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 (atrichosis), mainly congenital, is associated with various non-progressive neurological and sensory disorders. Several relevant cases reported so far show intrainfamilial 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.

FIGURE Patients at the age of 16.
Table  Summary of published observations of atrichosis associated with neurological disorders.

<table>
<thead>
<tr>
<th>Authors</th>
<th>Mental deficiency</th>
<th>Microcephaly</th>
<th>Epilepsy (abnormal EEG) Other features</th>
<th>Occurrence</th>
<th>Transmission</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mosavy</td>
<td>+</td>
<td>+</td>
<td>-</td>
<td>Short stature (?) Sibs AR</td>
<td>AR</td>
</tr>
<tr>
<td>This observation</td>
<td>+</td>
<td>+</td>
<td>-</td>
<td>Short stature Sibs AR</td>
<td>AR</td>
</tr>
<tr>
<td>Molyahan1</td>
<td>+</td>
<td>-</td>
<td>+ (±)</td>
<td>-</td>
<td>Sibs AR</td>
</tr>
<tr>
<td>Perniola et al8</td>
<td>+</td>
<td>-</td>
<td>+ (±)</td>
<td>-</td>
<td>Sibs AR</td>
</tr>
<tr>
<td>Richieri-Costa and</td>
<td>+</td>
<td>-</td>
<td>+ (±)</td>
<td>-</td>
<td>Sibs AR</td>
</tr>
<tr>
<td>Frota-Pessoa2</td>
<td>+</td>
<td>-</td>
<td>+</td>
<td>-</td>
<td>Sibs AR</td>
</tr>
<tr>
<td>Shokeir10</td>
<td>+</td>
<td>-</td>
<td>+</td>
<td>-</td>
<td>4 generations AD</td>
</tr>
<tr>
<td>Del Castillo et al11</td>
<td>+</td>
<td>+</td>
<td>(±)</td>
<td>Papular lesions Sibs AR</td>
<td>AR</td>
</tr>
<tr>
<td>Damste and Prakken12</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>Papular lesions Single</td>
<td>AR</td>
</tr>
<tr>
<td>Loewenthal and Prakken13</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>Papular lesions Single</td>
<td>AR</td>
</tr>
</tbody>
</table>

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,9 AR inheritance is probable. Since, in the case of Molyahan,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 al11 links these groups to cases with specific skin manifestations, but the interrelationship 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).

References

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