A kindred of seven affected individuals in three generations is described with autosomal dominant inheritance of bilateral five-fingered hands, pedal polydactyly with syndactyly and agenesis of the tibia and of the lower end of the radius.

Aplasia of the tibia associated with polydactyly, with or without syndactyly, and five-fingered hands was described in 1915 by Werner and in 1918 by Davidson. Reber (1968), Eaton and McKusick (1969), Pashayan et al (1971) and Temtamy and McKusick (1978) described similar cases with a familial incidence and Lamb, Wynne-Davies and Whitmore (1983) reported 15 affected members in five generations, strongly suggesting autosomal dominant inheritance with variable expressivity.

Figure 1 – The maternal grandfather aged 65 years showing five-fingered hands, hypoplastic right radius, absent tibia and pedal polydactyly with syndactyly. His grandson aged 11 has five-fingered hands and pedal polydactyly with syndactyly. The boy is the elder brother of the children in Figure 2. Figure 2 – The three children are unable to stand or walk while the mother has five-fingered hands and pedal polydactyly with syndactyly.

Radiograph of upper limb showing a five-fingered hand with absence of the lower end of the radius and ulnar hypertrophy.
We have examined a family of seven affected individuals in three generations. Partial agenesis of the distal radius was also present in some of them.

### CLINICAL AND GENETIC FINDINGS

Table I lists the anomalies of each patient. One member of the previous generation was said to be affected but she had died.

Bilateral five-fingered hands and absence of the thumb were seen in all cases (Figs 1 to 3). Pedal polydactyly with syndactyly of the big toes occurred in all seven patients (Figs 1 and 2). Six metatarsals were present (Fig. 4).

Complete agenesis of the tibia was seen in four patients (Figs 1, 2 and 4), and was bilateral in three. The affected leg was very short and rotated with contracture at the knee and ankle. There was rigid inversion and adduction of the foot. Proximal migration of the head of the fibula and hypoplasia of the distal femur were apparent. The fibula showed marked hypertrophy with lateral bowing. Agenesis of the

<table>
<thead>
<tr>
<th>Case</th>
<th>Age (yr)</th>
<th>Gender</th>
<th>Deformity</th>
<th>Lower limb</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>65</td>
<td>M</td>
<td>Bilateral five-fingered hands and absent lower end of radius (R)</td>
<td>Absent tibia and patella (R), bilateral polydactyly, syndactyly, six metatarsals and equinovarus foot (R)</td>
</tr>
<tr>
<td>2</td>
<td>35</td>
<td>F</td>
<td>Bilateral five-fingered hands</td>
<td>Bilateral polydactyly, syndactyly and six metatarsals</td>
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<tr>
<td>3</td>
<td>14</td>
<td>F</td>
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<td>Bilateral polydactyly, syndactyly and six metatarsals</td>
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<tr>
<td>4</td>
<td>11</td>
<td>M</td>
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<td>Bilateral polydactyly, syndactyly and six metatarsals</td>
</tr>
<tr>
<td>5</td>
<td>7</td>
<td>M</td>
<td>Bilateral five-fingered hands</td>
<td>Bilateral absent tibiae and patellae, polydactyly, syndactyly, equinovarus feet and six metatarsals</td>
</tr>
<tr>
<td>6</td>
<td>5</td>
<td>M</td>
<td>Bilateral five-fingered hands and absent lower end of the radii</td>
<td>Bilateral absent tibiae and patellae, polydactyly, syndactyly, equinovarus feet and six metatarsals</td>
</tr>
<tr>
<td>7</td>
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<td>M</td>
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<td>Bilateral absent tibiae and patellae, polydactyly, syndactyly, equinovarus feet and six metatarsals</td>
</tr>
</tbody>
</table>

Fig. 4
Radiograph of the lower limbs, showing bilateral absence of the tibiae and patellae, proximal migration of the head of the fibula and six metatarsals and phalanges.

Fig. 5
Diagram of the family tree.
lower one-third of the radius was present in three patients (Figs 1 and 2), unilateral in one and bilateral in the others, with hypertrophy and bowing of the ulna and a valgus hand (Fig. 3).

**DISCUSSION**

This family (Fig. 5) illustrates the autosomal dominant mode of inheritance of this syndrome with variable expressivity. Partial radial agenesis has not been described before in such patients.

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**References**


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