

## Rare case of tibial hemimelia, preaxial polydactyly, and club foot

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### Abstract

A seven-month old female presented with left tibial hemimelia (or congenital tibial aplasia; Weber type VIIb, Jones *et al*/ type 1a), seven-toed preaxial polydactyly, and severe club foot (congenital talipes equinovarus). Definitive amputation surgery disarticulated the lower limb at the knee. This case report describes the anatomical findings of a systematic post-amputation examination of the lower limb's superficial dissection, X-rays, and computed tomography (CT) scans. From the X-rays and CT scans, we found curved and overlapping preaxial supernumerary toes, hypoplastic first metatarsal, lack of middle and distal phalanges in one supernumerary toe, three tarsal bones, hypoplastic middle phalanx and no distal phalanx for fourth toe, and

no middle or distal phalanges for fifth toe. The fibula articulated with the anteromedial calcaneus and the tibia was completely absent. We identified numerous muscles and nerves in the superficial dissection that are described in the results section of the case report. Due to the rarity of this combination of anatomical findings, descriptions of such cases are very infrequent in the literature.

**Key words:** Club foot; Congenital talipes equinovarus; Preaxial polydactyly; Tibial hemimelia; Congenital tibial aplasia

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**Core tip:** This case report analyzes the anatomical findings associated with systematic post-surgical examination of the amputated lower leg of a seven-month old female presenting with a very rare combination of lower limb malformations. These include left tibial hemimelia (or congenital tibial aplasia; Weber type VIIb, Jones *et al* type 1a), seven-toed preaxial polydactyly, and severe club foot (congenital talipes equinovarus). This case report describes the results of the amputated limb's superficial dissection, X-rays, and computed tomography scans. Due to the rarity of this combination of anatomical findings, descriptions of such cases are very infrequent in the literature.

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## INTRODUCTION

Tibial hemimelia (TH), also known as congenital tibial aplasia, is a rare congenital anomaly characterized by deficiency of the tibia with a relatively intact fibula<sup>[1-4]</sup>. The incidence of TH in the United States is approximately one in 1000000 live births<sup>[1-3]</sup>. It is further subdivided into different types, making each subtype even rarer.

TH can occur as an isolated hereditary malformation as well as a feature of several autosomal recessive and autosomal dominant syndromes, involving accompanying congenital defects<sup>[2,4,5]</sup>. Weber<sup>[3]</sup> has created the most detailed classification system for tibial malformations to date, classifying and scoring the main pathological findings of the complete leg, which include coxa, femur, patella, tibia, fibula, and pes. Potential causes of TH include errors in several different aspects of limb development<sup>[5]</sup>. Other known causes of TH are ingestion of thalidomide or inheritance of an autosomal dominant gene; more sporadic cases are idiopathic<sup>[6]</sup>.

Polydactyly is a common congenital hand and/or foot anomaly that is characterized by the presence of supernumerary digits<sup>[7]</sup>. This condition has an incidence rate of 1.7 per 1000 births. It may be isolated or associated with established genetic syndromes<sup>[7,8]</sup>. Several morphologic classification types for polydactyly exist, including those based on metatarsal variations with duplicated distal phalangeal segments<sup>[7,9]</sup>, those expanding the two preaxial types to four<sup>[7,10]</sup>, and those that add mirror foot or diplopodia<sup>[7,11]</sup>.

Club foot or congenital talipes equinovarus (CTEV) is a complex, fixed deformity of the lower limb that is always present at birth and can be unilateral or bilateral<sup>[12]</sup>. CTEV is a multiplanar deformity of the lower leg with a prevalence of one to three per 1000 live births<sup>[13,14]</sup>. It is twice as common in males and at least half the cases are bilateral<sup>[14-16]</sup>.

