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Olgu Sunumu / Case Report

Trisomy 9 Mosaicism Presenting with Epilepsy, and Facial Dysmorphism: A Case Report

Epilepsi ve Fasiyal Dismorfizm ile Gelen Trizomi 9 Mozaisizm: Bir Olgu Sunumu

Faruk İncecik[1], M. Özlem Hergüner[1], Gülen Mert[1], Şakir Altunbaşak[1]

[1]Çukurova University Faculty of Medicine, Pediatric Neurology, Department of Pediatrics, ADANA

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ABSTRACT

Trisomy 9 syndrome is a rare genetic disorder. Trisomy 9 has two forms; 1) mosaic, 2) non-mosaic. The patients usually present similar clinical features, independent of the presence of mosaicism, characterized by growth retardation, mental deficiency and brain, facial, cardiac, renal and skeletal abnormalities. Developmental delay and mental retardation are the most common neurological symptom in trisomy 9 mosaicism in our knowledge. Epilepsy associated with this syndrome has not found in literature. We describe a 10-year-old boy with trisomy 9 mosaicism who presented seizures, and dysmorfic features.

Key Words: Trisomy 9 mosaicism, Developmental delay, Epilepsy, Children

ÖZET

Trizomi 9 sendromu, nadir görülen genetik bir bozukluktur. Trizomi 9 sendromu 2 forma sahiptir; 1) mozaik, 2) non-mozaik. Hastalar genellikle mozaisizm varlığından bağımsız olarak büyüme geriliği, mental retardasyon, serebral, yüz, kardiyak, renal ve iskelet anomalileri gibi benzer bulgular ile gelirler. Literatür bilgilerimize göre trizomi 9 mozaisizminde en sık görülen nörolojik semptom, gelişme geriliği ve mental retardasyondur. Literatürde epilepsinin bu sendrom ile ilişkisi bulunamadı. Trizomi 9 mozaisizmi saptanan, dismorfik özellikleri ve nöbetleri olan 10 yaşında bir erkek çocuk bildirdik.

Anahtar Kelimeler: Trizomi 9 mozaisizm, Gelişme geriliği, Epilepsi, Çocuklar

INTRODUCTION

Trisomy 9 is a rare chromosomal abnormality, characterized by mental retardation and growth retardation, facial dysmorphim (low-set ears, microphthalmia, a bulbous nose, a small mouth, a high-arched palate), central nervous system anomalies (hydrocephalus, Dandy-Walker malformation, and holoprosencephaly), congenital heart defects (most commonly ventricular septal defect), genitourinary anomalies (hypoplastic genitalia, cryptorchidism, cystic kidneys, hydronephrosis), and skeletal anomalies (joint dislocations, deformations)[1,2].

Trisomy 9 syndrome have been described mosaic and non-mosaic forms. Complete trisomy 9 results spontaneous abortion in first trimester and is rarely seen in live-born infants with multiple malformations and results in early death. Trisomy 9 mosaicism is associated with prolonged survival and variable clinical features[2,3].

Epilepsy and mental retardation are the most common neurological symptom in chromosomal abnormality[4]. However, we could not find epilepsy in trisomy 9 mosaicism in literature.

Here, we report a case of a 10-year-old boy with trisomy 9 mosaicism presenting seizures and facial dysmorfism.

CASE REPORT

A 10-year-old boy was admitted to our hospital because of seizures. He had generalized tonic clonic seizures since at age 7 years. He was started valproic acid for seizures, but it was ineffective. Then, levetiracetam was changed added for seizures, and his seizures stopped.

In past medical and family history, he was born after 38 weeks of gestation with a birth weight of 1900 g. His parents were nonconsanguineous. There was no family history of seizures or developmental or other neurologic disorders. He underwent orchidopexy for right cryptorchidism at the age of two years. His neuropsychomotor development was delayed, since he started to walk without support at the age of three years and spoke his first words at the age of four years and six months.

On physical examination, his weight was 28 kg (P25), length was 127 cm (< P3), and head circumference was 47 cm (< P3). He had microcephaly, prominent wide nose with bulbous nasal tip, epicanthal folds, small mouth, high arch palate, micropenis, and small testicles (Figure 1). Ophthalmologic examination was normal. We did not detect hearing loss. The remainder of the physical and neurological examination was unremarkable.

