CONGENITAL DIPLOPODIA WITH HYPOPLASIA OR APPLASIA OF THE TIBIA

A Report of Six Cases

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The syndrome described here is a combination of two congenital anomalies of the same leg, namely partial duplication of the foot or diplopodia and hypoplasia or aplasia of the tibia. The first report dates back to 1966 when Karchinov mentioned four cases and described one of them which is included in this account as Case 1.

Diplopodia as an independent entity is much more rare than diplocheiria or duplication of the hand. Cases have been published by Canale and Rizzo (1962), Guerzoni and Lenzi (1963) and Gandolli and Guerzoni (1964, 1965). Hypoplasia or aplasia of the tibia as an independent condition has long been recognised. Ollerenshaw (1925) stated that Otto made the first report in 1841. The cases reported in the literature now amount to about 120.

This paper reports six patients from Bulgaria, five female and one male, and aged from four months to fifty years. In two cases the right leg was affected, in two the left leg and in two both legs.

The diplopodia is manifested by partial duplication of the foot, the extra part being on the tibial aspect. The term polydactyly is not appropriate because the additional structures comprise not only two or three toes but also well-formed tarsal bones with corresponding metatarsals. The formation of an entire foot is never attained. The normal and the accessory feet together give a monstrous appearance resembling a fan. The two are in varus deviation because of the hypoplastic or aplastic tibia. In three of our six cases the accessory structure consisted of three toes, each with three phalanges and with corresponding metatarsal and tarsal bones, whereas each of the other three patients had two supernumerary toes and related structures. Occasionally the additional tarsal bones articulated independently with the inferomedial border of the hypoplastic tibia.

With regard to the tibia in our six patients, four were hypoplastic and two were totally absent. Normally the proximal tibial epiphysis develops in the last four months of intrauterine life and is visible in radiographs at birth. The centre for the distal epiphysis appears after one to one and a half years. In this syndrome the epiphysial nuclei of the tibia are either completely missing or develop in part after a substantial delay. When growth is complete the tibia presents as a short thick bone, cuboid or trapezoid in shape. In our oldest patient hypertrichosis of the skin was present over the hypoplastic bone. The fibula is not involved, but owing to the shortening of the tibia it projects proximally and distally, with the upper end displaced upwards and outwards in relation to the femur and the lower end displaced downwards and outwards in relation to the talus and calcaneum. In cases of total absence of the tibia, and rather more frequently in hypoplasia, the fibula is bowed under the effect of a fibrous band replacing the absent or underdeveloped tibia. Along with these changes in the tibial skeleton, congenital changes also involve the muscles and tendons of the leg and foot, with replication of some of them.

It is obvious that in this syndrome the tibial component of the lower limb is primarily involved. However, it is worth noting that whereas proximally the disorder leads to hypoplasia or aplasia, distally an additional or accessory structure results. This is a rare combination.
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Genetics—On tracing the genealogy of the six patients of the series, it was found that the syndrome is neither hereditary nor familial. The clarification of the etiopathogenesis is difficult. The hereditary theory, based on the assumption of primary alteration of genes, obviously cannot be accepted. Rather more acceptable is the morphogenic theory, according to which the gene is primarily normal, with faulty development of the tibial embryonal ray occurring within the first four to six months of foetal life. The teratogenous theory may also be considered. The malformation, more particularly the diplopodia, might be the result of excessive development of two or more genes which are divided originally but fuse. It is difficult to determine the validity of either theory.

Differential diagnosis—This should be made from the following congenital anomalies of the musculo-skeletal system: 1) hypoplasia or aplasia of the tibia associated with leg shortening and varus of the foot; diplopodia is absent; 2) ordinary polydactyly or diplopodia of the foot with supernumerary toes or a tendency towards duplication of the foot: aplasia or hypoplasia of the tibia is absent; and 3) the Laurence-Moon-Biedel syndrome characterised by polydactyly of the foot combined with retinitis pigmentosa, obesity, mental retardation and hypogenitalism: neither diplopodia nor hypoplasia of the tibia is observed.