There are three basic components of CTEV: Equinus, varus, and adduction deformities<sup>[14]</sup>. The severity of the club foot deformity is classified according to the Pirani scoring system<sup>[17]</sup>. There are three classifications of CTEV: Postural, idiopathic, and teratogenic or syndromic<sup>[15,16]</sup>. The cause of CTEV is unknown<sup>[14,16,18,19]</sup>, although there is a clear multifactorial inheritance pattern with many potential environmental influences<sup>[16]</sup>, including amniotic injury or hemorrhage, and positioning *in utero*<sup>[14]</sup>, and retracting fibrosis<sup>[18]</sup>.

Although each of the aforementioned syndromes is relatively rare, comorbidity of all three of these disorders is even rarer, if not unique. This case report discusses the anatomical findings associated with a 7-mo old female presenting with all three of these conditions occurring simultaneously.

## CASE REPORT

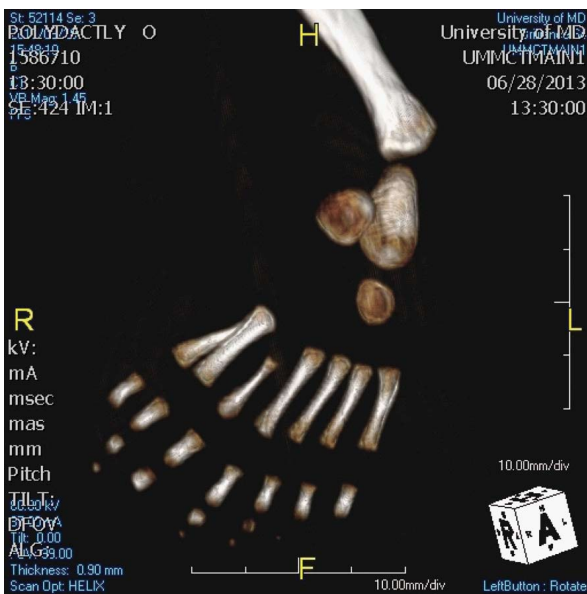
A seven-month old female presented with TH, seven-toed preaxial polydactyly and severe left CTEV. There was also medial contracture of the left foot and lower leg with left congenital hip dysplasia. In addition to having no left tibia, she also did not have a left patella or an active quadriceps mechanism. The left fibula was bowed and articulated with the lateral side of the femur. Due to the severity of the deformity, and the unlikelihood of a successful limb salvage procedure, amputation was advised.

Definitive amputation surgery was performed; the amputation involved a left knee disarticulation of the lower limb. During the surgery, the musculature of the anterior, lateral, and posterior compartments of the left lower leg was transected to provide coverage to the stump. This made post-mortem identification of muscles and vessels that were present prior to surgery challenging. The operative report described the presence of the posterior tibial nerve and deep peroneal nerve with the absence of a posterior tibial artery.

The family of the child elected to donate the amputated specimen to the Maryland State Anatomy



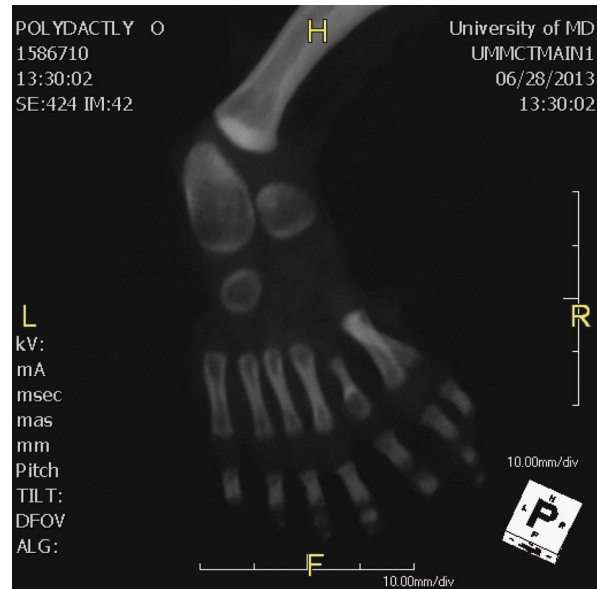
**Figure 1** Post-amputation superficial dissection demonstrating complete fibula, and supernumerary toes with toe A (purple arrow) and toe B (red arrow), with toe A being closest to the hallux.



