BILATERAL APLASIA OF THE TIBIA, POLYDACTYLY
AND ABSENT THUMB IN FATHER AND DAUGHTER

H. PASHAYAN, F. C. FRASER, J. M. MCINTYRE and J. S. DUNBAR, MONTREAL, CANADA

From the Departments of Medical Genetics, Orthopaedics and Radiology,
the Montreal Children's Hospital

The syndrome of bilateral aplasia of the tibia, polydactyly with or without syndactyly, and absence of the thumbs, was first reported in 1915 by Werner in a twenty-year-old woman in her sixth month of pregnancy who requested an abortion because the pregnancy was causing her difficulties. The patient had no thumbs and six fingers, each with a metacarpal bone. There were eight toes on the left foot and seven on the right. The lower limbs were very short and there was aplasia of the tibiae; movement was greatly reduced at the "knee-ankle" joint. She delivered a normal baby at term. There were no affected relatives.

In 1918 Davidson reported the second case, in a two-year-old baby. The patient had no thumbs and five well formed fingers of equal length, each with a metacarpal bone. He was able to use both hands well. Each foot had eight formed toes. The lower limbs were very short and there was a complete absence of the tibiae; the fibulae were present but articulation was faulty at both ends. The lower legs were unstable and the child walked with difficulty by inverting his feet and bearing weight on the outer sides of the legs. There were no affected relatives.

Fifty years later Reber (1968) described the first familial case. His propositus was an eight-year-old boy with bilateral aplasia of the tibiae, polydactyly of feet, and ten fingers with three phalanges each. The father also had ten fingers but no thumbs. Two fourth degree relatives had ectrodactyly, but were not themselves related. A "secondary case" with heptadactyly was only an eighth degree relative of the proband. These defects in distant relatives are probably unrelated to those of the proband and his father. A second familial case was reported by Eaton and McKusick (1969), who described a man with absent thumbs and pre-axial polydactyly of the feet, but with normal tibiae. His two daughters and one granddaughter showed polydactyly of the toes, hypoplastic tibiae, absent patellae, absent thumbs and polydactyly of the fingers. The findings in the above two families strongly suggest the existence of an autosomal dominant gene with variable expressivity, causing absent thumbs and manual polydactyly, with or without varying degrees of tibial hypoplasia and pedal polydactyly. This paper reports a case of virtually identical expression of the syndrome in father and daughter.

CASE REPORTS

Case 1. The daughter—The patient was a woman of French Canadian and Irish American descent, born to parents who were unrelated. Except for the father, there were no limb abnormalities among ten uncles and aunts, the grandparents, thirty first cousins or any other relative. One maternal first cousin was said to have no control of movement of the lower limbs and to be unable to walk, but was said not to have bone defects. The mother and father were twenty-five and twenty-nine years old respectively when the proband was born. She had a younger sister who was free of abnormalities.

At twelve weeks' gestation the mother fractured her left tibia in a car accident. The labour was unremarkable and the delivery was by breech. The baby's birth weight was 2,420 grammes.

The patient was seen periodically at the Montreal Children's Hospital throughout her childhood, and at seventeen years of age was recalled for re-examination. She was a good humoured, attractive looking girl with a rather awkward gait. Thanks largely to the good
sense of her mother she was psychologically well adjusted, accepting her handicap as a challenge. She was being trained to become a mechanical artist and a sample of her work seen by the authors showed precision and neatness. She could ski, drive a car and dance. She did, however, have difficulty in skating on ice.

![Fig. 1](image1)

**Fig. 1**
Case 1—The dorsal (Fig. 1) and palmar (Fig. 2) aspects of the hands, showing the five fingers and no thumbs.

![Fig. 2](image2)

**Fig. 2**

![Fig. 3](image3)

**Fig. 3**
Case 1—Anterior (Fig. 3) and posterior (Fig. 4) views of the legs which are mostly femur. The ankles are grossly abnormal and the scars of the amputated toes can be seen.

![Fig. 4](image4)

**Fig. 4**

Her height was 128·5 centimetres, her sitting height was 89·5 centimetres and her span was 152·5 centimetres. Each hand had five fingers, each with three phalanges and a metacarpal bone (Figs. 1 and 2). Opposition of the first to the second digit was not possible. Radiological
examination showed partial fusion of some of the carpal bones, and only one trapezium—or trapezoid—bone on each side. The head, cardiovascular, respiratory and gastro-intestinal systems, and secondary sexual characteristics were normal.

The lower limbs (Figs. 3 and 4) were extremely short (39 centimetres) ending with a knee joint with very little movement, a rudimentary tibia and an ankle joint with no medial malleolus. Most of the movement was at the ankle joint. She had had amputations of the supernumerary five toes—three on the right and two on the left—but this was the only surgery done. The mother did not want her to have amputation of the feet, or to wear prostheses like her father, even though it would bring her up to normal height. A diagram of the feet drawn before the amputation of the toes showed eight toes on the right foot and seven toes on the left (Fig. 5). Radiographs taken at the age of four years showed six metatarsal bones on each foot, with eight toes on the right and seven on the left, some of them partly fused.

Creases 10 centimetres long were seen on the postero-lateral aspect of the legs, resembling healed scars. They appear to mark the region of the interosseous membrane between the tibia and fibula.

