THE "SMALL PATELLA" SYNDROME

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Twelve closely related members of one family were found to have small or absent patellae. Seven of these patients also had abnormalities of the pelvic girdle and upper femora. There was a resemblance to the nail–patella syndrome in the patellar abnormalities but not in the associated conditions, and it would therefore appear that the syndrome described is a new variety of congenital abnormality of the knee.

A syndrome combining abnormalities of the patella and of the pelvic girdle has been found in twelve members of one family. This combination differs from other conditions of the knee which have a strong familial history.

DESCRIPTION OF THE FAMILY

Twelve members of the family were found to be affected: nine were interviewed by us in 1975 and the relevant information for the other three was kindly supplied by Dr J. H. Renwick, who had examined them while investigating the genetics of the nail–patella syndrome (Renwick 1956b). Of the remaining members of the family, twenty-one closely related individuals were examined; past hospital records and radiographs of those affected individuals who had had previous operations on the knee were reviewed. The significant abnormalities of the twelve patients are summarised in Table I.

In all cases the patellae were either small or absent (Table I) and, when present, the patella was laterally placed or dislocated (Figs. 1 to 4). Examination of the knees showed no predilection for genu varum or valgum. There was radiological evidence of abnormalities of the pelvic girdle and upper femora in seven of the affected individuals; of the remaining five, radiographs of the pelvis were not available for three and the other two were too young (seven years of age and eighteen months respectively) for radiological assessment to be made.

The abnormalities included coxa vara or valga with buttressing of the femoral neck and apparent hypoplasia of the lesser trochanter. There were also abnormalities of the ischiopubic junction, varying from unusually prominent ossification (Fig. 5) to defective formation (Fig. 6). Both sides of the pelvic girdle were equally affected. None of the patients in whom these abnormalities were found had any symptoms referable to the pelvic girdle.

Table I. Details of the twelve members of the family with the small patella syndrome

<table>
<thead>
<tr>
<th>Pedigree number</th>
<th>Date of birth</th>
<th>Sex</th>
<th>Patellar abnormalities</th>
<th>Abnormalities of the pelvic girdle</th>
</tr>
</thead>
<tbody>
<tr>
<td>III.32</td>
<td>1920</td>
<td>F</td>
<td>Rudimentary right patella. Absent left patella</td>
<td>Not known</td>
</tr>
<tr>
<td>III.33</td>
<td>1923</td>
<td>F</td>
<td>Right patella small and dislocated. Left patella small but central</td>
<td>Present</td>
</tr>
<tr>
<td>III.36</td>
<td>1916</td>
<td>M</td>
<td>Bilateral small laterally placed patellae. One episode of dislocation of patella at 20 years of age</td>
<td>Present</td>
</tr>
<tr>
<td>III.38</td>
<td>1920</td>
<td>M</td>
<td>Right patella small, bipartite, central. Left patella, small, multipartite, laterally placed</td>
<td>Not known</td>
</tr>
<tr>
<td>IV.6</td>
<td>1937</td>
<td>M</td>
<td>Bilateral absent patellae. Genu valgum. Multiple operations between 10 and 17 years of age including bilateral supracondylar femoral osteotomies and tendon transfers</td>
<td>Not known</td>
</tr>
<tr>
<td>IV.7</td>
<td>1944</td>
<td>F</td>
<td>Rudimentary right patella. Absent left patella</td>
<td>Present</td>
</tr>
<tr>
<td>IV.8</td>
<td>1943</td>
<td>M</td>
<td>Right patella small and laterally placed. Left patellectomy and patellar tendon transfer for recurrent dislocation of a small patella at 11 years of age</td>
<td>Present</td>
</tr>
<tr>
<td>IV.9</td>
<td>1945</td>
<td>F</td>
<td>Bilateral small and laterally placed patellae. Recurrent episodes of dislocation. No treatment</td>
<td>Present</td>
</tr>
<tr>
<td>IV.12</td>
<td>1945</td>
<td>M</td>
<td>Bilateral patellectomy and tibial tubercle transfer at 7 years of age for bilateral recurrent dislocation of rudimentary patellae</td>
<td>Present</td>
</tr>
<tr>
<td>IV.14</td>
<td>1951</td>
<td>M</td>
<td>Congenital absence of patellae. No dislocation of tendon</td>
<td>Present</td>
</tr>
<tr>
<td>V.8</td>
<td>1968</td>
<td>F</td>
<td>Bilateral small dislocated patellae</td>
<td>Too young to tell</td>
</tr>
<tr>
<td>V.9</td>
<td>1974</td>
<td>F</td>
<td>Patellar nuclei small for age and laterally placed</td>
<td>Too young to tell</td>
</tr>
</tbody>
</table>


