CONGENITAL ABSENCE OF TIBIA.

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This study was undertaken as an enquiry into the aetiology of congenital defects of bone, and into the indications for treatment in the particular case of partial or complete absence of the tibia. Although with regard to the former neither this nor any other of the recent investigations has succeeded in arriving at the whole truth, we hope that the ground has been cleared, several unsatisfactory hypotheses disproved, and the way opened up for the discovery of the ultimate cause in the course of our study. In addition, some light has been thrown on problems of embryology, comparative anatomy, and muscle physiology.

The material for this investigation is derived from a number of cases studied clinically and by radiographic examination, a series of dissections of specimens obtained from these cases, and an examination of the other specimens (except one recently described by Ollerenshaw(23)) in the Royal College of Surgeons Museum. A survey of all the available literature has also been made.

I.—DEFINITION AND GENERAL DESCRIPTION.

Congenital absence of the tibia is defined as "the absence at birth of the tibia in whole or in part, the lower limb being nevertheless present with its different segments well developed in their essential parts." This definition (modified from that of Launois and Küss(18)) excludes cases where the absence of the tibia is merely a part of a deformity such as absence of the...
entire lower limb, or of deficiency of the distal segments of the limb, or of "congenital amputation" through the leg or thigh.

The deformity is one of the rarest of the congenital malformations, and certainly the most uncommon of those of the lower limb.

Hoffa's statistics of 9,877 cases of congenital deformity of all sorts give 2,425 of the lower limbs, without mention of absence or defect of the tibia. The number of published cases now amounts to 102. Of these the first case described is stated by several writers to be that of Otto in 1841. Launois and Kies(18) in 1901, in a valuable paper, recorded 41 cases they had collected; and Nuzi(22) in 1920, including those in his series, brought the number up to 81. The three cases of Clutton(7) (1896) and one each of Openshaw(24) and Muirhead Little(19) (1913) were, however, omitted from this list, as were also those of Boetticher (1904), Munro (1904), Spaunau (1908), Davidson (1918), Tissier, and Broca (1914), since recorded by Bertaux(4) (1920) in publishing his own two cases. Further cases have recently been published by Rocher(26) (1920), Vallois and Roume(27) (1922) and Ollerenshaw(23) (three in 1925). The remaining three, those of Cautley(6) (1915), Elmslie(10) (1920) and Laming Evans's(11) second case (1923), are included in our own series, as, although they have been already reported clinically, they are now described anatomically for the first time. Finally to this list of 102 we add three more cases:—those of Thomas Bryant (1888), Laming Evans's first case (1915), and Fairbank (1924), which have not yet been published, bringing the total number up to 105.

Congenital absence of the tibia may be total or partial, unilateral or bilateral. Statistics given by different writers vary, and are not very reliable because many of the cases have been very imperfectly described; and, moreover, most of the descriptions have not been aided or confirmed by radiographic and anatomical examination. We agree with the conclusions of Bertaux(4), namely, that total absence is more frequent than partial, unilateral absence occurs more often than bilateral, and of the unilateral cases the defect is of the right limb more frequently than of the left. In none of these points, however, is the difference great. A noteworthy feature of the partial cases is that the defect is almost always of the distal end of the bone. Of the bilateral cases, there may be total absence on both sides, or total on one side and partial on the other.

With regard to sex there is a slight preponderance of males over females. In our own series there are three males, two females, and one with no record of the sex. Bertaux states that the affection is twice as common in males as in females; but our reading of the literature does not bear this out, and is in favour of the view above expressed.

The family history is usually negative for congenital abnormalities and other diseases. There are, however, a considerable number of instances of the occurrence of congenital defect of the tibia, or of other abnormalities, in near or remote relatives. In this connection Ollerenshaw's(23) recently published record of absence of the tibia in identical twin girls, the eleventh and twelfth children of a family otherwise normal, is interesting.

The nature of the birth is not stated in most cases. A few writers have noted deficiency of the liquor amnii or that labour was difficult. Others, however, have stated that birth was normal and pregnancy uneventful. Maternal injuries and impressions, and illness during pregnancy, have been noted in only a few instances.

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Associated abnormalities are present very frequently. Launois and Kiss (18) noted them in 24 out of their 41 collected cases. In five of the six cases in our series they were present. These abnormalities are not only confined to the limb where the tibia is absent, but may affect the upper limb, trunk or head. They are most common in the affected lower limb, occurring, for example, as congenital diallocation of the hip, ectro-, poly-, or syn-dactyly, or in the abnormalities of the bones and muscles of the thigh, leg, and foot, given in the anatomical description below. In the upper limb phocomelia, absence and partial defects of the forearm (of the radius most commonly), and of hand bones and fingers may occur; and with regard to the head and trunk, harelip and cleft palate, pseudo-hermaphroditism, cryptorchidism and hypospadias have all been observed in various cases.

It is rare to find any stigmata of fetal disease in cases of congenital absence of the tibia. This statement is supported by the records of almost all the cases yet published. It is a striking fact, common perhaps to most congenital deformities, that, while possessing a gross defect such as that of absence of the tibia, the individual is usually otherwise perfectly healthy. These facts seem to throw considerable doubt on fetal disease as a possible cause of the defect.

The subjects of this deformity are thus to be met with at any age, and it is worth while to observe their natural method of progression if untreated; this will be referred to again below.

II.—CLINICAL AND RADIOGRAPHIC FEATURES.

The characteristic signs of congenital absence of the tibia are:

1. Shortening of the leg. This is invariably present to a greater or less extent, the degree depending largely upon the age of the patient when seen, since post-natal growth as well as pre-natal development is defective, and the affected leg becomes progressively shorter as compared with the normal and relatively to the other segments of the limb, as age advances. There is frequently also shortening of the thigh to a less extent. The shortening of the leg may be so marked that the leg appears as a mere appendage of the thigh. There is not necessarily wasting of the leg or thigh; they are in fact frequently well formed and plump.

2. Flexion and adduction of the leg on the thigh. These take place at the knee joint and their degree depends on whether the tibial defect is total or partial. In cases of total absence the flexion and adduction are well marked, constituting a gross deformity in which the inner borders of the leg and thigh may even be in apposition. Accompanying this are limitation of extension at the knee, and, usually, luxation of the head of the fibula upwards and backwards, so that it comes into contact with the postero-lateral aspect of the lateral femoral condyle, to which it is articulated by a well formed diarthros's, or, less commonly, by a syndesmosis. When the tibia is only partially defective, and particularly when a considerable amount of
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3. Inversion of the foot on the leg. This also is a constant feature. It occurs mainly at the ankle joint and is apt to be very marked, and is usually irreducible.

4. Palpation. This reveals that there is but one bone in the leg (in the case of total absence of the tibia) and that having the form and position of the fibula; where the absence is partial, the defect may be recognisable. The absence of the normal bony points, e.g., the medial malleolus, is also determinable.

5. Appearances on Radiographic Examination. It is advisable that X-ray examination should be done in all cases, since it forms the most certain method of diagnosis, and the only sure way of determining whether or not the tibial defect is total or partial. It also gives valuable information of the state of osseous development of the other segments of the limb and of the pelvis.

6. Minor features. Certain other features, less constant and important, may be present. The fibula is often found to be not only shorter than normal, but also stouter and curved with convexity outwards, imparting a similar longitudinal curve to the leg. The prominence of the lateral malleolus is frequently a noticeable feature. The femoral condyles may be ill-formed or absent, as may be also the patella. Cutaneous dimples, grooves, constrictions and "scars" are often present; and abnormalities of the foot such as syndactyly, polydactyly, and ectrodactyly, or even more gross deformities are frequent occurrences.

When the tibial defect is partial, it is almost invariably the distal end which is absent. In one recorded case only (Parona\(^{(29)}\)), quoted by Bertaux\(^{(4)}\), has the proximal end been the absent portion. The proximal end may be a mere nodule, when it may not be palpable; it is, however, often larger, and is then apt to be conical in form with its distal end sharp-pointed and projecting forward beneath the skin of the leg. A puckering or mamillary elevation of the skin over this projection is not an uncommon feature.

Except in the uncommon cases of slight partial absence, the deformity resulting from the defect is so great as to make the natural method of progression impossible. The victims of this deformity are only able to move themselves either by jerking themselves forward on their buttocks with the aid of their hands, or by walking on their hands and knees. Some patients with unilateral defect learn to stand and walk upright with the aid of a crutch. One of the patients included in our series was able to walk well on her knees. The majority, however, are unable to move except in one of the ways described. In all cases of total defect, the leg is useless.
III.—Cases.

The series of cases here given includes three hitherto unpublished (Laming Evans's first case, Fairbank's and Bryant's cases); and five which have been already recorded elsewhere. The latter are included mainly for the sake of completeness, because in the anatomical section of the paper, descriptions are given for the first time of the specimens derived from them.

Case I. Laming Evans's first case (1915)—bilateral total absence of the tibia.

G. T., male aged 3 weeks (Fig. 1). Royal National Orthopaedic Hospital. Family history: Parents were healthy with no deformities in themselves or among their near relatives. There were ten previous children all without deformity, of whom eight are alive and well. Mother was aged 37 years. Pregnancy and birth were normal.