The dermatoglyphic patterns on the digits were five radial loops and five ulnar loops. The first digit showed a radial loop on the left hand and an ulnar loop on the right hand. A triradius was present at the base of each digit, but there were no triradii or patterns on the

FIG. 5
Diagram of the extra toes of Case 1 before amputation. The arrows show the toes that had no active movements.

FIGS. 6 AND 7
Case 1. Figure 6—Radiograph of the left hand. Figure 7—Radiograph of the legs.

FIG. 7

The dermatoglyphic patterns on the digits were five radial loops and five ulnar loops. The first digit showed a radial loop on the left hand and an ulnar loop on the right hand. A triradius was present at the base of each digit, but there were no triradii or patterns on the
palms. It is interesting that radial loops on the left thumb are rare in the general population (less than 0.5 per cent) (Holt 1968) and that the thumb does not normally have a triradius at its base. Thus the first digit in our case appears to be a finger rather than a thumb, on dermatoglyphic as well as anatomical grounds. Surgical removal of a number of toes had distorted the hallucal patterns.

Radiographic examination (Figs. 6 and 7)—A skeletal survey done at the age of seventeen years showed that the skull was normal, as was the vertebral column with the exception of a slight and probably insignificant thoraco-lumbar scoliosis convex to the right. The humeri were thick and rather massive, the distal end of the right humerus was slightly blunted and the head of the radius reciprocally broadened. The femora showed a moderate degree of coxa valga. Tiny and laterally displaced patellar ossification centres were present.

The tibiae were short and thick, about 6 centimetres in length and broader than the femora. The fibular heads were displaced upwards and situated laterally to the femoral shafts, as a result of unhindered growth in length of the fibulae. At an earlier age the fibular heads were opposite to the knees in a relatively normal situation. The distal end of each fibula was lower in relation to the foot than would normally be expected.

The tarsal bones showed an abnormal coalition between the talus and the navicular bone, and the talus and the calcaneus bilaterally. The calcaneo-cuboid joint was present, articulating with hypertrophied fourth and fifth metatarsals. Each first metatarsal was hypoplastic. There were five toes remaining on each side but the medial left and right toes were smaller than a normal great toe and had three phalanges instead of two.

Case 2. The proband’s father—The father was described by his wife as having had hands identical to those of his affected daughter. His mother and father were twenty-five and twenty-nine years old respectively when he was born. He had been very short and had had bilateral absence of the tibiae and polydactyly of the toes. His feet had been amputated in childhood. He had died of cancer of the rectum and the liver at forty-one years of age.

Radiographs taken in 1953 of his upper limbs showed five fingers and no thumbs on each side; on the right side the trapezium and trapezoid bones were fused. The scaphoid, lunate and triquetral bones were fused. There was almost complete synostosis of the entire radius and ulna; a small gap was present at about the junction of the middle and uppermost thirds. The fourth left metacarpal bone was greatly shortened and broadened, and there was a deformity of the proximal phalanx of the fourth finger which appeared to be partly segmented longitudinally in its distal half.

COMMENTS

Several theories, of varying degrees of credibility, have been advanced to account for congenital absence of the tibia (Young 1904, Gaensler 1914, Ferguson and Scott 1952). In the familial cases, trauma, virus, mechanical pressure and nutritional deficiency can be discounted as etiological factors. The older literature cites “maternal impressions” as a possible cause, and the proband, whose mother had a tibial fracture during pregnancy, might have been cited as a pertinent example if it were not that her father was affected. Amniotic adhesions resulting from early trauma are often invoked as a possible explanation of the defect (Young 1904, Warkany and Nelson 1941), particularly in view of the scars often seen overlying the lesion. In the present case the proband inherited the gene responsible for the defect from her father, and the lesion resulting in tibial absence must clearly have been in the tissues of the leg. If the scars were caused by amniotic adhesions these must have been a secondary result of the lesion, not a cause of it. The mechanism by which a mutant gene causes both aplasia specifically of the tibia and abnormal differentiation of the digits remains tantalisingly unclear.

The familial cases reported show a father to son (Reber 1966), father to daughter and daughter to daughter (Eaton and McKusick 1969), and in our case father to daughter transmission. Reber’s family and the first and second generation in Eaton’s family show
variable expressivity of the trait. Our family, and the cases in the second and third generation in Eaton's report, show the full expression of the syndrome. Even though the number of familial cases is very limited, the available evidence indicates that the inheritance of this syndrome is autosomal dominant, and the affected individuals therefore have a 50 per cent chance of transmitting the gene to their offspring. It is important to recognise that this applies to sporadic as well as familial cases if they represent fresh mutations. The father of the proband and his parents had no idea that the condition might be transmitted. His daughter recognises that she may some day have to decide whether to take the risk of transmitting this deformity to her children.

**SUMMARY**

1. A family is reported in which tibial aplasia with polydactyly of hands and feet and absent thumbs occurred in a father and daughter.
2. The evidence suggests the existence of an autosomal dominant mutant gene that causes absent thumbs and manual polydactyly, with or without varying degrees of tibial hypoplasia and pedal polydactyly.

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**REFERENCES**


