVA) (1294 nmol/L, normal range: 295-932)
- High HVA:HIAA ratio (3.86, normal range: 1.5-3.5).

SLC6A3 gene: homozygous mutation
Treatment: Baclofen and levodopa.
After six months → Slight symptomatic benefit

Case II Report:
Three-year-old girl.
At 4 months, axial hypotonia and feeding difficulty.
At 6 months, dystonia and peripheral hypertonia.
At the age of 10th month, The seizures developed.

On physical examination;
Weight: 9.9 kg (-3.3 SDS)
Height: 88 cm (-2.4 SDS)
Head Circumstance: 47 cm (-2.0 SDS)
Axial hypotonia
Spasticity in the extremities
Joint contracture

Cranial MRI and EEG were normal.

CSF analysis;
- Elevated homovanillic acid (HVA) (1840 nmol/L, normal range: 295-932)
- High HVA:HIAA ratio (8.6, normal range: 1.5-3.5).

SLC6A3 gene: homozygous mutation
Treatment: Trihexyphenidyl, clonazepam and levodopa
Trihexyphenidyl was discontinued due to sleep problems.

Results:
It is important to consider DTDS in the initial differential diagnosis for patients presenting with dystonia. In both cases, high ratio of HVA to 5-HIAA is detected. In the presence of clinical suspicion, it should also be evaluated by gene analysis. Dopamine agonists have had limited utility in few patients.