Yq— in a child with livedo reticularis, snub nose, microcephaly, and profound mental retardation

**Summary** A child with terminal deletion of the long arm of the Y chromosome (Yq—) presented with marked livedo reticularis, snub nose, microcephaly, short stature, and other dysmorphic features. He was profoundly mentally retarded. Most of the patients with Yq— have been reported as having varying dysmorphic features, mental retardation, and short stature. This child, in addition to the above, has livedo reticularis and microcephaly. He was of normal birthweight and, therefore, does not come into the syndrome of microcephaly, snub nose, livedo reticularis, and low birthweight dwarfism. Further information on Yq— should be obtained to ascertain if consistent patterns of abnormalities exist.

Cases with terminal deletion of the long arm of the Y chromosome (Yq—) have presented with a range of findings, from normal intelligence and normal male development, normal fertility, aspermia, varying dysmorphic features (major and minor), mental retardation, and short stature (table). In only one of these was the Yq— familial. The case described here, in addition to short stature, congenital abnormalities, and mental retardation, had marked livedo reticularis and microcephaly.

**Case report**

The proband (fig 1), born on 18.7.71 and seen at 5 years 7 months of age and at 8 years 7 months of age, was the youngest and third-born child. At the time of his birth, his father and mother were 26 and 27 years of age, respectively. The pregnancy and delivery were uncomplicated and the birthweight was 3200 g. Mild respiratory distress developed 24 hours after birth. His head circumference at birth was reported to be 35 cm. His head circumference at 10 months of age was 43 cm, and at 23 months of age


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### Table 378: Comparison of clinical features in the proband with those with Yq− in the literature.

<table>
<thead>
<tr>
<th>Reference</th>
<th>Normal male development</th>
<th>Y chromosomes of fathers or male sibs</th>
<th>Dysmorphic or unusual features</th>
<th>Mentally retarded</th>
<th>Short stature</th>
<th>Apparent breakpoint of Yq</th>
</tr>
</thead>
<tbody>
<tr>
<td>Langmaid and Laurence</td>
<td>+</td>
<td>+</td>
<td>Spina bifida occulta&lt;br&gt;Deep set eyes, bilateral frontal depression, low anterior hairline, Brushfield spots, small cavernous haemangiomas, hypoplastic 5th metacarpals, prominent chin, anteverision of auricles, short clubbed toes, short fingernails and toenails</td>
<td>−</td>
<td>+</td>
<td>del (Y) (q11)</td>
</tr>
<tr>
<td>Meisner and Inhorn</td>
<td>+</td>
<td>+</td>
<td>+</td>
<td>+</td>
<td>−</td>
<td>del (Y) (q11)</td>
</tr>
<tr>
<td>Telfer et al⁷</td>
<td>NS</td>
<td>NS</td>
<td>None</td>
<td>Slow: IQ 81</td>
<td>+</td>
<td>del (Y) (q11) or (Yp)</td>
</tr>
<tr>
<td>Nakagome et al⁶</td>
<td>+</td>
<td>+</td>
<td>Low nasal bridge, brachycephalic head, hypoplastic 5th metacarpals, unusual dermatoglyphs, livedo reticularis, puffy hands and feet.</td>
<td>+</td>
<td>+</td>
<td>No banding</td>
</tr>
<tr>
<td>Scherini et al⁵</td>
<td>NS</td>
<td>NS</td>
<td>Cleft lip, cleft palate, hypertelorism, Taussig-Bing syndrome</td>
<td>NS</td>
<td>+</td>
<td>del (Y) (q11)</td>
</tr>
<tr>
<td>Yunis et al³</td>
<td>+</td>
<td>+</td>
<td>None</td>
<td>+</td>
<td>+</td>
<td>del (Y) (q11)</td>
</tr>
<tr>
<td>Present case</td>
<td>+</td>
<td>+</td>
<td>Microcephaly, right ptosis, epicanthal folds, right Darwinian tubercle, small snub nose, hypoplastic 5th toenails, unusual dermatoglyphs, livedo reticularis, puffy hands and feet.</td>
<td>+</td>
<td>+</td>
<td>del (Y) (q11)</td>
</tr>
</tbody>
</table>

NS = Not stated

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**Figure 2**: GTG banded karyotype of proband, 46, X, del(Y) (q11). Arrow points to Yq−.
Case reports

FIG 3 QFQ banded metaphases of the proband (left) and proband's father (right). Arrows point to abnormal (left) and normal (right) Y chromosome, respectively.

it was 44.5 cm. He sat alone at 2 years of age. He stood and took a few steps with support at 8½ years of age. He was not toilet trained.

