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Case Report

A Rare Case of Chromosomal Disorder Wolf–Hirschhorn Syndrome at Teritary Care Centre

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ABSTRACT

Background: A rare chromosomal anomaly, Wolf–Hirschhorn syndrome (WHS), which results from the deletion of 4p (4p).

Case: The case was identified at AIIMS Raipur, Chhattisgarh. The diagnostic findings included a "Greek warrior helmet" appearance of the head, ptosis, and a flat nasal bridge.

Discussion: We reported patients with submicroscopic deletions at 4p16.3. In the current case, the diagnosis was on the basis of karyotype and clinical features. Our results showed the importance of testing with the conventional karyotype.

KEYWORDS

• Karyotype • Wolf–Wolf-Hirschhorn Syndrome • Ptosis • Micrognathia

INTRODUCTION

Wolf–Hirschhorn syndrome (WHS) is a rare chromosomal disorder. Incidence of WHS is approximately 1 in 50,000 births.[1] Female babies are more affected.[2,3]

Case: The female baby of five month who was hailing from Uttar Pradesh visited AIIMS, Raipur for complaints of poor weight gain and development and episode of generalized tonic-clonic seizure. The patient was admitted in the Pediatric ward Height and weight of the baby were 50cm and 2.5kg, respectively. Head circumference was 33.5cm. The patient had features of microcephaly, flat nasal bridge, low-set malformed ears, small chin-micrognathia, "Greek warrior helmet" appearance of the face, short philtrum, bilateral ptosis, weak

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muscle tone, long fingers and and delayed milestones. The child also had dilated right atrium and right ventricle, atrial septal defect and a small patent ductus arteriosus observed in the echocardiography. The diagnosis was done with karyotype and clinical correlation. Karyotype showed 4p-microdeletion.

DISCUSSION

The severity varied from mild to severe in WHS. Three phenotype were seen according to the deletion of the short arm of chromosome 4: Phenotype with deletion <3.5MB (mild), deletion with 5 to 18MB (moderate), large deletions with 22-25 MB.[2,3]



Figure 1: Baby with Wolf–Wolf-Hirschhorn syndrome showing a. Greek warrior helmet" b. Ptosis

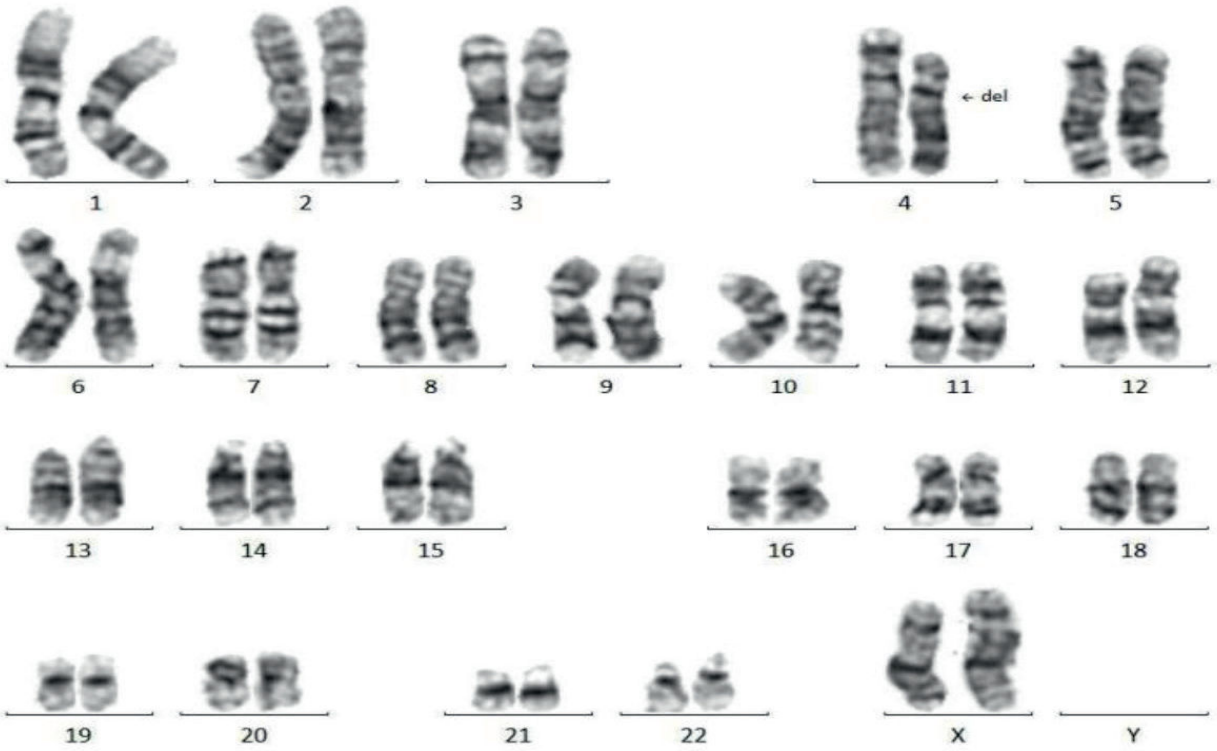


Figure 2: Karyotype 46,XX,del(4)(pter p15.3)

MSX1 gene (OMIM142983) located on the proximal part of chromosome 4p on 4p16.2 may be involved in dental, lip, and palate growth, absence or loss, leading to dental abnormalities, cleft lip and Cleft palate which is seen in WHS.[4,5]

Basic phenotype of cases with WHS is Growth retardation, Craniofacial dysmorphia, epilepsy, metal retardation. Moderate to severe psychomotor retardation with the presence of microcephaly and often structural abnormalities of the brain (fine corpus callosum) are well known findings. Epilepsy happens in 95% of children with time of onset within the first 3 years of life and peak time at around 6 to 12 months of age (6 Battaglia).

Congenital heart defects was found in 50% of children.[7,8] Ventricular septal defect and pulmonary stenosis was found by Vega *et al*[8] In our case right atrial dilatation, right ventricular dilatation, along with ASD and mild PDA was observed.

Ptosis was a very prominent finding in our case. Stabismus, hyperterolism, long eyelashes, broad nasal bridge, beaked nose, short philtrum, prominent philtrum column were also observed in Vega *et al*'s cases.[8]

CONCLUSION

In the current case, we have made the diagnosis on the basis of karyotype and clinical features. The observed finding was 4p16 microdeletion. Our results showed the importance of testing the conventional karyotype. This syndrome needs multidisciplinary approach to plan treatment.

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