Requests for reprints should be sent to Mr J. E. Scott.
Other miscellaneous abnormalities in the affected members of the family included four individuals with flat feet, two with significant syndactylyism of the toes, one haemophiliac and one individual with congenital deafness. There were no other skeletal abnormalities, in particular no iliac horns, no deformities of elbow or nail and no generalised laxity of the joints. Investigation of the blood groups was not performed on enough members of the family for deductions to be made about any possible genetic association with the ABO locus.

DISCUSSION

The syndrome described in this family appears to differ from other conditions of the knee which are associated with a strong family history. Congenital lateral dislocation of the patella is a rare condition which occurs shortly after birth and is caused by displacement of the extensor mechanism of the knee. Recurrent or habitual dislocation of the patella is much more common and occurs during childhood or adolescence; the commonest cause is an abnormally high patella (Anderssen 1958) although there may be other anomalies such as failure of development of the lateral femoral condyle (Smillie 1974) or an abnormal attachment of the iliotibial tract (Jeffreys 1963) and often there is an associated genu valgum. There is a familial tendency in congenital dislocation of the patella (De Palma 1954; Green and Waugh 1968) and more commonly in recurrent dislocation of the patella (Carter and Sweetnam 1958; Bowker and Thompson 1964). Familial absence of the patella has only been described twice: Kutz (1949) reported one case, and Bernhang and Levine (1973) reported two cases.

In none of the above conditions have other associated skeletal or soft tissue anomalies been described. However, the patellar abnormalities in the affected members of this family resemble those of the nail–patella syndrome, in which the patella may be
Fig. 7
Full pedigree of the family.
hypoelastic or absent, and there may be recurrent dislocation of the patella with characteristically no predilection for genu varum or valgum. The nail–patella syndrome was first described by Sedgewick (quoted by Little 1897). The clinical signs described by Osterreicher (1931) and by Turner (1933) included iliac horns, abnormalities of the elbow and dystrophy of the nails. A considerable number of other associated anatomical defects have been described and have been reviewed by Silverman, Goodman and Cuppage (1967).

The genetic linkage between the single defective gene locus of the nail–patella syndrome and ABO blood groups was described by Renwick and Lawler (1955); and the characteristics of an autosomal dominant gene with 100 per cent penetrance were described by Renwick (1956a). The small patella syndrome is the manifestation of an autosomal allele in the heterozygous state. The penetrance is complete for the patellar abnormalities but not for the ischiopubic changes. From the complete pedigree (Fig. 7), in which the nine sibs of II.11 and their descendants seem to be free of this lesion, it would appear that the allele was newly mutant in II.11 or alternatively that II.11 had a different father from that of her sibs.

We are extremely indebted to Dr J. H. Renwick (Reader in Human Genetics and Population Teratology, London School of Hygiene and Tropical Medicine) for his guidance on the genetic aspects of this syndrome and also for his permission to reprint the majority of the family tree from his London Ph.D. Thesis (1956b). We are also grateful to Dr H. Shawdon, Consultant Radiologist, Central Middlesex Hospital, for his help with the radiological aspects of this syndrome.

REFERENCES