**Figure 2** Computed tomography scan demonstrating skeletal characteristics with regard to the fibula, tarsal bones, metatarsals, and phalanges.

Board and releases were obtained. A post-amputation superficial dissection on the amputated lower limb of the child was followed by X-rays and CT scans. A detailed superficial dissection was conducted to identify any and all anatomical structures present.

For description purposes, the supernumerary toes were identified as toe A and toe B, with toe A being closest to the hallux (Figure 1). Using the Weber method, the child's TH condition can be classified as type VIIb, class 4 (total points 8)<sup>[3]</sup>. Using the classification of Jones *et al.*<sup>[20]</sup>, this child's TH condition can be classified as type 1a<sup>[2]</sup>. Using Venn-Watson and Watanabe method, the polydactyly exhibited can be categorized as a complete medial ray duplication with a hypoplastic medial member<sup>[7,9,21]</sup>.



**Figure 3** Plain radiograph showing skeletal characteristics with regard to the fibula, tarsal bones, metatarsals, and phalanges.

From studying the plain X-rays and CT scans, we found the following skeletal characteristics with regard to the fibula, tarsal bones, metatarsals and foot phalanges: Curved and overlapping supernumerary toes, hypoplastic first metatarsal, lack of middle and distal phalanges in toe B, only three tarsal bones present: Calcaneus, talus, and navicular, no distal phalanx for fourth toe, hypoplastic middle phalanx for 4<sup>th</sup> toe, and no middle or distal phalanges for fifth toe. The fibula articulated with the anteromedial portion of the calcaneus (Figures 2 and 3).

The extensor muscles visible after superficial dissection of the lower leg and foot include peroneus longus, peroneus brevis, and peroneus tertius. Extensor digitorum longus is present with its tendons connecting to between toes one and two, and three through five. The extensor digitorum brevis is also present with its tendons connecting to toes two through five, and extensor hallucis longus with its tendon connecting to toe A and not the hallux (Figure 4).

The flexor muscles visible include a portion of the gastrocnemius that is attached to the Achilles tendon. The Achilles tendon itself appears to be attached to the medial side of the calcaneus. Flexor digitorum brevis appears to connect with toes A and B then toes two through four. The flexor hallucis longus tendon does not appear to be present. These two characteristics may contribute to the hyperextensibility of the hallux on manipulation. We were unable to find the tendons of flexor digitorum longus, the muscle bellies of quadratus plantae, or the muscle belly of flexor hallucis brevis. Due to transection during surgery, we cannot identify tibialis anterior as being present. The tendon to tibialis posterior is visible running alongside the medial plantar nerve and deep to the flexor digitorum brevis muscle.



Figure 4 Superficial dissection demonstrating extensor muscles visible in the lower leg and foot.



Figure 6 Superficial venous arch visible upon initial superficial dissection (white arrows).



Figure 5 Superficial dissection demonstrating flexor muscles visible in the lower leg and foot, and nerves showing a supernumerary branch of the lateral plantar nerve connecting to toe A (light blue arrow).

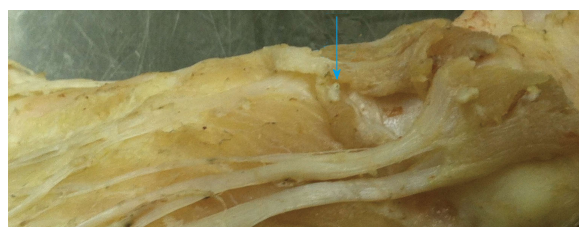


Figure 7 Dorsalis pedis artery penetrating into the deep compartment of the foot (blue arrow).

lumen with a relatively thick wall suggestive of an artery (Figure 7). We cannot determine if the deep venous arch is present from the superficial dissection. Posterior tibial artery is not visible and is also specifically described as absent in the operative report.