Both of the child's legs were flexed on the thighs at an angle of 80°. Scars were present over the prominence of the femoral condyles. The upper ends of the fibulae were displaced backwards relatively to the femora; while the distal ends formed sharp-pointed prominences at the lowest points of the legs. Both tibiae were completely absent. Each foot was displaced markedly inwards and upwards on the leg so as to lie along the medial side of the leg, which it overlapped at one point. The hallux was absent from both feet; and there were two toes only on the right foot. The right testicle was incompletely descended; the left hand had only three digits, the right hand two only with a deep cleft between them; and the right radius and ulna were abnormally short.
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X-ray examination: In both legs one bone only is shown. This bone is well ossified and straight and almost certainly from its position relative to the foot, the fibula. There may be a slight doubt as to its identity because the proximal end lies not, as usual, to the lateral or posterolateral aspect of, but in the middle line immediately below, the distal end of the femur.

No treatment was adopted at the time, and the child was not brought to hospital again.

Case II. Laming Evans's second case (1923)—left unilateral total absence of the tibia. B. B., male, aged one year and five months (Fig. 2). Royal National Orthopedic Hospital. Patient was the first child of young parents both without deformity and with no deformities in either of their families. Pregnancy and birth were normal. A second child had been born since without deformity.

The child was well developed and healthy. The left thigh was \( \frac{1}{2} \)" shorter and smaller in circumference than the right. The right patella was well developed; the left impalpable. The left leg was shorter than the right and was flexed upon the thigh. Flexion was complete, but extension was short by 45°. The upper end of the fibula was articulated with the upper part of the lateral femoral condyle. The fibula was palpable through its whole extent, was curved with convexity outwards, was thicker and one inch shorter than the right fibula. The lateral malleolus was unduly prominent. A deeply recessed dimple was present over the head of the fibula and was continued downwards over the outer aspect of the fibula as a linear scar-like depression.
The foot lay in complete supination along the inner border of the leg. The general conformation of the foot was normal, but the great toe was bifid in its ungual segment. The circulation was good and the limb not wasted. As far as one could tell sensation was normal. The left testicle lay in the high scrotal position; but there were no other abnormalities.

X-ray examination (Figs. 3 & 4). (i) The pubic segment of the os innominatum is still without sign of ossification (normally this centre appears in the fourth month of intra-uterine life); (ii) the ossific centre for the head of the femur is absent on the left side; (iii) the left tibia is completely absent; (iv) ossification of the tarsus is normal; (v) the accessory toe lies on the medial side of the ungual phalanx of the hallux, and has two centres of ossification, thus resembling the four outer toes.

The treatment adopted was amputation at the knee joint. The specimen was dissected (see below).

Case III. R. C. Elmslie's case (1920)—bilateral total absence of the tibia. M. M., female, aged 13 years (Figs. 5 & 6). St. Bartholomew's Hospital. The tibia was totally absent in both lower limbs producing gross deformity, the legs being flexed to a right angle and displaced outwards and backwards on the thighs, and externally rotated. The patellae were present and the femoral condyles well developed. The heads of the fibulae were large, and articulated with the outer side of the lateral femoral condyles. Severe talipes equino-varus was present on both sides. The second toe was absent from the right foot, a deep cleft existing in its place. The hallux was missing from the left foot. The child walked well on her knees.

X-ray examination (Figs. 7 & 8). (i) Total absence of the tibiae; (ii) both patellae present and well formed; (iii) luxation of the heads of the fibulae to the postero-lateral aspect of the distal ends of the femora; (iv) absence of the left hallux and all but the base of the first left metatarsal, and in the right foot absence of the second toe and fusion of the first and second phalanges of the third and fourth toes.
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Fig. 5.

Fig. 6.

Fig. 7.

Fig. 8.
Amputation at the knee joint on both sides was performed and peg-legs subsequently fitted. A note eighteen months later stated that the child was walking well. (For description of the specimens see below.)

Case IV. H. A. T. Fairbank's case (1924)—right unilateral absence of the tibia. J. R., male, aged two years (Fig. 9). Hospital for Sick Children. The right tibia was totally absent. The patella was palpable and the femoral condyles well developed. The head of the fibula articulated with the lateral condyle. The foot was in extreme varus; only one toe and two metatarsals were present, and, of the tarsus, the calcaneus was alone determinable. Left talipes varus was also present, with fusion of the second and third toes, and absence of the first, fourth and fifth metatarsals, and absence of the navicular. The right hand showed the "lobster claw" deformity with associated deficiency of the carpus. The left hand bore only one digit (a thumb) and the left carpus was also deficient.

Fig. 9.

X-ray examination (Fig. 10). (i) In the right lower limb, the hip joint is normal, the tibia totally absent, the head of the fibula luxated to the postero-lateral aspect of the distal end of the femur, the calcaneus and cuboid present; (ii) in the left foot, the calcaneus, cuboid, talus, two metatarsals, and two digits are shown, and the lower epiphysis of the tibia appears to be in two portions; (iii) in the right hand, the outer digits consist of two phalanges and one metacarpal, the carpal region casts no shadow, and the lower end of the radius and ulna are normal; (iv) in the left hand the digit consists of three bones, two phalanges and one metacarpal, and there is no shadow in the carpal region; (v) the pelvis is normal.
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Amputation of the right leg through the knee joint was carried out, the patella being left in the flap. (For dissection of specimen see below.)

Case V. E. Cautley's case (1915)—right unilateral partial absence of the tibia. Female, died at six months, from "zymotic enteritis," weighing 6½ lbs. at death. The child was the fifth of a family in which there had been no member with deformity. The right lower limb was shorter than the left, and was inwardly curved. The foot had seven toes, the accessory digits lying to the inner side of the hallux and being identical with the normal second and third toes. The hallux was short, and the second and third toes were closely united almost to the proximal end of the terminal phalanges. The fibula was thought at the time to be absent and the tibia to be very much curved; but dissection (see below) has shown the fibula to be present and the tibia defective. The left foot closely resembled the right in appearance, presenting similar extra toes. There was also webbing of the extra toes and the normal second and third toes.

Case VI. Thos. Bryant's case (1888)—total absence of the right tibia. No clinical record is available of this case which is reported for the first time here. A description of the dissected specimen is given below.

Case VII. Clutton's second case (1896)—bilateral total absence of the tibia. Female, aged eight years, in whose family there were no deformities. (Fig. 11). The tibia was totally absent in both limbs. The femoral condyles were under-developed, the femoral shafts ending in rounded knobs. The proximal end of each fibula was attached to the outer side of the distal end of the femur. The left great toe was very small, with associated deficiency of the tarsus and metatarsus on the inner side. Four right toes only were present; and there was also a deficiency of the inner part of the tarsus and metatarsus.

(Note.—These were clinical observations only, not confirmed by X-ray examination.)

Amputation at the knee was performed in both limbs. At operation it was noted that the patella and the ligamenta patellæ were absent, and that there was no joint
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cavity on either side, the ends of the femora and fibulae being cartilaginous and bound together by fibrous tissue. The specimen has been dissected and the description is given below.

Case VIII. Bland-Sutton's case (1892)—left unilateral total absence of the tibia.
Female, aged 18 years, healthy. The left leg was relatively short. The tibia was totally absent, and the fibula short, but otherwise well developed. The distal end of the fibula articulated with the talus; and the foot was in an extreme equino-varus position.

Amputation through the knee was adopted. A description of the macerated specimen is given below.

IV.—Diagnosis.

The diagnosis is rarely difficult. The deformity, with its three features of shortening of the leg, flexion and adduction at the knee, and marked varus of the foot, is, except in cases of slight partial defect, a gross one and pathognomonic in itself. Palpation reveals that one bone alone is in the leg, or that the tibia is partially defective; and X-ray examination makes the diagnosis certain. Difficulty has been reported in deciding whether the bone felt in the leg is the fibula or the tibia. This difficulty is strongly insisted on by some authors, who point out that it is increased by the tendency for modifications to occur in the size and shape of the fibula when the tibia is absent, and vice versa. One of the cases in our series was first shown at a clinical meeting as a case of absence of the fibula, and on subsequent dissection has been proved to be one of partial defect of the tibia. Ordinarily, however, there should be no difficulty; when the fibula is absent the foot is everted and the knee joint is well formed, though there may be flexion at the knee. Shortening of the leg is common to both affections.

Where the distal third only of the tibia is deficient the resulting deformity might be mistaken on a cursory examination for the ordinary congenital talipes equino-varus of a severe kind, but the absence on palpation of the medial malleolus and the radiographic examination easily determine the diagnosis.

V.—Anatomical Description.

1. Laming Evans's case (1924)—R.C.S. Museum, Teratological Series No. 439.52.

The skin is normal and bears no scars. Over the outer aspect of the head of the fibula is a dimple, from which a groove runs downwards on the outer side of the leg in the line of the fibula to the lateral malleolus. (Microscopic examination of the skin of the groove shows no scar, but diminution in the thickness of the basal layers of the epidermis.) The nails are fully grown and the skin is hairless. The superficial fascia is abundant. No bursae are present. The deep fascia is very thin and the special bands poorly developed: the plantar aponeurosis is ill defined and the ligamenta transversum cruris and laciniatum indistinguishable; but the ligamentum cruciatum cruris and the peroneal retinacula are clearly marked.

