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A Rare Chromosome 18p Deletion Syndrome in Abha City: A Case Study

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Abstract

Background: The 18p deletion disorder is a chromosomal condition due to the absence or deletion of a short arm or all the chromosome. The symptoms and signs vary among patients which may include mental and post-natal growth retardation that ranged from moderate to severe. The cause of 18p deletion among 85% of cases is due to *de novo* deletions and unwarranted hereditary transmission of structural rearrangements among the rest of cases.

Case presentation: A 16-years-old male was presented with dysmorphic features and refereed for chromosomal analysis. He was born full term by vacuum assisted vaginal delivery with a birth weight of 4.5 kg with no gestational complications. His gestation was without any complication. The pre-natal ultrasound showed vesico-urethral reflux on the left side and hypocalcemia. He had speech delay at 11 months, alopecia areata at 13-years-old and at age of 14-years-old was diagnosed with celiac disease. The MRI brain showed normal brain and chromosomal analysis for dysmorphic features showed brachycephaly, ptosis, squint (intorsion). The study revealed Deletion of a segment of the short arm of chromosome 18 at a breakpoint 18p11.2 band. Chromosomal analysis was of patient's mother revealed normal karyotype 46-XX.

Conclusion: This case is quite variable than other studies reported in literature as that patient showed mild dysmorphic features with normal brain function. The clinical presentation is associated with 18p deletion which need further diagnosis and essential management as the symptoms vary regarding the quantity of chromosomal loss.

Keywords: Deletion of 18p syndrome; Dysmorphic features; Chromosomal analysis; Genetic syndrome; Speech delay; Developmental disorder

Introduction

The 18p deletion disorder is a chromosomal condition due to the absence or deletion of a short arm or all the chromosome. The worldwide prevalence of 18p deletion were found among 150 cases but most of the cases were not published [1]. Its incidence rated from 1 patient out of 50 thousand infants with a ration ranging from 3:2 among females to males [2]. Also, several studies have been shown in literature regarding patients with 18p deletion suffering from deficiency of growth hormone [3,4].

The symptoms and signs vary among patients which may include mental and post-natal growth retardation that ranged from moderate to severe. The clinical features also may include dysplastic ears, drooped corners of mouth and round face. Some other features are less frequently as micrognathia, ptosis, microcephaly, hypertelorism, epicanthic folds, short neck, and dental anomalies [5]. Also, some other congenital malformations have been reported including such as congenital heart defects or brain malformations [6].

The cause of 18p deletion among 85% of cases is due to *de novo* deletions. Also, unwarranted hereditary transmission of structural rearrangements may be another cause among the rest of cases [7]. We present the results of an evaluation of a case with 18p deletion syndrome.

Case Presentation

The patient is a 16-years-old male, who was presented with dysmorphic features and refereed for chromosomal analysis. As for the previous history, the patient was born full term by vacuum assisted vaginal delivery with a birth weight of 4.5 kg. His gestation was without any complication, pre-natal ultrasound showed vesico-urethral reflux on the left side. The age of mother was 35-years-old during his birth. He was diagnosed with hypocalcemia. Regarding developmental millstones, after delivery his developmental progression was as his siblings till 11 months with speech delay. At age of 13, he was diagnosed with alopecia areata and at age of 14 years old, he was diagnosed with celiac disease.

While following up with nephrology for VUR he was advised to do MRI brain and chromosomal analysis for dysmorphic features. His dysmorphic features were as following brachycephaly, ptosis, squint (intorsion). The brain MRI was normal, while his chromosomal analysis revealed Deletion of a segment of the short arm of chromosome 18 at a breakpoint 18p11.2 band.

Chromosomal analysis was done by Standard stimulated blood lymphocyte culture with synchronization followed by chromosome metaphase slide preparation, GTG staining, and image analysis by CytoVision software. Number of Cells Counted: 20, number of Cells Analyzed: 5, number of Cells karyotyped: 2 Band Resolution: 450-500 Karyotype 46, XY, del (18) (p11.2).

The chromosomal analysis of patient's mother revealed normal karyotype 46-XX, while the study wasn't done for the patient's father. And there isn't any family history of similar condition or any other chromosomal anomaly.

Discussion

This is a case of a boy presented with an 18p deletion. The chromosomal analysis revealed normal parents' analysis thus the cause of 18p deletion among in this case is due to *de novo* deletions [7]. As for the development it was normal as his siblings, but he had a speech delay this was consistent with many studies showings that the patients with 18p deletion achieved normal milestone except for speech delay [1,2].

Also, this study showed that the child was suffering from alopecia areata and celiac disease. There has also been a reported association

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between alopecia and other skin and hair disease with the deletion of chromosome 18p [8,9].

Regarding the neurological function, the MRI revealed a normal brain, and this could be attributed to that who have smaller deletions originating at 18p11.21 are less impaired than those with larger deletions originating at 18p11.1 who would suffer from severe brain malformations [6-10].

The present study showed less dysmorphic features including brachycephaly, ptosis, squint (intorsion) which are less than other studies. This was consistent with many studies revealing that 18p deletion is associated with minor dysmorphic conditions which vary from mild to severe according to the severity of the deletion [1-11].

Conclusion

This case is quite variable than other studies reported in literature as that patient showed mild dysmorphic features with normal brain function. The clinical presentation is associated with 18p deletion which need further diagnosis and essential management as the symptoms vary regarding the quantity of chromosomal loss.

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