Duplication 9p Syndrome; Case Report

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ABSTRACT

Duplication 9p syndrome is a rare genetic disorder in which extra chromosomal material is found on the short arm of chromosome 9 (p). The extra chromosome may include the entire short arm, part of the short arm, or part of the long arm (q). Despite differences in duplication size, phenotypically, especially facial and digital anomalies are seen.[1]

The term “Duplication 9p syndrome” is often used when there is a partial rather than a complete copy of 9p. If it is an exact copy, it is called trisomy 9p or trisomy 9p syndrome.[2]

General phenotypic features duplication 9p syndrome includes short stature, microcephaly/brachycephaly, downward sloping palpebral fissure, hypertelorism, prominent and large nose and bulbous nasal tip, short broad neck, short philtrum, downward curved corners of the mouth, droopy ears, anomalies of fingers and toes. Mental retardation and/or developmental delay may accompany these dysmorphic findings. The phenotypic heterogeneity in most cases is due to a change in the size of the replicated segment or to the often-accompanying monosomy or other chromosome segment.[1,2]

CASE

A two-year-old girl was admitted to our outpatient clinic with the complaint of developmental delay. She was born 2215 gr with normal spontaneous vaginal delivery at 38th gestational week. The patient had a history of hospitalization in the neonatal intensive care unit due to SGA, feeding difficulties and neonatal jaundice. There was no consanguinity between the parents. In his examination, the head circumference z score was -2.22 SDS. In addition, frontal bossing, flattened nasal root, prominent large nose and bulbous nasal tip, low ear, hypertelerosis, exotropia, short neck was observed. (Photo 1)

Cranial MRI revealed nonspecific gliosis in the right parietal lobe. In the DNA microarray gene analysis performed from the patient's peripheral blood sample, approximately 38 mb duplication was detected in the 9p24.3p13.1 region. (Figure 1) The patient and his family were referred to the medical genetics’ outpatient clinic for genetic counseling.

Photo 1: frontal bossing, flattened nasal root, prominent large nose and bulbous nasal tip, low ear, hypertelerosis, exotropia, short neck.

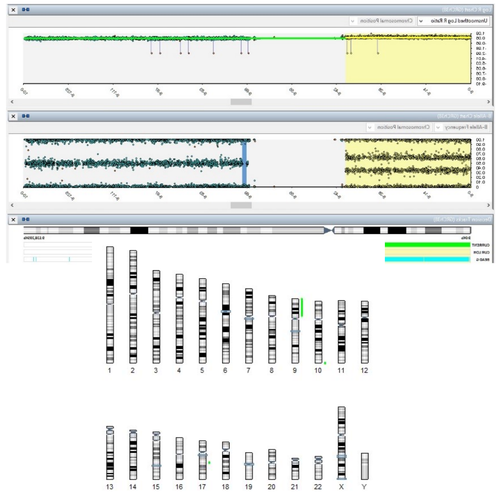

Figure 1: Duplication 9p syndrome result of the patient with the microarray method

CONCLUSİON

We wanted to draw attention to the phenotypic features of duplication 9p syndrome, which is a rare genetic syndrome, such as frontal bossing, flattened nasal root, prominent large nose and bulbous nasal tip, low ear, hypertelerosis, exotropia, and short neck.

REFERENCES

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