References


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Congenital dislocation of the knees in a child with Down-mosaic Turner syndrome

SUMMARY A further case of Down-mosaic Turner syndrome is discussed. Both the cytogenetic and the dermatoglyphic data support the clinical diagnosis. The association with dislocated knees and the diagnosis of this polysyndrome at birth have not been reported before.

Approximately one in 200 liveborn children have a chromosomal abnormality. It appears that aneuploidy and structural chromosomal anomalies are among the most common causes of abnormal fetal development. Klinefelter syndrome with Down syndrome is the most frequent double aneuploidy recognised.

In this paper a case of Down-mosaic Turner syndrome associated with congenitally dislocated knees is presented. No previous report of a similar combination could be traced.

Case report

After an uneventful pregnancy, the proband, a Caucasian female, was born weighing 3 kg and measuring 50 cm in length. The father was 28 years old and the mother 26 years at the time of the child’s birth. The proband had mongoloid features (fig 1a), bilaterally dislocated knees (fig 1b), webbed neck, a transverse palmar crease, low hairline, oedema of both feet, hypoplastic nipples, and cubitus valgus. X-rays confirmed bilateral dislocation of her knees. She is now 5 years old and has both mental and physical retardation. She is the only child and her mother has not had any miscarriages.

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Cytogenetic investigations

A buccal smear showed a single Barr body in 14% of the cells examined. Chromosomes from lymphocyte cultures taken from the proband and both parents were examined in standard and trypsin-Giemsa banded preparations. The proband had 46 chromosomes in 37 of the 50 cells analysed. In each of these cells there was an extra chromosome 21.

FIG 1a  Proband at the age of 5 years.

FIG 1b  Congenitally dislocated knees (only one shown).
Case reports

while one of the X chromosomes was missing (fig 2a). The remaining 13 cells contained 47 chromosomes with an additional chromosome 21 (fig 2b). As 75% of the cells analysed had one X chromosome, it is not surprising that the child had phenotypic features of both Turner and Down syndromes. Her karyotype was 46,X,+21/47,XX,+21. Both parents had normal karyotypes.

DERMATOGLYPHS
The fingerprint patterns were ten ulnar loops (fig 3). The total finger ridge count, 103 ridges, was below the average figure of 126 ridges for English female controls. On the right palm there was an incomplete four-finger crease and a peripheral loop on interdigital area III. The hallucal area on the left sole showed an open field tibial arch pattern without associated e or f triradii. Similar features are observed in Down syndrome.

On the left palm there was a peripheral loop on area I. The sum of the left and right a-b ridge counts, 91 ridges, was above the mean value of 85 ridges for females with Down syndrome and of 86.8 ridges for XX controls. On both soles there was a proximal

FIG 2 (a) Of 50 cells examined, 37 showed this karyotype; (b) of 50 cells examined, 13 showed this karyotype.
looph II and a zygodactylous z triradius. Thenar/first interdigital patterns, high a-b ridge counts, an excess of II loops, and zygodactyly are peculiarities of Turner syndrome.

The palmar configurations of this case also showed features common to both Down syndrome and single X females. On the hypothenar area of each palm there was a whorl. It was associated with a r' triradius on the left palm and a r' on the right palm. Whorls occur on this region of the hand with a frequency of 26.2% in Turner syndrome and 11.9% in females with Down syndrome, but with an incidence of 2.0% in XX controls. A distal deviation of the axial triradii is observed in both Down and Turner syndromes.

**Discussion**

The incidence of Down syndrome in the live born is approximately 1 in 700 and that of Turner syndrome is about 1 in 5000. If one assumes that mechanisms governing the two syndromes are separate, the expected incidence of them occurring together would be 1 in 3.5 million. As each syndrome contributes significantly to abortion material there could be an increased chance of the combination being aborted. Up to the present only 17 liveborn cases could be traced in published reports. A review of these is set out briefly in the table.

From the table it is apparent that of all the cases of double aneuploidy, only one, that reported by Townes et al. had no X mosaicism. Furthermore, of all the fully documented cases only two, case 10 and the present case, have clinical appearances of both Down and Turner syndromes and these were substantiated by cytogenetic studies. The unique feature of the present case, however, is its diagnosis at birth, the earliest diagnosis made previously being at 2 months.

The karyotype 46,X,+21/47,XX,+21 probably arose through a somatic error involving the X chromosome in a patient trisomic for chromosome 21 at conception. If this were true then there should be no excess risk of Turner syndrome in further

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**TABLE**

- **Previously described cases of Down-Turner polysyndrome in chronological order**