Muscles.—(Figs. 12 & 13). The quadriceps femoris tendon, thin and expanded, blends with the front of the capsule of the knee joint. The tendons of no other thigh muscles are present, except one on the medial side, which, from its insertion into the fibro-cartilaginous mass at the proximal end of the representative of the tibia, is presumably that of semimembranosus. All the muscles of the leg and foot are present;
but their arrangement is abnormal, and there are also present several muscles which have no place in the normal limb. Arising by a continuous fleshy origin from the upper four-fifths of the anterior border of the fibula is a muscle which divides in front of the ankle into three tendons running to the lateral three toes, a tendon to the base of the fifth metatarsal, and a fleshy remainder which is inserted into the dorsal surface of the calcaneus. This muscle therefore represents a part of extensor digitorum longus, peroneus tertius, and a peroneo-tarsus. The tendon of extensor hallucis longus is joined on the lateral side by the fibres of extensor digitorum brevis and is inserted by two slips into the two distal phalanges of the great toe. Extensor digitorum brevis is very large. The medial fibres join the tendon of extensor hallucis longus; in addition there are four tendons, two to the second toe and one each to the third and fourth toes. Abductor hallucis is small and has an abnormal origin from the side of a slender tendon which springs from the medial side of the tendon of extensor hallucis longus on the dorsum of the foot. This tendon, traced round the medial border of the foot, is found to proceed from a fleshy belly lying in the sole deep to the extensor tendons, superficial to the oblique head, and arising from the fascia covering the transverse head of adductor hallucis and the interossei. Flexor hallucis brevis also partly arises from the tendon; but the rest of its origin is normal. Peroneus longus and brevis are large relatively to the other muscles of the limb. Their attachments are normal.

On the posterior aspect of the limb the arrangement of the muscles is more abnormal and the value of each more difficult to determine. The tendons of three muscles unite, as in the specimen described by Symington and Craig in the sole to form a common tendon from which arise five tendons, one to each of the toes. Quadratus plantae joins the deep surface of the tendon and the four lumbricals arise between all five tendons. Of the three muscles entering into this arrangement one arises from the fibro-cartilaginous upper end of the tibial representative, from the anterior aspect of the interosseus membrane, and from the upper fourth of the anterior part of the medial surface of the fibula, and therefore appears to be tibialis anterior; the second muscle also derives
origin from the cartilaginous mass, and since the fibres it contributes to the common tendon go mainly to the tendons of the second, third and fourth toes it is presumably flexor digitorum longus; the third muscle is flexor hallucis longus, since it springs from the posterior surface of the fibula and its fibres are traceable into the slip to the great toe. The tibialis anterior element of the common tendon is given mainly to the tendon to the fifth toe. Tibialis posterior is recognisable in a muscle arising from the fibula posterior to the attachment of the interosseus membrane, and from the membrane itself, and inserted into the medial aspect of the fused talo-calcaneus. A peroneo-tarsus posterior is present, arising from the posterior surface of the fibula and inserted by tendon into the plantar surface of calcaneus deep to quadratus plantae. Triceps surae is present with normal attachments. Plantaris and popliteus are absent. The muscles of the foot not hitherto mentioned are present and normal.

**Blood Vessels and Nerves.**—These are healthy and have practically a normal arrangement.

**Bones.**—(Fig. 14). The patella is represented by a small flattened nodule of cartilage (confirmed by microscopical examination) in the anterior part of the capsule of the knee joint. (The distal end of the femur, seen at the operation of removal of the limb, had no condyles and no intercondylar notch, but was a rounded knob covered with articular cartilage on all aspects except the medial.) The fibula is short and stout, slightly curved longitudinally, with the convexity laterally. The shape, surfaces, and borders differ little from the normal. The proximal articular surface is on the medial aspect of the head and upper end of the shaft, thus extending across the line of the proximal epiphysis. (As seen at operation this surface lay in contact with the articular surface of the distal end of the femur.) The distal articular surface on the medial aspect of the distal end enters into the formation of the ankle joint, and is apposed to the articular surface on the lateral aspect of the talus.
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The tibia is represented by a strong cord-like tendinous structure attached to the fibula by a well-formed interosseous membrane. It contains no bony element; but at the proximal end it expands into an amorphous mass of dense fibrous tissue and cartilage, the proximal surface of which enters into the formation of the knee joint, while on the distal, medial and lateral aspects muscles are attached as described above. The deep aspect of the mass is attached to the medial surface of the fibula immediately distal to the proximal articular surface. The distal end of the tendinous structure is divided into two strong bands attached to the anterior and posterior borders of the distal end of the fibula, and entering into the formation of the ankle joint. The interosseous membrane extends proximally as far as the distal part of the fibro-cartilaginous mass, and ends in a free upper margin over which passes the anterior tibial artery. The talus and calcaneus are fused; but each is well formed and the sinus tarsi is present. The other bones of the tarsus and the metatarsus are present and normal. The proximal phalae of the hallux articulate distally with two nail-bearing elements: the lateral consists of a single phalanx, the medial of two phalanges.

Joints.—The joint representing the knee is formed by the distal end of the femur, the proximal end of the fibula, and the proximal end of the tibial representative. The articular surfaces and the relative positions of the parts entering into the joint have been described above. The capsule is well formed, attached below to the border of the amorphous mass described, and, elsewhere, close to the margins of the articular surfaces. The thin expansion of the quadriceps tendon forms the anterior part of the capsule. The menisci and ligamenta cruciata are absent. The ankle joint is formed between the upper surface of the talus, the distal end of the fibula, and the bifurcated distal end of the representative of the tibia. The capsule is thin and shows no specialised bands. The other joints of the foot are normal.

Summary.

1. The dimple in the skin over the head of the fibula with the continuing groove over the whole length of the outer border of the leg, the depressions not being due to scarring.
2. Absence of certain muscles, both of the thigh and leg.
3. Extensor digitorum longus which is partly a long extensor of the toes, but contains also peroneus tertius and peroneo-tarsus elements.
4. Origin of abductor hallucis indirectly from extensor hallucis longus.
5. Union of tendons of tibialis anterior, flexor digitorum longus and flexor hallucis longus to form a common tendon from which arise the flexor tendons for the five toes.
6. The presence of a peroneo-tarsus posterior.
7. The presence of a small patella.
8. The absence of the femoral condyles and intercondylar fossa.
9. The short hypertrophied fibula slightly outwardly curved, articulating with the distal end of the femur by an articular surface extending across the proximal epiphyseal line.
10. The representation of the tibia by a tendinous cord, ending proximally in an amorphous fibro-cartilaginous mass, and distally bifurcating to form the upper wall of the ankle joint.
11. The fusion of the talus and calcaneus.
12. The accessory hallux.
13. The absence of the menisci and cruciate ligaments from the knee joint, which is formed between the distal end of the femur, proximal end of the fibula, and the fibro-cartilaginous representative of the proximal end of the tibia.

2. Elmslie's case (1923)—R.C.S. Museum Teratological Series No. 439.5.

The specimen has been fully dissected to show the arrangement of the muscles and articulations.

Muscles.—No tendons of muscles of the thigh normally inserted around the knee are present in the specimen.
On the posterior aspect of the leg, the popliteus is absent. Plantaris, lateral head of gastrocnemius, and soleus arise from the head of the fibula. Plantaris tendon joins tendo calcaneus, which has a normal insertion. Tibialis posterior arises from the head of the fibula immediately below and in front of the upper articular surface, and is inserted into the upper surface of the fused talus and calcaneus. Flexor digitorum longus is double. One muscle arises in common with tibialis posterior; its tendon breaks into three slips—the largest divides into tendons for the fourth and fifth toes, one is inserted into the talo-calcaneus in front of tibialis posterior, and one joins that part of the tendon of flexor digitorum brevis which unites with the second flexor digitorum longus. The latter arises partly in common with extensor digitorum longus from the head of the fibula and partly with extensor hallucis longus from a shaft of the fibula; the tendon, after giving off a slip attached to the navicular, is joined by the medial tendon of flexor digitorum brevis, and then breaks into two tendons, one of which goes to the third toe, and the other is inserted into the head of the second metatarsal. (Note.—The second toe is absent.)

The flexor and extensor hallucis longus arise together from the shaft of the fibula. From a single fleshy belly arise in the foot two tendons, one of which joins flexor hallucis brevis, while the other, after receiving laterally a tendon from extensor digitorum brevis, is inserted into the dorsal surface of the great toe.

Anterior Muscles.—Of the other muscles of this group extensor digitorum longus, with origin as described above, gives off a tendon to each of the lateral three toes and to the head of the first metatarsal; the peroneus tertius and tibialis anterior are absent. An abnormal muscle similar in position and attachments to that found in Fairbank's specimen, and so to be designated a peroneus, is present. It is a large musculo-tendinous mass attached to the front of the upper two-thirds of the shaft of the fibula and with no other attachment beyond the fibula. (Microscopic examination shows that the muscle fibres are abnormally large and not degenerated in any way.)

Peronei longus and brevis are normal. In the foot abductor hallucis is absent, and the tendon of flexor hallucis longus joins the short flexor as described above. Flexor digitorum brevis has two tendons: the lateral is joined by the very small quadratus plantae, and itself joins the slip of flexor digitotum longus to the fourth toe; the medial, re-inforced by a slip from the lateral flexor digitotum longus, joins the tendon of the medial flexor of the toes.

