West Sendromu İle Prezente Olan Dup15q Sendromu Olgusu

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INTRODUCTION

Dup15q Syndrome is a syndrome with central hypotonia, autism, mental retardation, growth retardation, ataxia, seizures and behavioral problems, which is almost always secondary to maternal duplications in chromosome 15.

CASE

HISTORY: The patient's seizures started in the form of contractions and jerks when she was 1 years old. She was diagnosed West syndrome and received 13 doses of ACTH therapy. Then phenobarbital and topiramate were added to the patient's treatment. Topiramate was replaced with levetiracetam because dual therapy was ineffective.

BACKROUND & FAMILY HISTORY: Normal

PHYSICAL EXAMINATION: Mental and motor developmental delay, no speech and object tracking, depressed nasal root, low set ears, axial hypotonia, DTR hyperactive, spasticity in all extremities

LABORATORY

- Cranial MRI: Cerebral atrophy (1 year)
- Electroencephalography (EEG): Generalized sharp slow, spike and multiple spike wave activity (2 years old)
- Chromosome analysis: 47, XX, mar.ish der (15)(D15Z1+SNRPN+)
- Microarray analysis: Four replicates of region 15q11.2

CONCLUSIONS

Three different neurodevelopmental disorders can occur depending on deletions or duplications in the 15q11-q13 region; Prader Willi Syndrome (PWS), Angelman Syndrome (AS) and Dup15q Syndrome. Although it has some features of both PWS and AS, some features are specific to the disease. Duplications causing this syndrome are almost always of maternal origin. Syndrome is characterised atypical facial appearance, central hypotonia, autism, mental retardation, growth retardation, ataxia, seizures, and behavioral problems. Seizures in Dup15q Syndrome occur in 60% of patients and often begin before the age of 5. Patients may have multiple seizure types such as infantile spasm, tonic, atonic, tonic-clonic, myoclonic, focal seizure in which awareness is lost, and atypical absence. These patients have an increased risk of 'Sudden Unexpected Death in Epilepsy'. There is no specific treatment for Dup15q Syndrome. Treatment is recommended for seizure control, physical therapy for hypotonia and spasticity, and speech and language therapy for the autism spectrum.

REFERENCES

1. Chamberlain SJ, Lalande M. Neurodevelopmental disorders involving genomic imprinting at human chromosome 15q11-q13. Neurobiol Dis. 2010;39(1):13-20.

2. Kalsner L, Chamberlain SJ. Prader-Willi, Angelman, and 15q11-q13 Duplication Syndromes. Pediatr Clin North Am. 2015;62(3):587-606.

3. Al Ageeli E, Drunat S, Delanoë C, et al. Duplication of the 15q11-q13 region: clinical and genetic study of 30 new cases. Eur J Med Genet. 2014;57(1):5-14.

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