Physical examination at 8 years 7 months of age revealed he was below the 3rd centile for height (110 cm), weight (19.5 kg), and head circumference (47 cm). Physical findings included: a small head; ptosis of the right eyelid; epicanthal folds; a hyper-optic refractive error; a Darwinian tubercle on the right ear; a relatively small snub nose; diastasis recti; hypoplastic toenails on the fifth toes bilaterally; slight limitation of supination; puffy dorsum of the hands and feet; testes palpable in the inguinal canal; penis somewhat small, but not significantly so; and marked livedo reticularis, not related to cold, circulatory, or cardiac problems.

Dermatoglyphic studies revealed a markedly distal palmar triradius of both palms with some hypoplasia of the ridges. White lines were present. A small dimple was noted in the lateral portion of the distal palm crease, opposite the head of the fifth metacarpal.

The urine amino-acid pattern and thyroid studies were normal. X-rays of the hands, wrists, and skull were normal. Both the father and child were H-Y antigen positive.

Psychological studies indicated the child functioned in the profound range of mental retardation. On the Bayley Scales, a motor scale age of 11 months was obtained and the mental scale age scattered to 14.2 months. A social age of 14 months was obtained.

Chromosome studies indicated that the child had a 46,X,del(Y)(q11) karyotype by both GTG and QFQ banding techniques from a peripheral leucocyte culture (figs 2, 3, 4). Chromosome studies on his parents revealed normal karyotypes. A normal Y chromosome was found in his father (fig 3, 4).

FIG 4 Partial karyotype of Y and G group chromosomes from the proband (top) and proband’s father (bottom), stained with GTG banding.

Discussion

The small Y chromosome is reported as Yq—variant in newborn surveys. The incidence of Yq—variant has been reported as 0.39%, 0.11%, 0.82%, and 0.99%. In a survey of 13,751 male infants, Walzer and Gerald reported 52 Yq—variants (0.38%). In addition, they reported Yq—in one phenotypically normal child whose father carried a normal sized Y chromosome. There is no information in newborn surveys indicating whether the small Y chromosome was a familial inherited
variation (Yq—variant) or a true terminal deletion of the long arm of a Y chromosome (Yq—), as the fathers were not studied apart from one child reported by Walzer and Gerald. Therefore, the incidence of Yq—(deletion) and Yq—(variant) is unknown.

Infertility or fertility, normal or short stature, normal intelligence or mental retardation, and varying dysmorphic features have been observed in Yq—. This child presented with different dysmorphic features, short stature, and mental retardation. In addition, he also had marked lipoed reticulare and microcephaly. He was of normal birthweight and, therefore, does not come into the syndrome of microcephaly, snub nose, lipoed reticulare, and low birthweight dwarfism. The dysmorphic features, apart from short stature, described in the reported patients with Yq— are not similar and thus not of a specific phenotype. Whether they are polygenic, spontaneous, or related to Yq— is unknown. A differentiation should be made between Yq—(deletion) and Yq—(variant) to determine whether clinical findings are specific.

The proband recently died at 9 years 6 months of age from massive confluent bronchopneumonia. An aganglionic segment of the distal colon was noted at necropsy.

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References


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Gross congenital abnormality associated with an apparently balanced chromosomal translocation t(9;17)(q34;q11)

SUMMARY Gross mental and physical abnormality is described in an adult female who had some features similar to those of Ehlers-Danlos syndrome. There was no family history of the disorder. The patient also carried a balanced chromosomal translocation t(9;17)(q34;q11).

Surveys indicate that balanced chromosomal rearrangements are found in about 1.15% of newborn infants and that these rearrangements arise de novo in 0.02%. Therefore, a karyotypic imbalance is usually associated with mental retardation and multiple anomalies, whereas it is a general finding that persons with balanced chromosomal rearrangements are phenotypically normal. Thus, almost all the newborn infants with balanced de novo rearrangements found by the surveys were phenotypically normal. Nevertheless, some persons with balanced de novo chromosomal rearrangements have shown mental retardation or physical anomaly. Tharapel et al reviewed 25 cases from the literature and described a further six cases. Most were ascertained because of their physical or mental anomaly and it is therefore not possible to reach a reliable estimate of their incidence. However, the absence of such