Bones.—The tibia is completely absent and has no representative. The fibula is slightly hypertrophied, but is not bowed. On the postero-lateral aspect of the head is an articular surface for the femur. The distal end articulates with the talus. The talus and calcaneus are both malformed and are fused. The other bones of the tarsus and all the metatarsals are present; but the second toe is absent, and there is fusion of the proximal and middle phalanges of the third and fourth toes.

Articulations.—The knee joint is well-formed between the distal end of the femur and the head of the fibula. The articulations between the distal end of the fibula and the talus and those of the foot are normal.

Summary.

1. The absence of the tendons of the thigh muscles and of certain muscles, particularly peroneus tertius, tibialis anterior and abductor hallucis, of the leg and foot.
2. The duplication and partly abnormal insertion of flexor digitorum longus.
3. The origin of flexor and extensor hallucis longus tendons from a common muscle belly.
4. The presence of a peroneus identical in position and attachments and microscopic appearances to that of Fairbank's specimen.
5. The absence of any representative of the tibia.
6. The practically normal fibula.
7. The knee joints formed between the proximal end of the fibula and the distal end of the femur with absence of menisci and cruciate ligaments.
8. The malformation and fusion of talus and calcaneus.
9. The absence of the second toe and synostosis of the phalanges of the third and fourth toes.
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The skin, subcutaneous tissue and deep fascia had been removed before this examination was made.

Muscles.—(Fig. 15). The leg was amputated through the knee joint. The tendons of none of the thigh muscles, with the exception of the quadriceps extensor, are present.

Of the posterior muscles of the leg, popliteus and flexor hallucis longus are absent. The medial head of the gastrocnemius arises partly from the femur, partly from the medial side of the head of the fibula. The lateral head arises from the lateral side, and soleus from the posterior aspect of the head of the fibula. The tendon calcaneus is inserted as normally into the calcaneus. Plantaris arises, in conjunction with the medial head of gastrocnemius and tibialis anterior, from the medial side of the head of the fibula, and is inserted into the calcaneus alongside tendon calcaneus. Tibialis posterior, arising from the upper half of the posterior surface of the fibula, divides into a tendinous and a fleshy slip, which are inserted, the latter behind the former, into the upper surface of the calcaneus. Flexor digitorum longus, arising from the middle of the shaft of the fibula, has only one tendon, which is joined on the dorsum of the foot by extensor digitorum brevis, and is ultimately inserted into the terminal phalanx of the toe.

In the sole of the foot abductor and adductor hallucis, flexor hallucis brevis and quadratus plantae are absent. One lumbrical only is present, arising from the inner side of the tendon of flexor digitorum longus. Flexor digitorum brevis and abductor digiti quinti have a normal origin; but the former is inserted partly by a tendon into the toe and partly by a separate musculo-tendinous slip into the lateral side of extensor digitorum brevis, superficial to the attachments of flexor digitorum longus. Two plantar and one dorsal interosseal are present.

Peroneus longus is inserted into the plantar aspect of the bases of both the metatarsals present in the foot. Peroneus brevis gives rise to two tendons: the posterior is inserted into the base of the lateral metatarsal, and, by a continuing slip, into the proximal phalanx of the toe; the anterior is continuous with the tendon of a muscle (presumably peroneus tertius), arising from the lateral aspect of the fibula, immediately in front of the peronei longus and brevis. Thus is formed between two muscle bellies a strong tendinous loop, which lies in a groove on the plantar surface of the calcaneus beneath the articulation of that bone with the fibula.
On the anterior aspect of the limb and dorsum of the foot are recognisable: (i) A muscle arising from the fibula, in front of the muscles just described, which is probably an extensor digitorum longus. It is inserted by a tendinous expansion into the calcaneus; but definite strands can be traced forward through the fascia covering extensor digitorum brevis to join the tendons of that muscle which go to the toe and to the base of the medial of the two metatarsals. (ii) A double tibialis anterior. One arises from the whole length of the fibula in front of extensor digitorum longus and is inserted into the medial border of the calcaneus. The other takes origin by two bellies from the proximal end and upper part of the shaft of the fibula, and is inserted into the calcaneus immediately in front of the former muscle. (iii) A muscle which arises from the upper end of the fibula, has a very short belly and a relatively very long tendon, which passes distally in close contact with the bone to be inserted into the distal end of the fibula. This muscle probably corresponds to the peroneus described in other specimens. (A microscopic examination of the muscle fibres shows them to be larger than normal and without signs of degeneration.) (iv) An extensor digitorum brevis arising by two bellies from the calcaneus and inserted into (a) the side of flexor digitorum longus tendon, (b) the side of the tendon of flexor digitorum brevis to the head of the medial metatarsal, (c) the distal phalanx, (d) the proximal phalanx of the toe. The last is joined by a tendinous slip from extensor digitorum longus. Extensor hallucis longus is absent.

Arteries and Nerves.—Both are large and the general arrangement of each is normal. The anterior tibial artery ends in front of the distal end of the fibula. The posterior tibial artery runs in the foot in the situation of the lateral plantar artery. There is no medial plantar artery. The tibial nerve divides into medial and lateral plantar nerves; but the medial gives no muscular branches.

Bones.—The tibia is entirely absent and there are neither cartilaginous nodules nor a fibrous strand to denote its position. The fibula is slightly thicker than normally. It is straight. The upper extremity bears on the postero-superior aspect an articular facet for the distal end of the femur. The lower end forms the lowest part of the limb and has an articular surface on the postero-lateral aspect for the calcaneus. There is no sign of interosseus membrane.

In the tarsus, the talus, navicular and cuneiforms are absent. The calcaneus is malformed and almost inverted, so that the tuberosity is uppermost and lateral to the fibular articular surface, which is on the proximal part of the upper surface. The cuboid articulates with the calcaneus and supports the only two metatarsals present, which are presumably the fourth and fifth. In support of this are the characteristic shape and the muscular attachments of the lateral of the two bones. One toe, comprised of three phalanges, alone is present. It articulates with the lateral metatarsal and is therefore the fifth toe.

Articulations.—The knee joint is formed between the distal end of the femur and the proximal end of the fibula. The articular surfaces are covered with articular cartilage and the joint possesses a well-formed capsule. The ankle joint is well formed between the distal end of the fibula and the calcaneus; slight antero-posterior gliding movements only are allowed. Well-formed joints exist between the other bones of the foot.

(Operation note by Mr. H. A. T. Fairbank: There was a definite large joint cavity in the knee with the fibula articulating over a considerable area of the outer condyle. The patella was present. The femoral condyles were well formed.)

Summary.

1. The total absence of the tibia and of all the structures of the inner segment of the foot, viz., the talus, navicular, cuneiforms, medial three metatarsals and toes (and of the fourth toe), with the intrinsic muscles and the tendons of the leg muscles normally ending therein. In connection with this it is noteworthy that extensor and flexor hallucis longus are entirely absent, flexor digitorum longus has only one tendon and that is joined by extensor digitorum brevis and ultimately inserted into the only toe (the fifth) present, the medial plantar artery is absent and the medial plantar
nerve gives no muscular branches; while the bones, muscles, nerves, and vessels of the outer segment of the foot are practically normal.

2. The large size of peroneus tertius and the abnormal attachment of its tendon to form with that of a separate part of peroneus brevis a tendinous loop under the fibulo-calcaneal articulation.

3. The duplication of certain muscles, e.g., tibialis anterior.

4. The muscle called a peroneus which both arises from and is inserted into the shaft of the fibula and thus could neither have shortened nor subserved any function, yet shows, on microscopic examination, no degeneration, but actual hypertrophy of its fibres.

5. The absence of any representative of the tibia.

6. The absence of abnormal curvature or marked hypertrophy of the fibula.

7. The articulation of the proximal end of the fibula with the lateral condyle of the femur, and of the distal end with the calcaneus.

8. The absence of menisci and cruciate ligaments.

9. The patella is present and the femoral condyles well formed.

10. The malformation and inversion of the calcaneus.

4. Edmund Cautley's case.—R.C.S. Museum Teratological Series No. 410.2.

The specimen was removed from the body after death by amputation through the distal end of the femur.

External features.—The leg is greatly shortened both actually and relatively to the foot, which is slightly longer than the leg. The leg is turned inwards (adducted) upon the thigh so that its long axis lies at right angles to that of the thigh. The foot is completely inverted so that its transverse axis is parallel to the long axis of the leg, while its long axis is at right angles to that of the leg. There are seven toes, of which the two most medial have the characters of a third and second in that order from the medial border. Of the lateral five the most medial is like a great toe in form, though relatively short in comparison with the other toes; and there is syndactyly of the true second and third toes.

The skin and subcutaneous tissues had been removed before the present dissection was undertaken. No account of them therefore can be given.

Muscles.—(Fig. 16). The tendons of three only of the muscles of the thigh are present, namely, that of adductor magnus (very small) attached at the normal site on the femur, and those, presumably, of semimembranosus and semitendinosus, blending with the postero-medial part of the capsule (here fibro-cartilaginous) of the knee joint, and through it reaching insertions on the upper end of the tibia. Biceps femoris, gracilis, and sartorius are absent. The structures in front of the lower end of the femur have been removed in amputating the limb, so that the presence or absence of quadriceps femoris and of the patella cannot be stated; but it is certain that there is no ligamentum patellae.