TREATMENT

Treatment is surgical, being directed towards the two basic components of the syndrome. The supernumerary foot is amputated, with improvement of the form and function of the remainder. All accessory elements—tarsal, metatarsal and digital—should be removed. Not
infrequently the amputation is performed soon after birth on the insistence of the parents. The most opportune time, however, is just before the child begins to walk, when treatment of the tibia is commenced.

Reconstruction of the tibia using the available fibular tissue is the difficult part of the overall surgical treatment. To a great extent the outcome depends on the age when treatment is begun and on the degree of hypoplasia.

The various methods of reconstruction used may be summarised as follows: 1) creation of a new joint between the lower end of the femur and the head of the fibula; 2) lengthening of the hypoplastic tibia combined with proximal tibio-fibular synostosis; 3) simple proximal tibio-fibular synostosis; 4) angulation osteotomy of the fibula to permit weight-bearing by the lower end of the femur; 5) reconstruction of a tibial malleolus; 6) distal tibio-fibular synostosis; 7) implantation of the lower end of the fibula into the talus. Finally, amputation of the whole foot may be advisable.

CASE REPORTS

Case 1—This boy aged thirteen months presented with bilateral aplasia of the tibia with diplopodia and severe polysyndactyly of the hands (Figs. 1 and 2). Both legs were short with only the fibulae palpable. The broad inverted feet each had eight toes, all the accessory parts being medial. The radiograph showed bilateral and symmetrical complete absence of the tibia with dislocation of both upper and lower ends of the fibula, three extra toes and corresponding metatarsal and tarsal bones (Fig. 3).

In the first stage of reconstruction the accessory foot was removed (Fig. 4). At the second stage osteotomy of both fibulae was performed at the junction of upper and middle thirds, permitting alignment of each femur with the lower part of the fibula (Fig. 5). Orthotic devices of leather were constructed for the lower limbs, weight being transmitted through the lower ends of the femora. With these aids the child began to walk (Fig. 6).

Case 2—This girl aged eight presented with hypoplasia of the right tibia associated with bilateral diplopodia which had been treated by amputation of the accessory structures at the age of one month. Clinically, the right leg was short and each end of the fibula was prominent beneath the skin (Fig. 7).
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Case 2. Figure 7—Showing hypoplasia of the right tibia. Bilateral diplopodia had been treated by amputation of the supernumerary parts at the age of 1 month. Figure 8—The radiograph shows right-sided tibial hypoplasia with absence of the distal epiphysis and presence of additional tarsal bones in the two feet.

Case 2. Figure 9—A radiograph showing elongation of the tibia and superior tibio-fibular synostosis. Figure 10—To show the lengthened right leg with the upper end of the fibula no longer prominent.
The right foot was moderately inverted. The amputation scars were visible along the medial borders of the feet, with some remaining excess of skin and soft tissue. The radiograph showed hypoplasia of the right tibia with absence of the lower epiphysis, dislocation of each end of the fibula, and remnants of the supernumerary tarsal bones (Fig. 8).

In order to lengthen the tibia a long Z-osteotomy was performed, the fragments were distracted three centimetres and held by two wire loops. The proximal end of the fibula, which had already been dissected free, was implanted in a bed prepared in the upper part of the diaphysis, thus helping to maintain the elongation of the tibia. By five months consolidation had occurred with a useful amount of elongation of the limb and a pleasing cosmetic result (Figs. 9 and 10).

**Case 3**—This girl presented at the age of two years with hypoplasia of the right tibia and diplopodia. Clinically the right leg was 3 centimetres short with the foot inverted and on the medial border an accessory foot with three toes (Fig. 11). The radiograph showed absence of the distal part of the tibia (Fig. 12).

In the first stage of surgical treatment the accessory foot was amputated. In the second stage the head of the fibula was carefully resected, a proximal tibio-fibular synostosis was created, and the fibular head was used for construction of a tibial malleolus (Fig. 13).

**Case 4**—This girl aged eighteen months presented with hypoplasia of the left tibia and diplopodia. Clinically the left leg was 3 centimetres short; the foot was inverted and there were eight toes (Fig. 14). The radiograph showed hypoplasia of the tibia, the upper half of which was absent. The fibula was bowed; the upper end was postero-lateral to the prominent lower end of femur. The accessory foot had three toes and metatarsals and at least one tarsal bone (Fig. 15).