<table>
<thead>
<tr>
<th>Case</th>
<th>Patient report</th>
<th>Chromosome constitution</th>
<th>Clinical appearance</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Hanhardt (1960)</td>
<td>Not recorded</td>
<td>Down</td>
</tr>
<tr>
<td>*2</td>
<td>Mendis et al (1962)</td>
<td>46,XO,G+47,XX,G+</td>
<td>Down</td>
</tr>
<tr>
<td>*3</td>
<td>Van Wijck et al (1964)</td>
<td>46,XO,G+47,XX,G+</td>
<td>Down</td>
</tr>
<tr>
<td>*4</td>
<td>Root et al (1968)</td>
<td>46,XO,G+47,XX,G+</td>
<td>Down</td>
</tr>
<tr>
<td>*5</td>
<td>Zergottern and Hoefnagel (1964)</td>
<td>46,XO,G+47,XX,G+ /48,XXX,G+</td>
<td>Down</td>
</tr>
<tr>
<td>6</td>
<td>Candella et al (1965)</td>
<td>46,XX/46,XX,+G</td>
<td>Down</td>
</tr>
<tr>
<td>7</td>
<td>Candella et al (1966)</td>
<td>46,X,+G</td>
<td>Down</td>
</tr>
<tr>
<td>8</td>
<td>Pfeiffer et al (1968)</td>
<td>46,XX,21+47,XX,21+</td>
<td>Down</td>
</tr>
<tr>
<td>*9</td>
<td>Grosse et al (1971)</td>
<td>46,XX/45,XX,21+</td>
<td>Down/Turner</td>
</tr>
<tr>
<td>*10</td>
<td>Mikel'saar et al (1973)</td>
<td>47,XX,21+47,XXp-q-.21+</td>
<td>Down/Turner</td>
</tr>
<tr>
<td>12</td>
<td>Villaverde et al (1975)</td>
<td>Not recorded</td>
<td>Down/Turner</td>
</tr>
<tr>
<td>13</td>
<td>Villaverde et al (1976)</td>
<td>Not recorded</td>
<td>Down/Turner</td>
</tr>
<tr>
<td>14</td>
<td>Townes et al (1968)</td>
<td>45,XX/46,XX,21+</td>
<td>Down</td>
</tr>
<tr>
<td>15</td>
<td>Singh et al (1977)</td>
<td>45,XX/46,XX,21+</td>
<td>Down</td>
</tr>
<tr>
<td>16</td>
<td>Singh et al (1978)</td>
<td>45,XX/46,XX,21+</td>
<td>Down</td>
</tr>
<tr>
<td>17</td>
<td>Singh et al (1979)</td>
<td>47,XX,+G/46,XX,+G</td>
<td>Down</td>
</tr>
</tbody>
</table>

**Lack of uniformity in cytogenetic terminology is the result of differences in original reports.**

*Cases from Townes et al.*

Case 8 quoted by Mikel'saar et al.*

Case 1 quoted by Villaverde et al.*

Cases 6 and 7 quoted by Singh et al.*
children. The recurrence of Down syndrome is probably 1 in 100 and will increase with age.

The fact that clinical features of the two syndromes coexist in this patient shows the relative autonomy of these chromosomes in the determination of the processes of morphogenesis.

Congenital dislocation of the knees is 80 times less common than congenital dislocation of the hips. The congenital anomalies most frequently associated with it are dislocated hips and talipes equinovarus. Except in Larsen syndrome, there does not appear to be a hereditary basis.

Skeletal abnormalities such as short metacarpals, hypoplasia of the middle fifth phalanx, and clinodactyly are seen in Down syndrome, whereas cubitus valgus, medial tibial exostosis, and short fourth metacarpal are seen in Turner syndrome. There is thus a possibility that the association of the knee abnormality with the Down-mosaic Turner knee syndrome may not be a chance occurrence, as other skeletal defects do occur in the two syndromes.

I wish to thank Dr Saldaña-Garcia of Harperbury Hospital, Radlett, Herts, for his invaluable advice in the preparation of this manuscript, particularly in the analysis of the dermatoglyphs. I would also like to thank Dr M Super of the Department of Genetics, Royal Manchester Children’s Hospital for his constructive criticism and support.

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A new probably autosomal recessive cardiomeiolic dysplasia with mesoaxial hexadactyly

SUMMARY A distinct probably autosomal recessive syndrome was ascertained in a 17-year-old boy and his deceased sister. The main features were cardiac dysplasia, peculiar facies, central bilateral (mesoaxial) hexadactyly, synmetacarpalia, short stature, ocular torticollis, and delayed puberty.

We describe a patient with multiple congenital anomalies, some of which were indirectly ascertained in his deceased sister, suggesting autosomal recessive inheritance.

Case report

The proband, born in 1962, was the product of a term gestation and normal delivery. Birthweight and length were not recorded; however, he was smaller than his normal sibs. Bilateral hexadactyly, broad right hallux, acrocyanosis, the presence of teeth, and dyspnoea were observed at birth. Psychomotor retardation was evident from early infancy and he tilted his head to the left when he started walking at about 4 years of age. Excision of the supernumerary right finger was performed at 10 years of age. Physical examination at 13½ years (fig 1) showed delayed development, a weight of 28.7 kg, a height of 134 cm (both below the 3rd centile), an upper-to-lower-segment ratio of 0.97 (66/68), an arm span of 127 cm, and a head circumference of 53 cm. Other features included normocephaly, wide and flat forehead, hypotrichosis of the eyebrows distally (ulerythema ophryogenes), telecanthus, ocular torticollis as a result of alternate exophoria-tropia, a winking tick, long prominent nose with lateral bossing, tented nares, lateral pits in the nasal alae, short philtrum, macrostomia, everted lower lip, downturned mouth, multiple dental diastemata and malocclusion, large pinnae, prominent anhelix, two right Darwinian tubercles, short neck, prominent trapezius muscles, cylindrical thorax, hypoplastic nipples, underdeveloped external genitalia, normal spine, mesoaxial hexadactyly in both hands, digital clubbing and cutaneous syndactyly between digits 2 and 3, thenar and hypothenar hypoplasia, genu and pes valgus, right hallux polysyndactyly, and mild flattening of toe nails. Dermatoglyphic analysis did not reveal extra triradii at the base of the supernumerary fingers.

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References

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