Popliteus, plantaris, tibialis posterior, and flexor hallucis longus are absent. Triceps surae is represented by a single flat muscular belly arising from the head and upper end of the shaft of the fibula, the posterior part of the capsule of the knee joint, and, through the capsule, from the lateral condyle of the femur. The tendo calcaneus has a normal attachment. Flexor digitorum longus is double, consisting of a larger and a smaller belly arising together from the shaft of the fibula. The two tendons remain separate in the leg, but blend in the sole of the foot to give off five tendons to the lateral four toes and to the lateral of the two toes medial to the great toe. From these tendons six lumbricals arise and pass to the lateral side of the medial two and medial side of the lateral four toes. Flexor digitorum brevis divides into two parts, from the medial of which two tendons pass to the two most medial toes, while from the lateral two tendons pass to the true second and fourth toes. Abductor digiti quinti is normal; but abductor hallucis is absent, as is also quadratus plantae. Both oblique and transverse heads of adductor
hallucis are present. They join before being inserted into the base of the toe on the medial side of the hallux; and at their junction is attached a muscle—presumably the lateral head of flexor hallucis brevis—which arises from the metatarsal of the most medial toe. Six dorsal and four plantar interossei are present. The muscles of the lateral four interosseus spaces are normal. The additional three lie in the medial two inter-spaces, and consist of two dorsal interossei inserted on either side of the toe next to the hallux, and a plantar inserted on the lateral side of the most medial toe.

Tibialis anterior and extensor hallucis longus are absent. Two extensores digitorum longi are present. The medial and smaller muscle arises from the head of the fibula and front of the capsule of the knee and gives tendons to the medial two toes. The lateral has a normal origin from the fibula and supplies tendons for the lateral three toes and also to the base of the most lateral (i.e., the true fifth) metatarsal, though the line of separation of a peroneus tertius is not evident in the muscle itself. Extensor digitorum brevis is composed of three bands, of which two arise from the medial border and one from the lateral side of the dorsum of the tarsus. The lateral part gives off three tendons to the true second, third, and fifth toes, and then converges with the medial portions into an expanded tendon overlying the cuneiforms and inserted into the base of the metatarsal of the hallux.

Peroneus longus and brevis have a normal origin. The manner of insertion of their tendons is also normal, except that the tendon of peroneus longus gives off a slip attached to the calcaneus and extends across the sole to the bases of the most medial two metatarsals.

Three abnormal muscles are also present: (i) A large tibio-fibularus running downwards and medially from the upper half of the shaft of the fibula to the upper third of the tibia. (The proximal border of this muscle is attached to and separated from the cavity of the knee joint by a fibro-cartilaginous plate described below.) (ii) A small muscle arising from the front of the tibia close to the distal articular surface and passing distally to be inserted into the base of the most medial metatarsal. (iii) A slender muscle taking origin from the upper half of the shaft of the fibula deep to extensor digitorum longus, and inserted into the lateral side of the talus. This may be a peroneo-tarsus.

Bones.—The distal end of the femur consists of two imperfectly-formed condyles separated by a faintly-marked intercondyloid groove and fossa.

The tibia is represented by a short straight bone, stouter than but only one-third of the length of the fibula. Removal of a strip from its whole length shows that it is entirely cartilaginous. The normal subdivision of the bone into shaft and epiphyses is not evident, and it is probable that the epiphyses are entirely absent. The upper end is covered with articular cartilage on its proximal aspect, and articulates, a part of the cartilaginous plate intervening, with the side of the medial condyle of the femur. The distal end articulates directly with the talus.
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The fibula is outwardly curved in its long axis, but is not abnormally thick. The head is ill-defined and articulates with the lateral condyle of the femur, the cartilaginous plate intervening as in the case of the tibia. The distal end articulates with the lateral side of the talus.

Talus, calcaneus, and navicular are fused to form an irregularly-shaped bone. There are four cuneiforms all borne by the navicular part of this fused bone. The medial two cuneiforms support the medial two metatarsals (of which there are seven); the third cuneiform supports the metatarsal of the hallux and true second toe, and the fourth that of the true third toe. The cuboid articulates with the lateral two metatarsals.

The toes have been sufficiently described above. They all have the normal number of phalanges.

Articulations.—The articular surfaces entering into the knee joint are, as mentioned above, those on the distal end of the femur, the proximal end of the tibia and the head of the fibula. But the most interesting and important constituent of the joint is a plate of fibro-cartilage which covers the articular surfaces of both tibia and fibula, fills in the gap between these two surfaces, and blends with the capsule of the front and back of the joint. This cartilage is partly divided into two parts by a pad of fat and it receives part of the insertion of semitendinosus and semimembranosus. Probably it represents the menisci partly united. The transverse axis of the joint is abnormal, being parallel with instead of at right angles to the long axis of the leg. In other words, the femur articulates with the side instead of with the proximal end of the leg.

The tibia and fibula form separate articulations with the talus. No communication exists between the joints; but a strong ligament binds together the distal ends of the tibia and fibula.

In the articulations between the talo-calcaneo-navicular and the cuneiforms and the cuboid there is one joint cavity. Well-formed joints exist between the latter bones and the metatarsals, between those and the toes, and in the toes.

Summary.
1. The shortening of the leg.
2. The presence of an additional cuneiform and second and third metatarsals and toes, carrying additional interossei and lumbricals, on the medial side of the foot and to the medial side of the first metatarsal and hallux.
3. The absence of the tendons of some of the thigh muscles and of some of the muscles of the leg and foot.
4. The duplication of flexor and extensor digitorum longus and bifurcation of flexor digitorum brevis, corresponding to the additional medial segment of the foot.
5. The presence of a tibio-fibularus and of a probable peroneo-tarsus.
6. The imperfectly-formed femoral condyles and intercondyloid fossa.
7. The short cartilaginous tibia undifferentiated into its normal subdivisions and probably the shaft only, the epiphyses being absent.
8. The outward curvature and lack of hypertrophy of the fibula.
9. The formation of the knee joint between the distal end of the femur and the cartilaginous plate covering the medial side of the upper end of the fibula and the upper end of the tibia, the axis of the joint being in line with, instead of at right angles to, the long axis of the leg, and the cartilaginous plate probably representing the partly united menisci.
10. The separate articulation of tibia and fibula with the talus.
11. The fusion of talus, calcaneus, and navicular.

5. Thomas Bryant's case (1883)—R.C.S. Museum Teratological Series No. 439.

The specimen is the right leg of an infant. The leg has been removed by disarticulation at the knee. No description of the femur or knee joint is possible therefore. A superficial dissection had been carried out before the present dissection was undertaken; so that the condition of the skin and subcutaneous tissues cannot
be stated. Moreover it is not possible to state the difference (if any) of the size of the limb from the normal, as the age of the child from whom the specimen is derived is unknown.

The leg is straight, the fibula showing none of the curvature present in most specimens. The foot is inverted through 90°, its long axis remaining at right angles to the leg. There are five toes, of which the third and fourth show the condition of syndactyly; the first is short and only recognisable as the hallux because it possesses two segments.

Muscles.—(see Figs. 17, 18 & 19). The musculature of the limb is highly abnormal, in the absence of some muscles, in the irregularity of others, and in the presence of muscles which are not found in the normal limb. It is very difficult therefore to determine the place and value of the muscles seen on dissection. There are several very interesting and instructive features.

With the exception of gastrocnemius there is no trace of the thigh muscles which are inserted in the leg. In the leg all the muscles present are well developed, and were apparently healthy.

Peroneus longus and brevis are normal in size, origin, and insertion, except for the manner of insertion of the tendon of the former: this expands triangularly under the cuboid; the expansion is joined on the proximal side by the tendon of flexor hallucis longus (vide infra) while the lateral border merges into the fascia covering the muscles of the fourth interosseus space and the fifth metatarsal. The peroneus longus tendon runs through the expansion and then divides, one part ending in the medial cuneiform and base of the second metatarsal, the other running into the first dorsal interosseus muscle.

Extensor digitorum longus has a small fleshy belly arising from the anterolateral aspect of the upper end of the shaft and the head of the fibula, from which two tendons arise: the medial gives off a slip to the fourth toe, and another which, after partly fusing with the inner margin of extensor digitorum brevis, runs on to the third toe; the lateral supplies a tendon to the fourth toe, and another, which ends in the fascia covering the fourth interosseus space and undoubtedly represents a peroneus tertius tendon. The muscle arising in the position normal to peroneus tertius, however, has no connection with this tendon. It is a large muscle with a much extended origin, so that it arises from the whole length of the shaft of the
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fibula, immediately in front of the other peronei; but though the upper portion of its origin is in common with that of extensor digitorum longus, it soon separates from that muscle, and a strong tendon develops along its anterior border and runs to be inserted by two limbs into the under surface of the talus and the lateral surface of the calcaneus. This muscle is also undoubtedly peroneus tertius; apparently the muscle and its normal tendon have developed separately, or have become separated during development.