## DISCUSSION

TH, also known as congenital tibial aplasia, is a rare congenital anomaly characterized by variable degrees of deficiency of the tibia with a relatively intact fibula<sup>[1-6]</sup>. The incidence of TH in the United States is approximately one in 1000000 live births<sup>[1-3]</sup>. Children born with this abnormality usually exhibit marked shortening and bowing of the involved leg. There is also pronounced flexion contracture and instability of the knee, variable leg rotation, and marked inversion and adduction of the foot<sup>[1,2]</sup>. The first metatarsal is usually hypoplastic or markedly shortened and frequently associated with other medial ray defects and/or preaxial polydactyly<sup>[1,11]</sup>.

TH can occur as an isolated hereditary malformation as well as a feature of several autosomal recessive and autosomal dominant syndromes<sup>[4,5]</sup>. Seventy-five percent of patients who have TH have accompanying congenital defects. These can include congenital hip dislocation, polydactyly, medial foot defects, severe club foot, bifurcated femur, central hand defects, cleft palate, spinal anomalies, and a wide range of congenital

The transverse and oblique heads of the adductor hallucis muscle appear to span the supernumerary toes and hallux. In addition, the following muscles appear to be present: Abductor hallucis, flexor digiti minimi, abductor digiti minimi. Lumbricals appear to be present and there is a possible supernumerary lumbrical connecting with toe A and/or B. From this superficial dissection, none of the interossei is visible (Figure 5).

Nerves visible include posterior tibial, medial and lateral plantar nerves, with lateral plantar nerve having a supernumerary branch to Toe A while Toe B has a medial plantar nerve branch (Figure 5). Deep peroneal nerve is not visible within the superficial dissection but is apparently present as stated in operative report. Superficial peroneal nerve is not visible and is not mentioned in the operative report.

The vessels visible upon dissection are the components of the superficial venous arch (Figure 6). We have also identified a structure that could be dorsalis pedis due to the following characteristics: It penetrates into the deep compartment of the foot and it has a

anomalies of the cardiovascular, gastrointestinal, and genitourinary systems<sup>[2,4]</sup>.

Weber<sup>[3]</sup> has created the most detailed classification system for tibial malformations to date. He classified and scored the main pathological findings of the entire leg, which include coxa, femur, patella, tibia, fibula, and pes. Tibial malformations are divided into seven main groups and five of them are divided further into two subgroups. The seven main groups are: Type-I, hypoplasia; type-II, diastasis; type-III, distal aplasia; type-IV, proximal aplasia; type-V, bifocal aplasia; type-VI, agenesis with double fibula; type-VII, agenesis with a single fibula. The subgroup for types III through VII are a (with cartilaginous anlage), and b (without cartilaginous anlage). Three types of muscle function are described as +, present; (+), partly present; and -, absent for the coxa, femur, and tibia. Scoring of the various types ranges from zero to 39; the higher the score, the less the impairment grade<sup>[3]</sup>.

There is an older TH classification system described by Jones *et al.*<sup>[2,20]</sup>. In type 1a, the tibia is completely absent and the distal end of the femur is hypoplastic. In type 1b, a rudimentary tibia articulates with the distal end of a relatively normal femur. In type 2, the proximal end of the tibia is well developed and the distal end of the tibia is absent. In type 3, the tibia is represented by a characteristically amorphous bone segment that can be present more distally than proximally. In type 4, the proximal end of the tibia is normal with a shortened distal end and a characteristic congenital diastasis of the ankle joint<sup>[2]</sup>.

Potential causes of TH include errors in several different aspects of limb development. Cells from the primitive streak have been shown to migrate to the lateral plate mesoderm. Such an abnormality of this process could affect the limb bud which forms from the mesenchyme cells in the lateral plate mesoderm<sup>[5]</sup>. Other known causes of TH are ingestion of thalidomide or inheritance of an autosomal dominant gene; more sporadic cases are idiopathic<sup>[6]</sup>.