At the first operative stage the accessory foot was amputated. A new joint was then created between the distal end of the femur and the head of the fibula. The displaced fibular head was mobilised through a lateral incision. The tendinous part of the biceps was preserved, together with an extended fascial strip to serve for insertion of the muscle and for the fashioning of an external collateral ligament of the "knee". Through an anteromedial incision the distal end of the femur was next exposed; it

**Case 3**—A radiograph showing the appearances after amputation of the accessory foot, resection of the fibular head, proximal tibio-fibular synostosis, and formation of a tibial malleolus with the head of the fibula.

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Case 4. Figure 14—Showing hypoplasia of the left tibia with diplopodia. Figure 15—The radiograph showing the tibia represented by a small distal part. There are eight metatarsals and toes.

Case 4. Figure 16—A radiograph showing the creation of a new joint between the femur and the head of the fibula. The position of the two bones is maintained with a Kirschner wire. Figure 17—A radiograph to show the appearances after transfer of the lower shaft of the fibula to the residual tibia, with retention of the fibular malleolus.
Case 5. Figure 18—Showing severe hypoplasia of the tibia with diplopodia in a woman aged 50. Figure 19—The radiograph showing the tibia greatly shortened and trapezoid in form, the fibula bowed. The accessory foot comprising two digital rays can be seen articulating independently with the inner side of the deformed tibia.

Case 6. Figure 20—Showing aplasia of the left tibia with diplopodia. Figure 21—The radiograph showing total absence of the tibia, a bowed fibula and a foot with seven metatarsal rays and toes.
was oval in shape and lacked an intercondylar fossa. The patella was dissected out together with its related tendinous tissue, to be used for reconstruction of the extensor apparatus. With great care to protect the epiphysial cartilage, the head of the fibula proper was lowered beneath the distal end of the femur and fixed in situ by a Kirschner wire driven downwards through the lower end of the femur into the head and shaft (Fig. 16). The collateral ligament and the extensor apparatus were then fashioned. The wire was removed eight months later. Three months after its removal the range of movement in the newly formed knee joint amounted to 40 degrees.

At the second stage the lower shaft of the fibula was embedded in the residual part of the tibia. The fibular malleolus was preserved for the formation of an ankle mortise (Fig. 17). Histological investigation of the tibial bone demonstrated osteoporosis and a fibro-fatty medulla; scattered round-cell infiltration was also seen.

Case 5—A woman aged fifty presented with hypoplasia of the right tibia and diplopodia. She had never sought medical treatment. Clinically the leg was grossly short, with the head of the fibula prominent laterally (Fig. 18). The inverted foot had seven toes. There was hypertrichosis along the anterior aspect of the deformed leg. When walking her weight was borne on the lateral surface of the shortened leg and outer border of the foot. The radiograph showed a short trapezoid tibia and the fibula bowed outwards (Fig. 19). The elements of an accessory foot were present, with two toes and metatarsals and with tarsal bones, the latter articulating independently with the deformed tibia in a medial notch (Fig. 19). Because reconstructive measures were quite impractical, the foot was amputated and a below-knee type of prosthesis fitted.

Case 6—This girl presented at the age of one and a half months with an absent left tibia and diplopodia. Clinically there was gross shortening of the leg. The foot had seven toes and was so adducted in relation to the fibula that the medial border was touching the inner side of the thigh (Fig. 20). The radiograph showed absence of the tibia, a bowed fibula and a foot with seven metatarsals and toes (Fig. 21).

As a first stage the accessory foot was amputated. Reconstruction of the leg will be undertaken at the age of one year.

SUMMARY

1. The syndrome here described in six Bulgarian subjects is characterised by partial duplication of the foot or diplopodia, combined with either hyperplasia or aplasia of the tibia. The accessory elements are located along the medial border of the "normal" foot and consist of two or three toes with related metatarsal and tarsal bones. The fibula is not directly involved.

2. No evidence of familial inheritance was found in these cases.

3. Various reconstructive measures and the place of amputation of the whole foot are discussed in the course of the six case reports.

REFERENCES


Otto (1841): Quoted by Ollerenshaw (1925).