

Craniosynostosis, Ptosis, Hypodontia, Prominent and Everted Lower Lip, Mental Retardation; Is it a Second Case of Mehta-Lewis-Patton Syndrome?

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Abstract

We present an unknown case of an 11-year-old boy with mental retardation, microcephaly, prominent ears, unilateral ptosis, long philtrum, prominent and everted lower lip, abnormally shaped teeth and developmental delay. This is an unknown case with special facial features and mental retardation which can probably be the second case of Mehta-Lewis-Patton syndrome.

Key Words: Craniosynostosis; Mental retardation; Microcephaly; Ptosis; Everted lip

Case presentation

An 11-year-old boy was referred to us for further investigation because he had dysmorphic features and was intellectually disabled. He was the first child of healthy, non-consanguineous parents. At birth, the mother was 20 years old and the father 25 years old. His younger sister was healthy. The family history was unremarkable. There was no history of infections or medication during pregnancy. At sixteen weeks of pregnancy his mother was admitted with moderate vaginal bleeding which settled with bed rest.

He was born by vaginal delivery after 38 weeks' gestation with a low birth weight of

1300g (<3rd centile), length of 44cm (<3rd centile) and a head circumference of 32 cm (10-50 percentile). He was cyanosed at birth because of meconium aspiration but was discharged from hospital on the third day of life. There was a significant delay in developmental milestones. He sat at 12 months, walked at 2 years and said only a few words at 7 years of age. A febrile seizure was noted on one occasion at the age of 2 years.

On clinical examination, he had a slender body, weighed 27kg (<3rd centile), his height was 128cm (<3rd centile) and head circumference measured 48cm (<3rd centile). There was asymmetrical ptosis of the right eye

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(Fig 1). Ophthalmic examinations revealed refractive accommodative isotropia and strabismic amblyopia. Refractive error was OD +8.00 and OS +7.00. Visual acuity in both eyes was nearly 20/100. The ears were prominent (Fig 2). There was a long philtrum and prominent, everted lower lip (Fig 3). The teeth were small, conical and widely spaced (Fig 4). The structure and growth of the hair and nails were normal. No deformity was seen in his fingers, toes and joints. The external genitalia were unremarkable. There was a significant intellectual disability with an intelligence quotient between 50-69 (moderate to severe mental retardation)^[1].

On neurological examination, no abnormal findings were seen. Skull x-ray showed craniosynostosis. GTG-banded chromosome study demonstrated a normal male karyotype,

46 XY. Routine biochemistry studies, abdominal ultrasound and brain MRI were normal. Echocardiogram showed mitral valve prolapse with ejection fraction of 60%. No EEG abnormalities were seen. Moreover, metabolic evaluation for metabolic disease and molecular evaluation for fragile X were normal.

Discussion

The patient presented in this short report has the clinical features of prominent ears, long philtrum, abnormally shaped teeth, prominent and everted lower lip, unilateral ptosis, craniosynostosis with developmental delay.



Fig 1 A-D: AP and lateral view illustrating the facial appearance. Note the presence of ptosis (A), prominent ears and long philtrum (B), prominent and everted lower lip (C) and abnormally shaped teeth (D)

Some of the features are seen in recognized syndromes but it is difficult to find a complete match. In the report by Ohdo et al (1986)^[2] two sisters had a combination of congenital heart disease, ptosis, blepharophimosis, hypoplastic teeth, and severe mental retardation but craniosynostosis and the other facial features reported here were not present. In 2004, a case of 13-year-old girl with short stature, microcephalus, blepharophimosis, ptosis, bilateral microphthalmia, hypogonadism and severe mental retardation was reported by Hirayama et al. Her weight, height and head circumference were below -3D. She did not have hypoplastic teeth, heart anomalies and seizures^[3]. The case reported by us has special facial features, mild mental retardation with normal genitalia and abnormally shaped teeth.

Gorlin et al (1960) described two girls with craniosynostosis, oligodontia, and congenital heart disease with normal intelligence. Also, they showed additional features like labial hypoplasia, hypertrichosis and microphthalmia^[4]. While in our case, external genitalia were normal and intelligence quotient was significantly lower. The combination of blepharophimosis and congenital heart disease has been reported in the Marden-Walker syndrome^[5]. In this syndrome, joint contractures were a prominent feature, whereas congenital heart disease and joint contractures were not present in our case.

Garavelli et al (2000) reported a boy with a phenotype mostly resembling the condition named Marden-Walker syndrome. In addition, he had hypoplastic corpus callosum, cerebellar vermis hypoplasia, enlarged cisterna magna and vertebral abnormalities^[6], whereas MRI of the brain in our case was normal.

In 1989, Mehta et al described an unknown syndrome in a two year old male with congenital heart disease, bilateral ptosis, abnormally shaped teeth, craniosynostosis, prominent and low set ears, long philtrum, prominent lower lip, joint laxity, long fingers and toes and delay in psychomotor development^[7]. He had a repair of total anomalous pulmonary venous drainage at six days of age. Although many features of our

case are the same as that of Mehta and colleagues, congenital heart disease and joint laxity were not seen in this case.

We have used the London Dysmorphology Database^[8] and have not been able to place all of the clinical features of this case with a recognized syndrome. However the majority of clinical features appear to match with the syndrome described by Mehta et al. No further reports of this syndrome have been described since 1989 and we believe this report may represent a second case of this syndrome.

Conclusion

The patient presented in this report has mild mental retardation, prominent ears, long philtrum, abnormally shaped teeth, prominent and everted lower lip, unilateral ptosis, craniosynostosis with developmental delay. His karyotype is normal. It is probably the second case of Mehta syndrome.

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