Extensor digitorum brevis is normal except that its most medial tendon runs to be inserted like a long extensor tendon into the terminal phalanx of the second toe, instead of into the base of the first phalanx of the first toe. This tendon is joined in its course by a small tendon from a muscle presumed from its site of origin to be extensor hallucis longus. Its origin is from the upper three-fifths of the shaft of the fibula immediately medial and deep to extensor digitorum longus and peroneus tertius, and it crosses anterior to the deep peroneal nerve and anterior tibial artery. The above-mentioned slip constitutes only a tiny portion of its tendon, the major portion of which runs round the medial border of the tarsus into the sole, where it takes part in the formation of a tendon from which the flexor digitorum longus tendons arise. This then is a muscle whose tendon has become diverted to make the muscle subserve a function very different from that of the normal. In direct continuation to extensor hallucis longus in its line of origin is another muscle, arising from the distal two-fifths of the shaft, whose tendon ends in a dorsal expansion over the distal part of the tarsus. Whether or not this is to be regarded as an additional part of extensor hallucis longus cannot be stated. Neither can it be said whether a tibialis anterior is present or not: there is no muscle having an insertion similar to that muscle, though there is a small muscle arising from the head of the fibula immediately in front of and on the same level as extensor digitorum longus which may be regarded as a tibialis anterior. This gives off a slender tendon which ends in a fibrous mass to be described below, though its fibres are traceable onwards into a tendon which fuses with that of extensor hallucis longus to form the flexor digitorum longus tendon mentioned above. A similar small muscle adjoining and partly fused with the above muscle has a tendon also joining the fibrous mass, and is possibly an accessory tibialis anterior.
On the posterior aspect of the leg the muscles can be named with greater certainty. Of triceps surae both heads of gastrocnemius are present, and soleus is a large muscle arising from the posterior aspect of the head of the fibula; the tendo calcaneus is inserted normally. Plantaris is absent, as is also popliteus. The large tibialis posterior has a normal origin from the posterior aspect of the head of the bone, and is separated from the intervening bone by the anterior tibial artery, probably represents part of the tibial origin of the muscle. The insertion is into the fibrous mass mentioned above. This is merely a fused mass of tendons attached to the upper surface of the talus and the capsule of the ankle joint. Arising from the shaft of the fibula immediately lateral to tibialis posterior is a large muscle which must be regarded as flexor digitorum longus, though it is inserted entirely into the fibrous mass. Flexor hallucis longus, though it has a normal origin, is also peculiar in that some of its insertion is into this mass. But more peculiar is the remainder of its insertion, which is by two tendons, both of which run into the sole of the foot, the one to end in the expanded peroneus longus tendon, the other into the fascia covering the fourth interosseous space.

From the fibro-tendinous mass formed by the fusion of the tendons as described, the origin of the tendon entering into the formation of the flexor digitorum longus tendon has been already mentioned. From it also one head of flexor digitorum brevis arises, the other head having the normal origin of the muscle. There are three tendons only to this muscle, running to the second, fourth, and fifth toes. The flexor digitorum longus tendon gives off four tendons to the lateral four toes. The lumbricals are normal in position and attachments. From the deep surface of the long flexor tendon an abnormal muscle arises; it is inserted into the heads of the third and fourth metatarsals, and its significance is obscure. Abductor digiti quinti, flexor digiti quinti brevis and the interossei are normal; but quadratus plantae, adductor and abductor hallucis, flexor hallucis brevis and the flexor hallucis longus tendon are all absent.

**Fig. 19.**
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Vessels and Nerves.—The main vessels and nerves are present and the arrangement of the main trunks (having regard to the altered position of muscles) is practically normal.

Bones and Joints.—The tibia is entirely absent without fibrous or cartilaginous representative. The fibula is straight and not abnormally thick. The proximal end bears a slightly concave articular surface on the postero-medial aspect of the head, which doubtless articulated with the femur. The articular surface on the distal end is in the normal position and is applied to the facet on the postero-lateral aspect of the talus. The cavity of the joint thus formed is small, but the capsule and ligaments are very thick and strong. The plane of this modified ankle joint is vertical. It is at this joint that the inversion of the foot is effected.

Talus, calcaneus and navicular are fused to form a compound bone. Each portion is fairly well formed and clearly recognisable. The sinus tarsi is absent. The compound bone through its navicular part articulates with two cuneiforms only. These are presumably the second and third, as they support the second and third metatarsals. These, and the fourth and fifth borne by the cuboid (normal in form and articulation), are normal; but the first metatarsal is much smaller and shorter than the others. It is composed of two segments—with a joint between—the distal of which is bony and is probably the true metatarsal, while the proximal is cartilaginous and attached by fibrous tissue to the second cuneiform, and probably is a very poorly developed first cuneiform.

There are five toes, which have been sufficiently described above.

Summary.
1. The irregularity of attachment of some of the muscles, viz.:—
   (a) The dissociation between the muscle belly and the normal tendon, illustrated by peroneus tertius.
   (b) The diversion of the normal insertion so that the muscle is made to subserve an entirely different and even opposite function, e.g., extensor hallucis longus supplying the major part of the long flexor tendon of the toes.
   (c) The abnormal insertion of a tendon running an otherwise normal course, e.g., that of flexor hallucis longus into the tendon of peroneus longus and the fascia of the fourth interosseous space.
   (d) The termination of some of the leg muscles in a fibrous mass with the origin of short flexor tendons therefrom (a condition analogous to that which is found in the apes), e.g., tibialis posterior and flexor digitorum longus joining the fibrous mass, and flexor digitorum brevis deriving partial origin therefrom.
2. The presence of abnormal muscles: for example, the muscle described as possibly a separate part of extensor hallucis longus; and that which springs from the deep surface of the long flexor tendon of the toes and is inserted into the head of the third and fourth metatarsals.
3. The absence of normal muscles: for instance, popliteus, quadratus plantæ and the intrinsic muscles of the hallux.
4. The complete absence of the tibia.
5. The absence of hypertrophy and curvature of the fibula.
6. The fusion of the calcaneus, talus and navicular.
7. The partial failure of growth of the first cuneiform and first metatarsal.

The most striking thing about the specimen is that the abnormal features are mainly confined to the inner border of both the leg and foot. Moreover, not only are bones absent or poorly developed in this area, but here also muscles have gone astray in their attachments, have failed to form, or appear in abnormal situations. It is evident thus that the disturbance of the mesoderm which must have underlain these abnormal features was mainly confined to the inner side of the limb, and was not confined solely to the bones, but extended to all the skeletal structures of the part.
Summary of Anatomical Appearances and Abnormalities.

From a consideration of the above cases personally dissected and described with the remaining specimens of the Royal College of Surgeons Museum, together with a study of the anatomical descriptions given by other
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writers, a general survey of the structure of, and abnormalities which may be found in, a limb with congenital absence of the tibia, can be given. (It may be remarked how few detailed dissections of specimens of congenital tibial defect have been undertaken. Of the English cases hitherto published, full anatomical descriptions have been given only by Symington and Craig, and by J. S. McLaren. In the reviews of the subject by Launois and Küss, and by Nuzzi, fairly full accounts of the pathological anatomy are given; but the accounts are deficient with regard to the musculature, merely stating that the abnormalities of muscle are extremely variable, inconstant, and of no practical importance—a statement which our own study proves to be fallacious.)

The most striking features are:—

1. The alteration in structure is not confined to the partial or complete absence of the tibia. In all the specimens personally examined, and in all of those described by other workers where a minute dissection has been performed, all the parts, mesodermal and often even epidermal in origin, have been affected.

2. Moreover, the disturbance is not confined to the leg, or even to the leg and foot, but affects the whole lower limb.

3. The abnormal appearances are found in the leg and foot in the main, though not entirely, on the medial (tibial) side, where, moreover, they may either be limited to one part or affect the whole.

4. The changes are not solely suppressive or atrophic. It may, in fact, be doubted if any of them are to be looked upon as "atrophic" in the literal sense of the word. Suppression undoubtedly occurs, as illustrated by the absence of bone or muscle; but disturbance of development of an opposite kind also is found, often conjointly, in the presence of abnormal and double muscles, and of extra segments of the tarsus, metatarsus or toes. Noteworthy also is the constantly healthy condition of the parts, particularly the muscles, some of which may even be much larger than normal, and none of which show degeneration of any kind.

5. The curious and, in interpretation, often puzzling arrangement of the musculature in many specimens, particularly in regard to the dissociation between the muscle belly and the normal tendon, and to the extraordinary transposition of tendons into insertions which make muscles subserve functions which are the reverse of the normal.

6. The tendency to reversion to the structure of lower species, seen in the fibulo-femoral articulation, and in the musculature, e.g., by the presence of such muscles as tibio-fibularis.

7. The presence and persistence without wasting or degeneration of a muscle, seen in one or two specimens, unable to contract and apparently functionless: a physiological curiosity.

8. The tibia may be entirely or partly absent, or may be represented either by fibrous tissue, tendon, or cartilage.
9. The frequent association of dimples, grooves, constrictions and umbilications of the skin of the leg.

The investigation of the anatomy of the thigh has been confined in our own and all other recorded cases (except in one of Ollerenshaw's cases, where, however, no detailed account is given) to an examination of the structure of the distal end as seen at operation and of the tendons of thigh muscles which are inserted in the leg. With regard to the hip, however, Launois and Küss state that luxation is very frequent, and Nuzzi mentions it as an associated abnormality sometimes present. It was not present in any of the cases we have examined; but in Laming Evans's second case a skiagram showed absence of the ossific nuclei of the head of the femur and of the pubic bone on the same side. In most cases the distal end of the femur is imperfectly formed: the condyles and the intercondyloid fossa may be rudimentary or absent. In cases of partial defect of the tibia where the upper end has been present, the femur is usually normal. The thigh muscles which pass to the leg are frequently abnormal, absence of one or more being a constant feature in our specimens and alterations of insertion being seen frequently. The length and circumference of the thigh are commonly slightly smaller than those of the opposite side.

