Case reports

The acrocallosal syndrome in a Turkish boy

Memnune Yüksel, Mine Çalışkan, Gönül Oğur, Meral Özmen, Gülderen Dolunay, Selçuk Apak

Abstract
A 6 month old Turkish boy with the acrocallosal syndrome is reported. The patient, born to consanguineous, healthy parents, presented with macrocephaly, a prominent forehead, hypertelorism, polydactyly of the fingers and toes, severe motor and mental retardation, hypotonia, and absence of the corpus callosum. The mode of inheritance is discussed and our case is compared with previously reported cases of the syndrome.

The acrocallosal syndrome was first described by Schinzel in 1979. We report a male infant born to consanguineous parents with similar craniofacial dysmorphism, postaxial polydactyly of the hands and feet, severe motor and mental retardation, hypotonia, cyanotic spells, and absence of the corpus callosum.

Case report
The patient was first examined at 40 days of age because of polydactyly and dysmorphic features. He was the second child of healthy, young, Turkish parents who were first cousins. The first pregnancy had ended in spontaneous abortion during the first trimester.

Delivery was uneventful, birth weight was 3500 g, length 50 cm, and occipitofrontal head circumference 36 cm. Neonatally he was placed in an incubator because of several brief cyanotic spells. When first examined at 40 days of age the patient showed the following dysmorphic features (fig 1): large anterior fontanelle, high and prominent forehead, hypertelorism (ICD 35 mm, >97th centile), downward slanting palpebral fissures, epicanthic folds, a broad nasal bridge, short nose and philtrum, prominent ears, and an open mouth with protruding tongue. The hands showed bilateral postaxial polydactyly (fig 2). Scars from the postaxial supernumerary toes that were removed at birth could be seen. There was a right undescended testis. Neurological examination showed marked generalised hypotonia.

Cranial ultrasound and CT scan (fig 3) indicated absence of the corpus callosum. EEG showed asynchrony with abnormal background activity. X rays of

Figure 1 The patient at 4½ months of age.

Figure 2 Hands of the patient showing bilateral postaxial polydactyly.
the skeleton were normal except for postaxial polydactyly of the hands (fig 4); the supernumerary rays originated at the bases of the proximal fifth phalanges and contained two phalanges. Ophthalmological and cardiological examinations including echocardiography were normal as were GTG banded chromosomes (46,XY). The patient was followed up to 6 months of age. During this period he was admitted to hospital several times for cyanotic spells, hyperthermia, and respiratory tract infections.

At his last admittance to hospital with bronchopneumonia at 4½ months his weight was 6 kg (10th to 25th centile), height 62 cm (10th to 25th centile), and head circumference 43 cm (50th to 75th centile). Mental and motor retardation were obvious, with marked hypotonia and poor social contact. He stayed in hospital for around one and a half months. The family informed us later that the baby had died 20 days after his discharge from hospital. No necropsy was performed.

**Discussion**

Our patient increases the number of reported cases with the acrocallosal syndrome to 13.2-8 A comparison of the main features of the previous cases with our patient's is shown in the table. Total or partial absence of the corpus callosum, craniofacial dysmorphism, severe mental retardation, and postaxial polydactyly constitute the most consistent clinical features. All reported cases of the acrocallosal syndrome, apart from two families,7 8 were sporadic. It was previously assumed that the acrocallosal syndrome is caused by a fresh mutation of a dominant gene. Parental consanguinity in this and other reported cases4 and two affected cases in two families (sibs and first cousins) strongly suggest autosomal recessive inheritance. However, the rate of consanguinity in Turkey is among the highest (21-2%) in western countries7 and thus consanguinity is not such a strong argument for a recessive gene as it would be in a population with a low consanguinity rate.

### Major clinical features of our patient and previously reported cases

<table>
<thead>
<tr>
<th>Clinical findings</th>
<th>Published cases (n = 12)</th>
<th>Our case</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hypoplastic or absent corpus callosum</td>
<td>11/12</td>
<td>+</td>
</tr>
<tr>
<td>Postaxial polydactyly of hands</td>
<td>11/12</td>
<td>+</td>
</tr>
<tr>
<td>Postaxial polydactyly of feet</td>
<td>7/12</td>
<td>+</td>
</tr>
<tr>
<td>Duplication of phalanges of big toe</td>
<td>8/12</td>
<td>-</td>
</tr>
<tr>
<td>Craniofacial dysmorphm</td>
<td>12/12</td>
<td>+</td>
</tr>
<tr>
<td>Severe mental retardation</td>
<td>11/12</td>
<td>+</td>
</tr>
<tr>
<td>Cryptorchidism/hypospadias</td>
<td>2/6</td>
<td>+</td>
</tr>
<tr>
<td>Inguinal hernias</td>
<td>8/12</td>
<td>-</td>
</tr>
<tr>
<td>Cyanotic spells</td>
<td>3/12</td>
<td>+</td>
</tr>
<tr>
<td>Frequent infections</td>
<td>5/12</td>
<td>+</td>
</tr>
<tr>
<td>Hypotonia</td>
<td>9/12</td>
<td>+</td>
</tr>
</tbody>
</table>

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