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*¹ 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.

M BARAITSER
Clinical Genetics Unit,
The Hospital for Sick Children,
Great Ormond Street,
London WC1N 3JH

References

- ¹ Bixler D, Christian JC, Gorlin RJ. Hypertelorism, microtia, and facial clefting. A newly described inherited syndrome. *Am J Dis Child* 1969;**118**:495-8.
- ² Schweckendiek W, Hillig U, Kruse E, Rodeck G, Wendt GG. HMC syndrome in identical twins. *Hum Genet* 1976;**33**:315-8.

Requests for reprints to Dr M Baraitser, Clinical Genetics Unit, The Hospital for Sick Children, Great Ormond Street, London WC1N 3JH.

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

Received for publication 22 February 1982

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 1962.¹ Its main feature, general alopecia (atrachosis), 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-Pessoa² 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

FIGURE Patients at the age of 16.

TABLE Summary of published observations of atrichosis associated with neurological disorders.

Authors	Mental deficiency	Microcephaly	Epilepsy (abnormal EEG)	Other features	Occurrence	Transmission
Mosavy ⁸	+	+	—	Short stature (?)	Sibs	AR
This observation	+	+	—	Short stature	Sibs	AR
Moynahan ¹	+	—	+	—	Sibs	AR
Perniola <i>et al</i> ⁹	+	—	(+)	—	Sibs	AR
Richieri-Costa and Frota-Pessoa ²	+	—	+	—	Sibs	AR
Shokeir ¹⁰	+	—	+	—	4 generations	AD
Del Castillo <i>et al</i> ¹¹	+	+	(+)	Papular lesions	Sibs	AR
Damste and Prakken ¹²	—	—	—	Papular lesions	Single	?
Loewenthal and Prakken ¹³	—	—	—	Papular lesions	Single	AR

apart from zygodactyly. Teeth and nails were normal. There was no evidence of reduced sweating.

Discussion

Universal congenital alopecia has rarely been reported even in textbooks and handbooks of dermatology.^{3,4} Familial occurrence in one or more generations suggests AR or AD inheritance. Since the pathogenesis of the disorder is unknown further delineation has not been possible. Associations with various disorders, all sharing a (neuro) ectodermal substrate, have been reported. One type includes sensorineural deafness and either hypogonadism⁵ or ectodermal dysplasia⁶ or spiny hyperkeratosis.^{2,7}

Associations with neurological disorders are summarised in the table, and the cases are subdivided according to particular clinical features, though they are not very specific. Our observation is similar to that of Mosavy.⁸ In all but one,¹⁰ AR inheritance is probable. Since, in the case of Moynahan,¹ three relatives were known to exhibit a minor degree or even temporary alopecia, variable expressivity should be kept in mind. The observation of Del Castillo *et al*¹¹ links these groups to cases with specific skin manifestations, but the inter-relationship is not yet understood. Biochemical studies in these patients have been unsatisfactory or negative when undertaken.

The collaboration of Dr E Haneke, Department of Dermatology, is acknowledged. Additional clinical examinations were performed in the Department of Pediatrics, University of Hamburg (Director Prof Dr Schulte).

R A PFEIFFER AND J VÖLKLEIN

*Institut für Humangenetik und Anthropologie,
Universität Erlangen-Nürnberg,
Bismarckstrasse 10, D-8520 Erlangen,
West Germany*

References

- Moynahan EJ. Familial congenital alopecia, epilepsy, mental retardation with unusual electroencephalograms. *Proc R Soc Med* 1962;**55**:11–2.
- Richieri-Costa A, Frota-Pessoa O. Atrichia, abnormal EEG, epilepsy and mental retardation in two sisters. *Hum Hered* 1979;**29**:293–7.
- Rook A, Wilkinson DS, Ebling FJQ. *Textbook of dermatology*. 2nd ed. Blackwell: Oxford, 1972.
- Gottron HA, Schoenfeld W, eds. *Dermatologie und Venerologie*. Vol III/2. Stuttgart: Thieme, 1959.
- Crandall B, Samec L, Sparkes RS, Wright SW. A familial syndrome of deafness, alopecia and hypogonadism. *J Pediatr* 1973;**82**:461–5.
- Freire-Maia N, Cat I, Rapone-Gaidzinski R. An ectodermal dysplasia syndrome of alopecia, onychodysplasia, hypohidrosis, hyperkeratosis, deafness and other manifestations. *Hum Hered* 1977;**27**:127–33.
- Morris J, Ackerman AB, Koblenzer PJ. Generalized spiny hyperkeratosis, universal alopecia and deafness. *Arch Dermatol* 1969;**100**:692–8.
- Mosavy SH. Universal alopecia and microcephaly in 4 siblings. *S Afr Med J* 1975;**49**:172.
- Perniola T, Krajewska G, Carnevale L. Le neuroectodermosi con alopecia o ipotricosi. *Acta Neurol (Napoli)* 1977;**32**:357–79.
- Shokeir MHK. Universal permanent alopecia, psychomotor epilepsy, pyorrhoia and mental subnormality. *Clin Genet* 1977;**11**:13–7.
- Del Castillo V, Ruiz-Maldonado R, Carnevale A. Atrichia with papular lesions and mental retardation in two sisters. *Int J Dermatol* 1974;**13**:261–5.
- Damste TJ, Prakken JR. Atrichia with papular lesions; a variant of congenital ectodermal dysplasia. *Dermatologica* 1954;**108**:114–21.
- Loewenthal LJA, Prakken JR. Atrichia with papular lesions. *Dermatologica* 1961;**122**:85–9.

Requests for reprints to Professor R A Pfeiffer, Institut für Humangenetik und Anthropologie, Universität Erlangen-Nürnberg, Bismarckstrasse 10, D-8520 Erlangen, West Germany.