Polydactyly is a common congenital hand and/or foot anomaly that is characterized by the presence of supernumerary digits. It is classified as preaxial, central, or postaxial depending on the location of the duplication. Approximately 15% of all toe duplications are preaxial at the first ray, about 79% of duplications are postaxial and involve the lateral-most rays of the foot, and the remaining 6% are central including the second through fourth rays<sup>[7]</sup>. This condition has an incidence rate of 1.7 per 1000 births. It may be isolated or associated with established genetic syndromes. If polydactyly is non-syndromic, 30% of patients have a positive family history, which is most often expressed as autosomal-dominant inheritance with variable penetrance. No sex predilections have been identified<sup>[7,8]</sup>.

Several researchers have devised morphologic classification types for polydactyly. Venn-Watson<sup>[9]</sup>

created a classification system based upon the anatomic configuration of the fifth and first metatarsals and the duplicated bony parts. It recognized six metatarsal variations with duplicated distal phalangeal segments: Y-shaped fifth metatarsal; T-shaped fifth metatarsal; widened fifth metatarsal head; complete fifth metatarsal duplication; short, block first metatarsal; and widened first metatarsal head<sup>[7,9]</sup>. Masada *et al.*<sup>[10]</sup> created a variation of the Venn-Watson classification, expanding the two preaxial types to four: Type 1, complete metatarsal (ray) duplication; type 2, complete phalangeal duplication; type 3, incomplete metatarsal duplication; and type 4, incomplete phalangeal duplication<sup>[7,10]</sup>. Watanabe *et al.*<sup>[21]</sup> devised their own morphologic classification based upon ray involvements and levels of duplication. They classified foot polydactyly into three broad groups: Medial ray, central ray, and lateral ray with each group further subdivided into tarsal, metatarsal, proximal phalangeal, and distal phalangeal subgroups<sup>[7,21]</sup>. Belthur *et al.*<sup>[7]</sup> includes an additional categorization to that of the Watanabe *et al.*<sup>[21]</sup>'s morphologic classification known as mirror foot or diplopodia<sup>[7,11]</sup>.

Club foot or CTEV is a complex, fixed deformity of the lower limb that is always present at birth and can be unilateral or bilateral<sup>[12]</sup>. CTEV is a multiplanar deformity of the lower leg with a prevalence of one to three per 1000 live births<sup>[13,14]</sup>. It is twice as common in males and at least half the cases are bilateral<sup>[14-16]</sup>. Most occurrences of CTEV are sporadic, but cases of families with CTEV as an autosomal dominant trait have been reported<sup>[14]</sup>. In addition, the risk of having a child with CTEV is around 20 to 30 times greater when a first-degree relative is affected<sup>[16]</sup>. Approximately 20% of CTEV cases are associated with other congenital abnormalities<sup>[19]</sup>.

There are three basic components of CTEV: Equinus, varus, and adduction deformities<sup>[14]</sup>. The talus is severely flexed, causing the calcaneus, and subsequently the entire foot, to adopt an equinus (horse-foot) posture. In this adducted and inverted position, the anterior portion of the calcaneus is lying directly beneath the head of the talus<sup>[16,18]</sup>. This displacement is responsible for the severe varus deformity or oblique displacement toward the midline of the heel<sup>[18,22]</sup>. The navicular is smaller than normal and articulates with the medial aspect of the neck of the talus, which forces the forefoot to adduct toward the midline. In addition, the cuboid is medially displaced and inverted in front of the calcaneus<sup>[16,18,22]</sup>. The cuneiforms are displaced downward and inward in front of the navicular. The medial displacements of the navicular, cuboid, cuneiforms, and metatarsals contribute in varying degrees to the severe adduction of the club foot. The varus deformity of the calcaneus and the adducted mid-tarsometatarsal bones together are the cause of the foot inversion<sup>[18,22]</sup>. Generalized hypoplasia of the major foot bones causes the affected foot to be smaller than normal<sup>[16,19]</sup>. Commonly, the first

metatarsal ray is short<sup>[12]</sup>. Hypoplasia of the calf muscles is another well-recognized anatomical abnormality of CTEV<sup>[19]</sup>. In addition, the abnormal positioning of the foot can create a deep medial and/or posterior foot crease on the plantar aspect of the foot<sup>[15,16,23]</sup>.

