A new camptodactyly syndrome

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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.¹ 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

FIG 1 Pedigree of the family

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.

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TABLE 1 Summary of clinical findings

	III.4	IV.1	IV.2	IV.4	IV.5	V.2	V.3	V.4	V.5
Scoliosis	+	+	+	+	+	+	+	+	+
Camptodactyly	+	+	+	+	+	+	+	+	+
Torticollis	+	+	+	+	+	+	+	+	+
Bony fusion in cervical spine	X-rays not available	—	—	—	Fusion of atlas on axis	Fusion of atlas on axis and L4/L5 on S1	Fusion of atlas on axis	Fusion of C2 on C3	Fusion of cervical vertebrae
Other joint involvement	—	—	—	Unable to straighten elbow	Unable to straighten elbow	Unable to straighten elbow	—	—	Unable to straighten elbow
Mental retardation	—	—	—	—	—	—	—	—	+

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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·4, (b) IV·5, (c) V·3, (d) V·4, (e) V·5.

(a)

(b)

(c)

(d)

FIG 3 *Scoliosis* in (a) III·4, (b) IV·1 and IV·2, (c) V·3, (d) V·5.

TABLE 2 *Camptodactyly syndromes*

	<i>Inheritance</i>	<i>Facial features</i>	<i>Hands</i>	<i>Scoliosis</i>	<i>Other features</i>
Cantú <i>et al</i> ²	Recessive	Flat face, brachycephaly, wide forehead, myopia	Camptodactyly	—	Intrauterine growth retardation with proportionate dwarfism
Emery and Nelson ³	Dominant	High forehead, flat nasal bridge, long philtrum, flat malar region	Camptodactyly	—	Mental retardation in one
Goodman <i>et al</i> ⁴	Recessive	Prominent forehead, facial asymmetry, ocular hypertelorism, small mouth, long philtrum	Camptodactyly	+	—
Gordon <i>et al</i> ⁵	Dominant	Cleft palate	Camptodactyly	—	Club foot

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*⁵ 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.

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