Acquired alopecia, mental retardation, short stature, microcephaly, and optic atrophy

R C M Hennekam, E G C M Renckens-Wennen

Abstract
We report on a female patient who had acquired total alopecia, short stature, microcephaly, optic atrophy, severe myopia, and mental retardation. A survey of published reports failed to show an identical patient, despite various similar cases.

Case report
The proband was the first child of a healthy 26 year old mother and non-consanguineous 37 year old father. The parents and sibs did not wear glasses, had normal heights, and showed no signs of an ectodermal dysplasia.

Pregnancy and delivery were uneventful and at birth no anomalies were seen. At the age of 5 years her hair started to fall out and she was completely bald by the age of 8 years. She had severely retarded psychomotor development. She had no specific illnesses, particularly no seizures or skin infections. Menarche was at 18 years, and she had regular menses thereafter. Physical examination at 42 years showed a friendly, retarded woman. Height was 150·3 cm (<3rd centile) and head circumference 50·1 cm (<3rd centile). She had fleshy nasal alae, deep nasolabial grooves, full lips, and downturned corners of the mouth (figure). There was no scalp hair, eyebrows, or eyelashes, virtually no pubic hair, but some axillary hair. Her skin, nails, breasts, and sweating pattern were normal. She had a partial dental prosthesis, probably because of former inadequate dental care.

The remaining teeth were normal. She had severe myopia bilaterally (−10 dioptres). Fundoscopy showed atrophic, pale discs. She had narrow hands with long fingers (total hand length 16·3 cm (3rd to 10th centile), middle finger length 7·1 cm (25th centile), and small feet (length 21 cm, 3rd centile). Chromosome analysis, using GTG banding, showed a normal female 46,XX karyotype. EEG, audiometry, and urinary metabolic screening gave normal results. Radiography of the skull showed no anomalies, especially no unerupted teeth.

Discussion
The present patient is compared with other relevant published cases in the table. Although close similarities can be seen, heterogeneity within this nosological group probably exists. Our patient resembles most the cases of Mosavy,1 Pfeiffer and Völklein,2 Baraitser et al,7 and Benke and Hajianpour.8 The alopecia was congenital in all these reports. The patients reported by Baraitser et al7 and Benke and Hajianpour8 did not have growth retardation, optic atrophy, or myopia. Schinzel13 and Van Gelderen14 each described a patient with mental retardation, alopecia, myopia, short stature, and microcephaly. Their patients had different facial features, contractures, and numerous skeletal anomalies. Optic atrophy together with growth retardation and acquired alopecia are features of the GAPO syndrome.15-18 However, patients with this condition are usually not retarded and have in addition pseudoanodontia, prominent scalp veins, and characteristic facial features. We conclude that our case probably has a different condition from the other reports. It represents possibly a distinct neuro-ectodermal disorder of unknown cause.

We thank Dr M Baraitser (London) and Dr R A Pfeiffer (Erlangen) for their help in studying this patient.

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Received for publication 13 March 1990.
Accepted for publication 2 April 1990.

3 Moyahan EJ. Familial congenital alopecia, epilepsy, mental
**Main differential diagnostic features compared with present patient.**

<table>
<thead>
<tr>
<th>Reference</th>
<th>No of patients</th>
<th>Mental retardation</th>
<th>Alopecia*</th>
<th>Short stature</th>
<th>Microcephaly</th>
<th>Epilepsy</th>
<th>Optic atrophy</th>
<th>Other features</th>
<th>Inheritance</th>
</tr>
</thead>
<tbody>
<tr>
<td>1 2</td>
<td>6</td>
<td>+</td>
<td>C</td>
<td>+</td>
<td>+</td>
<td>-</td>
<td>-</td>
<td>Deafness, stubby fingers, coarse features</td>
<td>AR</td>
</tr>
<tr>
<td>3 4</td>
<td>4</td>
<td>+</td>
<td>C,R</td>
<td>+/-</td>
<td>+</td>
<td>+</td>
<td>-</td>
<td>Periodontal disease</td>
<td>AR</td>
</tr>
<tr>
<td>5 6</td>
<td>14</td>
<td>+</td>
<td>C</td>
<td>-</td>
<td>-</td>
<td>+</td>
<td>-</td>
<td>Papular lesions</td>
<td>AR</td>
</tr>
<tr>
<td>7 8</td>
<td>6</td>
<td>+</td>
<td>C</td>
<td>-</td>
<td>+</td>
<td>+</td>
<td>-</td>
<td>Decreased fertility, nail abnormalities, sulphur deficient hair</td>
<td>AR</td>
</tr>
<tr>
<td>9</td>
<td>4</td>
<td>+</td>
<td>C</td>
<td>+</td>
<td>+</td>
<td>-</td>
<td>-</td>
<td>Turriccephaly, dysmorphic face, contractures, myopia</td>
<td>AR?</td>
</tr>
<tr>
<td>10</td>
<td>2</td>
<td>+</td>
<td>C</td>
<td>-</td>
<td>+</td>
<td>-</td>
<td>-</td>
<td>Pseudoanodontia, prominent scalp veins, dysmorphic face, umbilical hernia</td>
<td>AR</td>
</tr>
<tr>
<td>11</td>
<td>10</td>
<td>+</td>
<td>TN</td>
<td>+</td>
<td>+</td>
<td>-</td>
<td>-</td>
<td>Myopia, typical face</td>
<td>?</td>
</tr>
<tr>
<td>Present case</td>
<td>1</td>
<td>+</td>
<td>A</td>
<td>+</td>
<td>+</td>
<td>+</td>
<td>-</td>
<td></td>
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</tr>
</tbody>
</table>

*C=congenital or in first weeks of life, R=regrowing in infancy, TN=trichorrhexis nodosa, A=acquired.

†One case reported with borderline intelligence.17

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*J Med Genet* 1990 27: 635-636
doi: 10.1136/jmg.27.10.635

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