The severity of the club foot deformity may be classified according to the Pirani scoring system<sup>[16]</sup>. It includes six clinical features to describe the abnormality, with each being graded as 0, 0.5, or 1 depending on its severity. The scores are summed to give a score between zero and six with six being the most severe<sup>[17]</sup>. Clinical characteristics of children with CTEV include a hypotrophic anterior tibial artery and atrophy of the musculature around the calf<sup>[14]</sup>. In many patients, the dorsalis pedis artery is also affected or even absent<sup>[24]</sup>.

There are three classifications of CTEV: Postural, idiopathic, and teratogenic or syndromic. Postural foot is a molding abnormality that is resolved with simple maternal manipulation shortly after birth. Idiopathic CTEV represents the majority of cases and has no known cause. Teratogenic or syndromic CTEV is associated with other diagnoses, such as arthrogryposis, spina bifida, and/or chromosomal anomalies<sup>[15,16]</sup>.

The cause of CTEV is unknown<sup>[16,19]</sup>. There is a clear multifactorial inheritance pattern with many potential environmental influences<sup>[16]</sup>. Possible causes of CTEV include an arrest during fetal development, a primary germ plasm defect in the talus causing continued plantar flexion and inversion of this bone with subsequent soft tissue changes in the joints and musculotendinous complexes, or an enteroviral infection affecting anterior horn cells resulting in club foot. Other potential causes include attempted abortion causing amniotic injury or hemorrhage which affect bone development, positioning *in utero*, or primary soft tissue abnormalities within the neuromuscular units causing secondary bony changes<sup>[14]</sup>. Retracting fibrosis has also been identified as a primary etiological factor of the club foot deformity<sup>[18]</sup>.

Although each of the aforementioned syndromes is relatively rare, comorbidity of all three of these disorders is even rarer, if not unique ( $1.7\text{--}5.1 \times 10^{12}$ ). In this individual, all three of these conditions occurred simultaneously. One other study (Hootnick *et al.*<sup>[6]</sup>) with similar presentation was found in our literature search. Both similarities and differences were observed and noted.

Similar to the findings of Hootnick *et al.*<sup>[6]</sup>, we found the limb displayed hip dysplasia with equal femur lengths. The femur displayed no visible condyles or epiphyses on the radiographs. The tibia was absent (TH), and the fibula was complete, but bowed, shortened, and thickened. The navicular was small and deficient. There were three tarsal bones identifiable, but no cuboid or cuneiforms were visible on the radiographs.

Our findings differ from those of Hootnick *et al.*<sup>[6]</sup> in that our case involved a seven-month old female;

theirs, a 15-mo old male. In our case, the left limb was affected; in theirs, the right limb was affected. In our case, there was proximal articulation of the fibula and femur; in theirs, there was no evident proximal articulation. In our case, there was no tendinous band in place of the tibia; in theirs, a tendinous band connected the medial head of the fibula to the medial side of the lateral malleolus. In our case, there were seven metatarsals; in theirs, there were six. In our case, the metatarsals and phalanges two and three were normal, but four and five were abnormal; in theirs, metatarsals and phalanges were normal for digits three through five, with a normal second metatarsal with bifid proximal phalanx. In our case, there were seven digits with a hallux; in theirs, no hallux was identifiable. In our case, there were two supernumerary toes: Toes A and B; in theirs, three supernumerary toes: Toes A, B, and C, with partial syndactyly between toes two, A, and B. In our case, toe A was normal, but toe B was missing the middle and distal phalanges; in theirs, the second metatarsal was normal, except a medial bony projection.