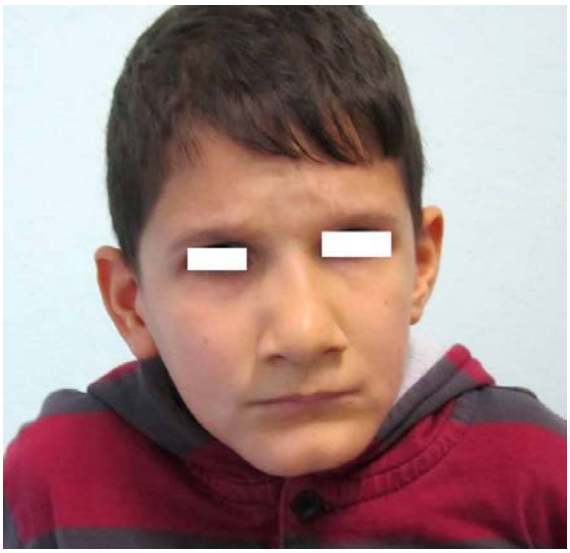

Figure 1. A 10-year-old boy with a trisomy 9 mosaicism with dysmorphic features**.**

Laboratory investigations were normal (serum lactate, pyruvate, biotinidase levels, urine and blood amino acid chromatography, urinary organic acids). Her cranial magnetic resonance imaging, initial and following interictal EEGs, echocardiogram, and abdominal ultrasound were also normal.

A combination of microcephaly, mild dysmorphic features, developmental delay, and seizures suggested the presence of a chromosomal abnormality. Karyotype analysis was performed from peripheral blood cell culture and GTG banding was applied. The karyotype was 47,XY,+9[38]/46,XY[5] (Figure 2).

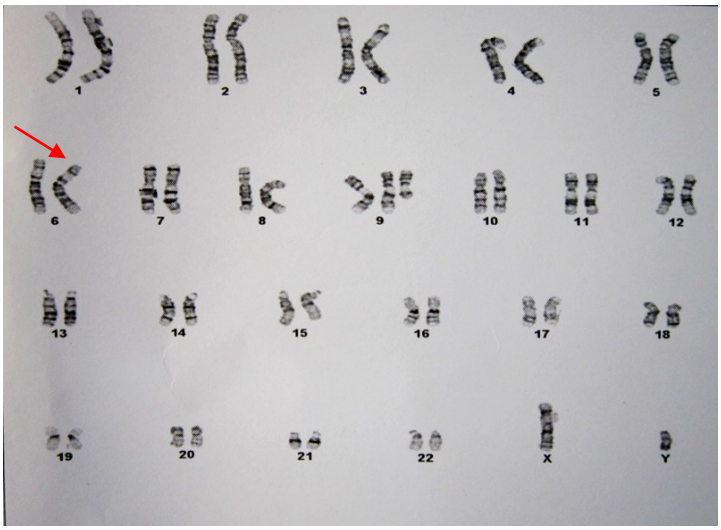

Figure 2. The karyotype of the patient showing 47,XY,+9[38]/46,XY[5].

DISCUSSION

Trisomy 9 is considered to be a rare chromosomal abnormality first described in 1973, more than 50 patients have been described in the literature so far[5]. Trisomy 9 has been reported both alone and especially, in mosaic with a normal cell line. Growth retardation, mental retardation, brain, facial, cardiac, renal and skeletal abnormalities are seen in trisomy 9. The mosaic forms of trisomy 9 are less severe symptoms and good prognosis in the literature[3,6].

Facial abnormalities are common in cases of mosaic trisomy 9. These abnormalities are microcephaly, hypertelorism, high or narrow forehead, short and upslanting palpebral fissures, deep-set eyes, microphthalmus, epicanthal folds, broad nasal bridge, bulbous nose, high arched palate, cleft lip or palate, and micrognathia[1,5]. Cardiac abnormalities and genitourinary abnormalities are seen 70% and 73%of the cases with this syndrome, respectively. Gastrointestinal abnormalities are also infrequent[7,8]. Motor and mental retardation are the most common neurological symptom in this syndrome. But, normal neurodevelopmental state also have been reported in mosaic form of this syndrome in some previously reports[7]. Our patient had neurodevelopmental delay. Central nervous system abnormalities, hearing loss, cardiac abnormalities, and gastrointestinal abnormalities were not detected in our patient. However, we described epilepsy. We could not find epilepsy in this syndrome in literature. To our knowledge, this is the first report with epilepsy.

In conclusion, mosaic trisomy 9 is a rare chromosomal abnormality that appears to present significant phenotypic variability. Chromosomal analysis should be performed on children with epilepsy with dysmorphic features.

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Yazışma Adresi / Address for Correspondence:
Dr. Faruk İNCECİK
Cukurova University Faculty of Medicine
Pediatric Neurology Department of Pediatricsı
01330 Balcalı-ADANA
Email: fincecik@yahoo.com

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