The patella, contrary to the statements made on clinical observation alone in the earlier recorded cases, is present in most cases, though it may be small and its ossification delayed. This is what one would expect, since the patella is not to be regarded as a homologue of the olecranon depending on the existence of the tibia, but as a true sesamoid of the quadriceps tendon, and thereby a part of the first and not of the second segment of the limb. That it is frequently small and sometimes absent is probably due to the fact that the quadriceps itself may be ill-developed or deficient.

A well-formed joint between the thigh and leg is usually present. Where the upper end of the tibia is present the knee joint differs little if at all from the normal. When the tibia is completely absent the joint is formed between the proximal end of the fibula and the distal end of the femur, on its posterolateral aspect in most cases. Where no joint exists the fibula is bound to the femur by fibrous tissue on its posterior or lateral aspect, and may be displaced upwards some distance into the thigh. A point of practical importance is that the articular surface on the fibula frequently transgresses the epiphyseal line, which may therefore be interfered with by attempted arthrodesis. The menisci and cruciate ligaments are absent in cases of total defect of the tibia.

The tibia may be totally or partially absent, or represented in whole or in part by fibrous, tendinous or cartilaginous elements. In the specimens which have been examined by ourselves and others every grade of tibial defect and representation has been observed: (a) total absence; (b) absence of the lower third or two-thirds (the usual condition in cases of partial defect), or
of middle third; (c) proximal epiphysis or upper end only (often conical with pointed projecting lower end attached to an umbilicated patch of skin); (d) absence of part of the shaft with a fibrous band uniting the upper and lower fragments; (e) fibrous or tendinous strands with and without attachment to fibro-cartilaginous masses at the either end of the leg. In one specimen the cartilaginous shaft without sign of ossification appears to be alone present, the epiphyses being absent.

The fibula is always shorter than normal and usually stouter and incurved. The latter two features are not invariably present: they are absent in three of the specimens we describe. The interosseous membrane may be present where the tibia is merely represented by tendon, or absent and its place taken by muscle where the tibia is actually present. In cases of total absence the membrane is not found.

The ankle joint is formed when the tibia is totally absent between the fibula and the talus, or when the latter bone is absent, the fibula and calcaneus. A representative of the tibia may take part in the formation of the joint. Usually the medial side of the distal end of the fibula articulates with the postero-lateral aspect of the body of the talus, which is thus luxated: it is at this joint, in fact, that most of the varus deformity of the foot takes place.

The bones of the foot share in the abnormality. Almost always there is malformation and fusion of the calcaneus and talus and sometimes also of the navicular. Absence of one or more of the bones of the inner segment of the foot—talus, navicular, cuneiforms, inner three metatarsals and toes, especially the hallux and its metatarsal—is common; but almost equally so is the presence of extra elements—a pre-hallux, cuneiform and particularly a supernumary great toe. Moreover the two conditions may occur in the same limb, a state of affairs which clearly indicates that the cause is not merely an arrest of growth.

The musculature also always shows marked abnormality of structure and arrangement. There may be absence of normal muscles, duplication of normal muscles, presence of additional abnormal muscles, or irregularities of insertion, ranging from mere diversion of tendons running an otherwise normal course, to actual dissociation between muscle belly and the normally inserted tendon. Finally, attention may be drawn again to the occurrence of muscles attached entirely to one bone and persisting, apparently healthy though functionless.

The blood vessels and nerves have been poorly described in all published accounts. In our own specimens the parts were too small to allow of a complete dissection of the nerves and vessels; and in fact such a dissection was considered unnecessary. In these specimens the vessels and nerves appeared perfectly healthy and the main trunks were present and little if at all abnormal in their arrangement. Where they are mentioned in the writings of others they are usually stated to be "practically normal."
Bertaux, who used the method of electrical stimulation to study the anatomy of his specimens, noted the absence of the reaction of degeneration, and found in four legs no abnormality except in one case where the deep peroneal nerve was absent with the muscles it normally supplies.

Cutaneous grooves, constrictions, dimples, umbilications and "scars" occur in the affected limb frequently, particularly over the head and shaft of the fibula, and over the free end of the tibia in the case of partial defect. These skin markings are not true scars: as was proved by histological examination, which showed the absence of scar tissue and mere diminution of the deeper layers of the epidermis at the site of the depression.

VI.—\Etiology.

The current theories of the cause of congenital bone defects are:

1. The germinal; (2) the mechanical (including the postural and the amniotic); (3) the traumatic.

The germinal theory regards as primary a defect of the mesoblast from which the skeleton of the affected limb is developed. To what this defect is due has not been ascertained; but vascular, microbial, toxic, and endocrinal influences are, without proof, invoked.

The mechanical theories state that interference with normal development and growth is brought about by external pressure on the fetus, either, according to the postural theory, by malposition in utero; or, according to the amniotic theory of Dareste\(^9\) by the abnormal tension of the amnion, itself arrested in development, or by amniotic bands or adhesions formed between the amnion and fetus as the result of inflammation of the amnion, or by constrictions effected by the umbilical cord.

The traumatic theory regards the cause as violence sustained by the mother during pregnancy and transmitted to the fetus as it lies in utero.

Our study has led us to believe that the theory of primary mesoblastic defect is correct, and that mechanical and traumatic influences are not concerned in the production of congenital bone defects. This view, we believe, is the one now held most widely, and we claim that its truth is now established.

Our investigation of the anatomy has made it clear that the disturbance of development is not confined merely to the tibia, but extends to all the mesoblastic structures of the limb. It has shown also that the disorder is not only of the leg, but of the whole of the limb and even of the pelvis. And further, it has demonstrated that the changes are not solely suppressive, supernumerary segments, bones and muscles, being almost as often present as normal parts are absent.

The clinical study has revealed that other abnormalities so widely different from the particular one we are considering as cleft palate and cryptorchidism are very frequently associated; and that there is a definite, though not invariable, tendency for the defect to be hereditary and familial.
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in occurrence. In the latter case, moreover, the defect may be identical in two or more members of the family. Another point has also come out: that when the defect is bilateral there is frequently remarkable symmetry of the affected limbs.

It is manifest from these facts that the cause of congenital bone defects is intrinsic and not extrinsic in action. It is inconceivable that external pressure or violence could bring about the anatomical abnormalities. The suggestion that either of these influences do so becomes highly improbable when we call to mind that they would have to act as early as the sixth week, when the foetus is only three or four centimetres in length, for by this time the muscles and bones are clearly differentiated.

The traumatic theory is mainly based on the assumption that the "scarring" of the skin and marked angulation of the fibula sometimes seen are due to intra-uterine laceration and fracture, followed by resorption of the tibia. The above clinical and anatomical facts, the frequent absence of any history of trauma, lack of angulation or "scarring," and the histological proof that the skin markings are not true scars, render this theory untenable.

There is little evidence also to support the mechanical theories. They seem to rest principally on the frequent presence of cutaneous markings on the outer side of the leg and foot and of outward curvature of the fibula, and on the usual limitation of the defects to the lower limbs with associated normal development of the rest of the body. The theories are thus based on very slender evidence; and the facts cited above are enough to disprove them. It is probable that the amniotic anomalies are associated abnormalities due to the same cause as the foetal mesoblastic defects. Although the cutaneous constrictions may be caused by an amniotic band or the umbilical cord, the dimples, cutaneous scars and grooves are more probably not due to these agents, but are caused directly by the disturbance of limb growth. The facts that they are not true cicatrices, are not present in very many cases, and, when present, are not always at sites where pressure would have occurred, favour the latter view. If these theories were true one would expect to find in records of cases a note that birth was difficult or otherwise abnormal. Such a note is absent in most cases, while in not a few the reverse is stated.

The evidence is, in our opinion, overwhelmingly in favour of the theory of a primary defect of growth of the mesoblast. This opinion is strongly supported by the recently published investigations of Hovelacque and Noel(49). They observed the development of the lower limbs in a breed of mice commenced by Rabaud in 1909, in which members afflicted with congenital absence of the tibia made their appearance first in 1913, and occurred continuously from then till 1920. They studied both normal and abnormal embryos of every age and size; and their conclusions are summarised here:
1. The first manifestations of the anomaly are revealed when the undifferentiated blastema first commences to undergo modification.

2. There is no modification in the outline of the fibula; at most there is a slight delay in the appearance of the successive stages.

3. The anomaly is manifested solely in the zone of blastema where the tibia would normally develop. A fibrous tract is formed here, and between it and the fibula the interosseous membrane appears.

4. In certain embryos cartilaginous nodules develop in this zone of blastema, mainly at the upper part near the femur. The continuity of the nodules and the fibrous tract, and the fact that they are parts of the same structure, have been definitely established.

5. Throughout the complete evolution, vascularisation is normally developed in the whole extent of the structure. Thus the idea of a vascular origin of the anomaly may be cast aside.

