Case reports


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Note
After this report was submitted for publication, Dr M Seabright (personal communication) found full trisomy 9 syndrome by prenatal diagnosis in a fetus of a woman aged 42 years.

The hypertelorism microtia clefting syndrome

SUMMARY A single case of the hypertelorism microtia clefting (HMC) syndrome, a rare autosomal recessive condition, is reported.

Since the original report by Bixler et al1 of two sibs with hypertelorism, cleft lip and palate, and microtia, there have been few reports of the syndrome. Identical twins were documented by Schweckendiek et al2 but the syndrome has not yet emerged from being a 'provisional private syndrome'. Other cases are needed to establish the condition as important for the geneticist. The purpose of this paper is to add a case to the sparse literature.

Case report
The proband, now one month old, was the product of an uneventful pregnancy. He was delivered at 38 1/2 weeks and at birth he was seen to have a unilateral left-sided cleft lip, a cleft palate, a naevus centrally placed on his face (which was fading at one month), gross hypertelorism, narrow palpebral fissures, and a broad bifid nose (figure). Both pinnae were malformed and the external meati were stenosed. At one month he was clearly able to see. There was no cardiac murmur and the renal tract appeared normal. At 2 months he was behaving in a way appropriate for age.

Discussion
The two sisters described by Bixler et al1 had in addition to hypertelorism, microtia, and clefting, cardiac defects, mild retardation, and renal anomalies. Both sisters were below the 3rd centile for height and were mildly microcephalic. One had a bifid nose and the other a broad nasal tip. The ears were small and deformed and, in particular, the tragus and anterior-superior helix were abnormal in shape. The external auditory meati were absent and inner ear malformations occurred in both. The cardiac defects were an abnormality of the endocardial cushion in one and an ASD in the other. However, other members in this family also had cardiac defects, making interpretation difficult. The renal anomalies were mild, consisting of a pelvic kidney and crossed ectopia.

In the infant reported in this communication, hypertelorism was marked. As with the sisters described by Bixler et al1 the nose was broad and bifid and the cleft lip and palate was unilateral. The malformation of the ears involved the whole of the pinna but especially the helix. It was too early to
assess the child's developmental status and so far no obvious renal anomalies have been noted. The only important differential diagnosis is frontonasal dysplasia, but in that spectrum of conditions the ears are not severely deformed.

The lateral cleft syndromes, which include the first and second branchial arch syndromes, hemifacial microstoma, and Goldenhar syndrome, commonly have malformation of the ears but cleft lip and palate is unusual (7% of cases) and severe hypertelorism is not a feature.

Clinically the patient in this report and those in the report of Bixler et al have the same rare autosomal recessive condition referred to as the HMC syndrome.

The author is grateful to Dr Joan Slack, Consultant in Genetics, Royal Free Hospital, to Dr Stephen Ware, Consultant Paediatrician, Basildon Hospital for the referral of the patient, to the photographic department of Basildon Hospital for sending us the pictures, and to the photographic department, The Hospital for Sick Children, Great Ormond Street, for help with the processing of the photograph shown.

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References


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Congenital universal alopecia, mental deficiency, and microcephaly in two sibs

SUMMARY A brother and sister are reported who had congenital universal atrichosis, microcephaly, and mental retardation. Similar observations representing a rare nosological group are summarised. Heterogeneity is suggested. The pathogenesis of the individual syndromes is unknown.

This observation fits into a nosological group the first example of which seems to have been recognised by Moynahan in 1962. Its main feature, general alopecia (atrichosia), mainly congenital, is associated with various non-progressive neurological and sensory disorders. Several relevant cases reported so far show intrafamilial similarity but interfamilial variation and suggest heterogeneity. Therefore, the summary in the table of observations published by Richieri-Costa and Frota-Pessoa may be far from being final.

Case report

The patients, a brother and sister, were born in 1964 and 1965 to normal unrelated parents. A younger brother was unaffected. Birthweight was 2550 g and 2900 g, respectively, and length was 48 cm. Motor and particularly mental and speech development have been severely retarded. Heights at the ages of 7 and 6 and again at 15 and 14 years were on the 3rd centile, and head circumferences (46 cm) were below the 2.5th centile. Bone age was within the normal range and puberty was delayed. At the age of 16, the girl was still premenarchal. Single scalp hairs were present at birth in the male but soon fell out, as in the female who was born with 'normal looking' hair. Scalp and body hair never reappeared. Eyebrows and lashes were extremely scanty, and there was virtually no pubic or axillary hair (figure).

Microscopical examination of single hairs showed no specific abnormalities. Histological studies were not permitted. It is noteworthy that neither child has ever been seriously ill and that no seizures have been observed. An EEG was performed in the female and found to be normal; however, she suffers from a VSD. Aminoaciduria was normal. No particular dysmorphic signs were noted in either sib.

CASE REPORTS

Patients at the age of 16.
The hypertelorism microtia clefting syndrome

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doi: 10.1136/jmg.19.5.387

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