A new camptodactyly syndrome

M BARAITSER

From the Clinical Genetics Unit, The Hospital for Sick Children, Great Ormond Street, London

SUMMARY A family is reported in which scoliosis and camptodactyly occurred in members over four generations. Additional features were torticollis, fusion of cervical vertebrae, and occasional limitation of joint movement in the upper limbs. Inheritance is autosomal dominant.

Flexion contracture of the proximal interphalangeal joints is known as camptodactyly. As an isolated malformation it affects one in 300 in the population and can be inherited as an autosomal dominant trait with variable expression. A small number of syndromes is known in which camptodactyly is a major feature. In this report, many members of a family are described who show camptodactyly, scoliosis, and torticollis, a combination that has not been previously reported.

Clinical features

The pedigree (fig 1) shows members over four generations with the main features transmitted in an autosomal dominant manner. The clinical signs present in each member are listed in table 1. All nine members had scoliosis to a variable degree. In the proband (V·3) it was noted in early childhood and, despite adequate treatment, progressed, whereas in the other members of the family the scoliosis varied from mild to severe. Four members needed an orthopaedic operation in an attempt to correct the deformity. Seven of the nine had camptodactyly. Torticollis was present in five and one required surgical treatment in infancy. At least five members had fusion of cervical vertebrae. One out of nine was severely mentally retarded and this boy (V·5) had all the major features of the syndrome. Four members were unable to extend their elbows totally but other joints were not involved.

Received for publication 9 April 1981

TABLE 1 Summary of clinical findings

<table>
<thead>
<tr>
<th>III·4</th>
<th>IV·1</th>
<th>IV·2</th>
<th>IV·4</th>
<th>IV·5</th>
<th>V·2</th>
<th>V·3</th>
<th>V·4</th>
<th>V·5</th>
</tr>
</thead>
<tbody>
<tr>
<td>Scoliosis</td>
<td>+</td>
<td>+</td>
<td>+</td>
<td>+</td>
<td>+</td>
<td>+</td>
<td>+</td>
<td>+</td>
</tr>
<tr>
<td>Camptodactyly</td>
<td>+</td>
<td>+</td>
<td>+</td>
<td>+</td>
<td>+</td>
<td>+</td>
<td>+</td>
<td>+</td>
</tr>
<tr>
<td>Torticollis</td>
<td>X-rays not available</td>
<td>Fusion of atlas on axis</td>
<td>Fusion of atlas on axis and L4/L5 on S1</td>
<td>Fusion of atlas on axis</td>
<td>Fusion of C2 on C3</td>
<td>Fusion of cervical vertebrae</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Bony fusion in cervical spine</td>
<td>Unable to straighten elbow</td>
<td>Unable to straighten elbow</td>
<td>Unable to straighten elbow</td>
<td>Unable to straighten elbow</td>
<td>Unable to straighten elbow</td>
<td>+</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Other joint involvement</td>
<td>Mental retardation</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
A new camptodactyly syndrome

Discussion

Camptodactyly, scoliosis, and torticollis are the main manifestations of a dominantly inherited syndrome which might occasionally include severe mental retardation. The 'whistling face' syndrome was considered, but this is characterised by camptodactyly with ulnar deviation ('windmill-vane')

FIG 2 Camptodactyly in (a) III\cdot4, (b) IV\cdot5, (c) V\cdot3, (d) V\cdot4, (e) V\cdot5.

FIG 3 Scoliosis in (a) III\cdot4, (b) IV\cdot1 and IV\cdot2, (c) V\cdot3, (d) V\cdot5.
### Table 2  Camptodactyly syndromes

<table>
<thead>
<tr>
<th>Inheritance</th>
<th>Facial features</th>
<th>Hands</th>
<th>Scoliosis</th>
<th>Other features</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cantú <em>et al</em>²</td>
<td>Recessive</td>
<td>Flat face, brachycephaly, wide forehead, myopia</td>
<td>Camptodactyly</td>
<td>---</td>
</tr>
<tr>
<td>Emery and Nelson³</td>
<td>Dominant</td>
<td>High forehead, flat nasal bridge, long philtrum, flat malar region</td>
<td>Camptodactyly</td>
<td>---</td>
</tr>
<tr>
<td>Goodman <em>et al</em>⁴</td>
<td>Recessive</td>
<td>Prominent forehead, facial asymmetry, ocular hypertelorism, small mouth, long philtrum</td>
<td>Camptodactyly</td>
<td>+</td>
</tr>
<tr>
<td>Gordon <em>et al</em>⁵</td>
<td>Dominant</td>
<td>Cleft palate</td>
<td>Camptodactyly</td>
<td>---</td>
</tr>
</tbody>
</table>

**FIG 4  Radiological findings.** *(a) IV-1, severe scoliosis of thoracic spine. (b) V-3, x-ray of neck showing fusion of upper cervical vertebrae. (c) V-3, x-ray showing severe scoliosis of thoracic spine.*

deformity), small mouth with inverted H-shaped dimpling of the chin, hypoplasia of the nasal alae, an immobile face, small eye openings, broad nasal bridge, and epicanthus. Neither scoliosis nor torticollis are prominent diagnostic criteria. In the present family, dysmorphic facial features were unremarkable except for one member who had a prominent jaw, wide forehead, and mild hypertelorism.

Other camptodactyly syndromes are listed in table 2. Two are dominantly inherited but are unlike that in the family described here in that neither is associated with scoliosis. Goodman *et al*⁶ and Cantú *et al*² described recessively inherited syndromes.

The main question that the proband asked was about risks of severe scoliosis and mental retardation in her offspring. The risk for an affected child is clearly 50% but the risk of mental retardation is probably small. There is, however, a considerable chance that an affected child could have a severe and progressive scoliosis needing operative treatment.

The author is grateful to Mr P Webb, FRCS, Consultant Orthopaedic Surgeon, The Hospital for Sick Children, Great Ormond Street, Mr M Pilcher, the Medical Illustration departments of The Royal National Orthopaedic Hospital, Great Portland Street, London, and The Hospital for Sick Children, and Mrs M Bravery for expert help with the preparation of this report.
A new camptodactyly syndrome

References

4. Goodman RM, Katznelson MBM, Katznelson A.


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

M Baraitser

doi: 10.1136/jmg.19.1.40

Updated information and services can be found at:
http://jmg.bmj.com/content/19/1/40

**Email alerting service**

*These include:*

Receive free email alerts when new articles cite this article. Sign up in the box at the top right corner of the online article.

**Notes**

To request permissions go to:
http://group.bmj.com/group/rights-licensing/permissions

To order reprints go to:
http://journals.bmj.com/cgi/reprintform

To subscribe to BMJ go to:
http://group.bmj.com/subscribe/