These observations are very convincing, and with the anatomical and clinical facts we have been able to bring forward, establish, we think, the truth of the germinal theory. With regard to this we feel that it is important to emphasise that the mesoblastic defect is not merely one of temporary or permanent cessation of growth. It is obviously a more profound disturbance than this, since it involves arrest of growth in some elements of the limb with associated abnormal or excessive development of other elements. The underlying cause of the defect is quite unknown. There are no reasons for assuming that it is endocrinal, microscopic or toxic action. The subjects of congenital bone defects rarely show evidence of syphilitic or other faetal disease, and are often born of perfectly healthy parents. Nor are there any strong reasons to support an atavistic theory. Occasional features in dissected specimens suggest reversion to those of lower animal orders; but these are not sufficient evidence on which to base an atavistic theory. Certainly there is nothing to support Gegenbauer's suggestion of suppression of a tibial "fin-ray." Apart from the fact that at no moment in the development of the limbs of the human embryo is there a state comparable to that of the fin of fishes, it is impossible by this theory to reconcile absence of the tibia with the presence of a normal foot and still more of polydactyly. Thus, while it is clear that the cause of congenital bone defects is primarily in the mesoblast itself, the underlying reason for this remains obscure.

VII.—TREATMENT.

Congenital absence of the tibia has always formed a difficult problem for surgeons, many of whom have taxed their ingenuity in endeavouring to devise means of converting the useless limb into one useful by itself or when aided by an appliance. The great majority of conservative measures attempted have failed; and the reason for this has been the deficient knowledge of the structure and manner of growth of limbs affected with the anomaly. Our study has made the indications for treatment clearer.
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In considering these it is necessary to bear in mind that the anomaly is a gross one involving the whole lower limb and not merely the obviously affected segment; that with the tibial bone defect are associated almost invariably defects or other abnormalities of other bones of the limb, and a musculature highly irregular in arrangement and development; and that the deficiency in the limb is not only one of development, but also one of growth. The last is a cardinal point, for, because of it, the leg, abnormally short at birth, becomes progressively shorter relatively to the sound limb as the age of the patient advances towards maturity. This fact has a very important bearing on the question of prosthesis both without and following conservative operative interference, and on that of bone-grafting to replace the congenital defect. Reference will be made to it again.

Until 1875 amputation seems to have been the only surgical procedure undertaken. The credit of being the first to attempt a conservative operation belongs to Albert(3), who in 1877 performed arthrodesis of the knee in a case of total tibial defect by grafting the fibula into the intercondylloid fossa of the femur. He has been followed since then by a number of other surgeons who have either adopted his method or effected the arthrodesis by removal of the cartilage of the fibular and femoral articular surfaces. Combined with one or other of these procedures arthrodesis of the ankle joint by similar methods, and correction of deformities of the knee and foot, were sometimes performed. Attempts have been also made, in cases of partial absence of the tibia, on the same principle of making the fibula take the place of the tibia, to effect union between the free end of the tibia and the fibula—either to the side of the bone or to the distal fragment after osteotomy (Nové-Josserand(21)), or to the head of the bone freed and drawn down (P. Bade(50)). The result is said to have been satisfactory in several cases where one of these measures was done, and in one or two excellent, the patient walking without the aid of an instrument. In a few cases, notably that of Fraser and Robarts(12), another principle, the replacement of the defect by bone-grafting, has been followed. In the case quoted both proximal and distal ends of the tibia were present and the fibula of the normal limb was transplanted into the gap with, it is claimed, a satisfactory result. Of recent years the tendency has been again towards amputation except in selected cases of small partial defects.

It is important to distinguish between cases of total and cases of partial defect, and to consider the question of operative or non-operative treatment of each.

In cases of total defect our opinion is that operative intervention is indicated in all cases whether the abnormality is unilateral or bilateral. We do not agree with the statement of Nuzzi(22) that orthopaedic apparatus assures good and correct functioning of the affected limb. An appliance is unavoidably cumbersome and must be progressively raised to compensate for the shortening and we do not advise it. The operation should be that
of amputation at the knee. We are opposed to arthrodesis of the knee and ankle. In the young subject they involve a great danger of damage to the epiphyseal cartilages, and thereby of interference with the already deficient growth; they do not abolish the necessity for an orthopaedic appliance; and the anatomical peculiarities prevent the formation of a useful limb by this procedure even when combined with the correction of deformities. A bone graft, or transplantation, of the opposite fibula, is also obviously inadvisable in total tibial defects: even if the graft survives it cannot prevent the progressive shortening of the limb upon which depends a great part of the deficiency of function. Except in adult subjects with well-formed femoral condyles we favour disarticulation at the knee rather than supracondylar amputation. By disarticulation interference with the growth of the femur is avoided. The disadvantages of this procedure in a normal limb, namely, the difficulty of fitting a prosthesis over the broad distal end of the femur and of making the artificial knee joint at the same level as the knee joint of the sound limb, do not apply in most cases of congenital absence of the tibia. In them the thigh is usually half an inch or more shorter than that of the
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healthy side, and the femoral condyles absent or poorly developed. Even if platymeria or forked platymeria of the condyles were present we would prefer to excise or round off the condyles rather than to amputate above them. It is of little importance whether or not the patella is left in the flap; on the whole it is better to remove it. The operation should be undertaken early, preferably during the second year when the child is attempting to walk. If it is done then and an extensible peg-leg applied, and later, if desired, an artificial limb, the normal education of the child can proceed uninterruptedly, and the femur and thigh muscles will not suffer from disuse atrophy. The operation of disarticulation at the knee in infancy is attended with no or very little shock, and the only objection to it at this age is the possibility of the later formation of a conical stump. There is, however, no need to fear this. (See Fig. 20.)

In partial defects we also advise amputation, except when there is present a considerable portion of the proximal part of the tibia with a well-formed knee and a foot which is plantigrade or can be made so. The same objections to an orthopaedic appliance and to conservative operations hold good, though to a less degree, for partial as for total defects. Unless the defect is such as stated in the above reservation conservative measures hold out little hope of achieving a good functional result. The modern artificial limb is far more useful, less noticeable, and less cumbersome than any orthopaedic appliance which can be fitted to one of these cases, whether a conservative operation has or has not been performed. A prosthesis is almost always needed after such an operation, because the operation cannot correct the shortening of the leg, and it cannot make the leg strong enough to bear the weight of the body steadily on a raised boot. The prosthesis is difficult to apply, inevitably heavy and clumsy, and cannot compare in efficiency with an artificial limb.

In the type of case mentioned above as a reservation it may be worth while to attempt a reconstructive operation. The principle of this will be either to make the fibula do the work of the tibia by uniting the distal end of the defective tibia to the fibula, or to obliterate the defect of the tibia by transplanting the fibula from the sound limb into it, or by a grafting of an autogenous piece of bone. Operations to stabilise the ankle and to correct deformity of the foot and knee should be undertaken later if necessary.

The best results from conservative procedures are to be expected where there is present a large portion of the tibia, especially if both the proximal and distal ends exist, and the proximal end with the knee joint is well-formed. Success cannot be expected if the defect is of the proximal end.

VIII.—CONCLUSIONS.

1. Congenital absence of the tibia is one of the rarest of congenital malformations and the rarest malformation of the lower limb. The number of recorded cases is now 105.
2. Tibial absence is rather more commonly total than partial, unilateral than bilateral, and right-sided than left sided.

3. Other abnormalities are frequently associated; but rarely is there any evidence of fetal disease.

4. The characteristic signs are: (a) Shortening of the leg; (b) flexion and adduction of the knee; (c) inversion of the foot. Radiographic examination is very valuable to establish the diagnosis and to reveal other anomalies of the bones of the limb.

5. The diagnosis is not difficult. Fibular defects and congenital equinovarus need elimination.

6. The anomaly involves the whole lower limb and not merely the tibia. Defects and other abnormalities of other bones of the limb and a musculature highly irregular in development and arrangement, accompany the defect of the tibia.

7. The defect is of both development and subsequent growth, so that there is progressive shortening relative to the sound leg till maturity is reached.

8. The changes are not solely suppressive. Supernumerary bones and muscles and abnormal muscles are almost as often present as others are absent. Moreover, the changes are not atrophic or degenerative.

9. The tibia may be totally or partially absent, or represented in whole or in part by fibrous tissue, tendon, or cartilage. In partial defects it is almost always the distal part of the bone which is absent. Absence of the proximal end and of the middle portion have been each recorded once only.

10. The cause is defective development of the mesoblast under an influence at present unknown, but not traumatic, mechanical, vascular, or atavistic. Amniotic anomalies when present are probably due to the same influence. The mesoblastic changes have been observed in fetal mice from the earliest stages of tissue differentiation.

11. Disarticulation at the knee followed by the application of a peg-leg or artificial limb is the best line of treatment. Conservative operations are indicated only in cases of partial defect when the gap is small and limited to the middle portion or distal end of the tibia. In all other cases treatment by conservative operations or by orthopaedic appliances is inadvisable. Conservative operations should be based on one of the following principles: (1) The utilisation of the fibula to take the place of the tibia; or (2) the insertion of an autogenous graft to fill the gap in the tibia. An appliance is almost always needed subsequently and the results are rarely satisfactory.

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IX.— Bibliography.

(Note.—Only works of reference and names and cases referred to in the text are given here. Good bibliographies are given by Launois and Küss, Nuzzi, and Bertaux).