Thus, this child seems to have partial mirror foot polydactyly consisting of only mirrored copies of the second and third toes, but not the hallux. This child exhibited preaxial polydactyly and CTEV as a result of having TH. As a result of the absence of the tibia, the foot became severely inverted and adducted as it articulated with the fibula. Since the patient's parents had no identifiable limb or other malformations, it is most likely that this disorder may have been the result of a rare autosomal recessive gene<sup>[12]</sup>, *de novo* dominant mutation, or localized vascular malformation during limb formation. A possible teratogenic event may have occurred in the affected leg at a time when limb developmental specifications had proceeded to the lower leg and foot<sup>[5]</sup>.

In conclusion, this case report analyzes the anatomical findings associated with systematic post-surgical examination of the foot of a seven-month old female presenting with a very rare combination of lower limb malformations. These observations include superficial dissection, X-rays and CT scans. Hootnick *et al.*<sup>[6]</sup> also described a similar case in the right lower limb of a 15-mo old male with comparable rare findings to our case, but with some notable and noteworthy differences. Due to the rarity of this combination of anatomical findings, descriptions of such cases are very infrequent in the literature.

## ACKNOWLEDGMENTS

The authors would like to thank the family for this generous donation.

## COMMENTS

### Case characteristics

A seven-month-old female presented with tibial hemimelia (TH), seven-toed

preaxial polydactyly, and severe left congenital talipes equinovarus (CTEV).

### Clinical diagnosis

Medial contracture of the left foot and lower leg with left congenital hip dysplasia, the absence of the left tibia, patella, active quadriceps mechanism and posterior tibial artery, and left fibular bowing with articulation to the lateral side of the left femur.

### Differential diagnosis

Interuterine toxic exposure to an agent, such as thalidomide; no evidence of such exposure, amniotic band stricture syndrome, rare autosomal recessive genetic mutation, *de novo* dominant, or localized vascular malformation during limb formation.

### Laboratory diagnosis

All labs are within normal limits.

### Imaging diagnosis

X-rays and computed tomography scans showed curved and overlapping preaxial supernumerary toes, hypoplastic first metatarsal, lack of middle and distal phalanges in one supernumerary toe, three tarsal bones, hypoplastic middle phalanx and no distal phalanx for fourth toe, no middle or distal phalanges for fifth toe, and the fibula articulated with the anteromedial calcaneus with an absent tibia.

### Pathological diagnosis

TH with preaxial polydactyly and CTEV of the left lower limb.

### Treatment

Due to the severity of the deformity, and the unlikelihood of a successful limb salvage procedure, amputation was advised and performed.

### Related reports

Hootnick's article (1983) also described a similar case in the right lower limb of a 15-mo old male with comparable rare findings to our case, but with some notable and noteworthy differences.

### Term explanation

TH or congenital tibial aplasia is a rare congenital anomaly characterized by variable degrees of deficiency of the tibia with a relatively intact fibula. Preaxial polydactyly is a congenital physical anomaly involving superfluous digits found on the lateral aspect of the affected upper limb or medial aspect of the affected lower limb. Club foot or CTEV is a complex, fixed deformity of the lower limb that is always present at birth and can be unilateral or bilateral.

### Experiences and lessons

Although each of the aforementioned syndromes is relatively rare, comorbidity of all three disorders is even rarer, if not unique ( $1.7-5.1 \times 10^{12}$ ).

### Peer-review

The authors describe a rare and very interesting case of a longitudinal deficiency of the lower extremity including a clubfoot-like deformity and polydactyly. The manuscript is well written and easy to understand, despite the complexity of the topic. The discussion is comprehensive, which enables the reader to understand the three components of the deformity. This case adds valuable information to the current literature that might help in decision making and counseling in similar cases.

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