RING CHROMOSOME 18 CASE WITH 18p DELETION

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INTRODUCTION

The ring chromosome is formed by the fusion of the break points and the loss of the distal fragments as a result of the fracture in both arms of the affected chromosome. The typical clinical signs of 18p and 18q syndromes in the ring chromosome of the 18th chromosome vary depending on the size of the deletion in 18p and 18q (1). Due to deletions in ring chromosome 18p and 18q: speech delay, growth retardation, motor mental retardation (MMR), microcephaly; skeletal anomalies accompanied by chest deformities such as short neck, pectus excavatus; dysmorphic facial findings accompanied by ptosis, high palate, micrognathia, dental anomalies, low flattened ear and nasal root; heart anomalies, holoprosencephaly, corpus callosum anomalies and rarely convulsions may accompany (2).

CASE PRESENTATION

A 5-year-old girl presented with the complaints of growth retardation and delayed speech. On examination, narrow forehead, long eyelashes, sparse teeth, microcephaly (head circumference: 46 cm - 2.82 SDS), faint philtrum, strabismus, short neck, high palate, sandal finger, syndactyly between the 2nd and 3rd fingers, and a low set ear were found. Gross and fine motor areas and language development were markedly retarded in the Denver developmental test. In Cranial Magnetic Resonance Imaging, the anterior posterior length of the corpus callosum was normal except that it was observed to be slightly shorter than normal for the age group. Magnetic Resonance Spectroscopy was normal. In echocardiography, no pathology was detected except for the patent foramen ovale. In karyotype analysis; The ring chromosome formed by the union between the p11.2-q23 regions of the 18th chromosome in 32 (64%) of the 50 metaphases examined (Figure-1) , and the deletion of the p11.2 region of the chromosome (46,XX,r(18)(p11.2q23)[32]/46,XX,del(18)(p11.2)[18]) in 18 of them were detected (Figure-2).

CONCLUSION

Our patient is presented because it is genetically and clinically compatible with 18p deletion and ring chromosome 18. Although not seen in our patient, It should be considered that rarely convulsions may accompany 18p deletion syndromes.

REFERENCES

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Figure 1: In karyotype analysis; The deletion of chromosome 18 was demonstrated.

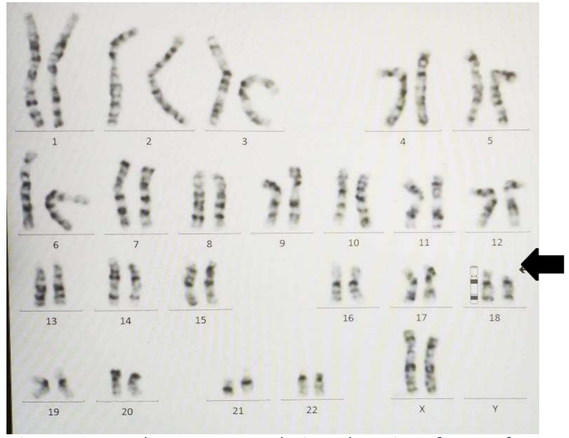

Figure 2: In karyotype analysis; The ring form of chromosome 18 was demonstrated.