Dysmorphology report

Another case of microcephaly, facial clefting, and preaxial polydactyly

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Abstract

We describe a nine month old boy with failure to thrive, developmental delay, bilateral cleft lip and palate, and left preaxial polydactyly. The features are similar to those in a child described by Howard and Young¹ and may be the second case of a previously unknown syndrome.

Case report

This boy was the first born of healthy, non-consanguineous parents of French-Canadian and British ancestry. The pregnancy was complicated by polyhydramnios confirmed on ultrasound examination. He was delivered by emergency caesarean section for fetal bradycardia and heavy meconium seen at the time of artificial rupture of the membranes. Apgar scores were 1 at one minute and 4 at five minutes. He required intubation; assisted ventilation was required for 48 hours. Hypotension and pleural and peritoneal effusions were treated medically. His birth weight was 3260 g (25th centile), length was 52 cm (60th centile), and head circumference was 36 cm (60th centile). In addition to the bilateral cleft lip and palate, a pedunculated extra digit was noted on the lateral aspect of his left thumb.

At the age of 9 months, his weight was 6.3 kg (<5th centile), length was 68.5 cm (5th centile), and head circumference was 39 cm (much less than the 5th centile). He was globally developmentally delayed with increased tone in all four limbs, more severe in

the left arm. He reached for toys but he had difficulty grasping, with a continually fistled left hand. He was beginning to roll but was unable to sit independently. He was able to bear weight on his legs. Occasional myoclonic jerks were noted but no seizures were documented. Vision and hearing were normal.

The bilateral cleft lip was repaired. He had a broad nasal bridge and bilateral epicanthic folds with mild hypotelorism (figure). Palpation of his skull indicated overriding lambdoidal sutures. X rays of the skull showed no evidence of craniosynostosis. The left preaxial polydactyly was removed surgically and the right inguinal hernia was repaired.

Routine haematological and biochemical investigation was normal. He has a normal male karyotype, 46,XY. TORCH titres were negative. A CT scan at 7 months of age showed severe atrophy of the right

Front view of the patient's face.

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hemisphere with mild atrophy of the left hemisphere. A renal and abdominal ultrasound examination was normal.

Discussion

The findings in our patient differ from those in Meckel's syndrome as well as the lethal, probably autosomal recessive syndrome described by Váradi *et al*,² with polydactyly, cleft lip/palate or lingual lump, severe developmental delay, and early death.

Our patient has similar features to the patient described by Howard and Young¹ with microcephaly, facial clefting, and preaxial polydactyly. Our patient's history was complicated by neonatal asphyxia which may have contributed to the developmental delay and cortical atrophy seen on CT scan. It is likely that

prenatal effects were major factors in the development of the microcephaly and global motor and cognitive delay.

This condition may represent a true MCA syndrome. Inheritance cannot be established at this time in our patient, although recessive inheritance is most likely.

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- 1 Howard FM, Young ID. Unknown syndrome: microcephaly, facial clefting, and preaxial polydactyly. *J Med Genet* 1988;25:272-3.
- 2 Váradi V, Szabó L, Papp Z. Syndrome of polydactyly, cleft lip/palate or lingual lump, and psychomotor retardation in endogamic gypsies. *J Med Genet* 1980;17:119-22.