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Tibial hemimelia with partially duplicated femur: a case report

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Tibial hemimelia is a rare congenital entity with a wide range of clinical manifestations affecting one or both lower extremities. The purpose of description of this case is to contribute to clinical insights into the variations associated with this rare entity. We present a case of a male newborn with tibial hemimelia who was referred to our clinic for further evaluation and treatment. According to our knowledge, this is the first description of tibial hemimelia with femoral duplication in medical literature. Multidisciplinary approach and thorough examination of patients with tibial hemimelia are required to understand full extent of deficiency and accompanying anomalies, and to ensure correct classification in order to provide the best treatment plan for each patient.

Key words: TIBIAL HEMIMELIA; FEMORAL DUPLICATION; CLUBFOOT; CONGENITAL LIMB DEFORMITY

INTRODUCTION

Tibial hemimelia is a rare congenital lower limb anomaly with an incidence of 1 in 1 million live births (1). The condition is defined as a spectrum of deformity characterized by deficiency of tibia and relatively unaffected fibula. The extent of pathology varies from mild hypoplasia of the tibia to its complete absence. In cases of complete tibial aplasia, flexion contracture of the knee and equinovarus deformity of the foot can be observed limiting knee and ankle range of motion (3). Most of the cases reported in literature were unilateral and 72% of them were affecting the right side (4, 5). It may present as an isolated disorder or as a component of a syndrome, such as Werner syndrome (polydactyly-triphalangeal thumb syndrome), Gollop-Wolfgang complex, tibial hemimelia diplopodia, tibial hemimelia-split hand/foot syndrome and tibial hemimelia-micromelia-trigonal brachycephaly syndrome (6-8). Prenatal ultrasound can be useful in diagnosis of tibial hemimelia as early as 16 weeks' gestation (9). Exact etiology of tibial hemimelia is still unclear, but both autosomal dominant and recessive inheritance have been reported for this malformation (10). Available classification systems take into account severity of deficiency and functional ability of the affected limb and while they are useful in indicating the severity of deficiency and difficulty of reconstruction, none of them can fit all types of tibial hemimelia, guide clinicians to optimal treatment modality or predict definite functional outcome of the affected limb. The case presented here does not meet classification criteria for any of the recognized syndromes, nor is there evidence of genetic inheritance. Informed consent was obtained from the patient's parents for publication of this case report.

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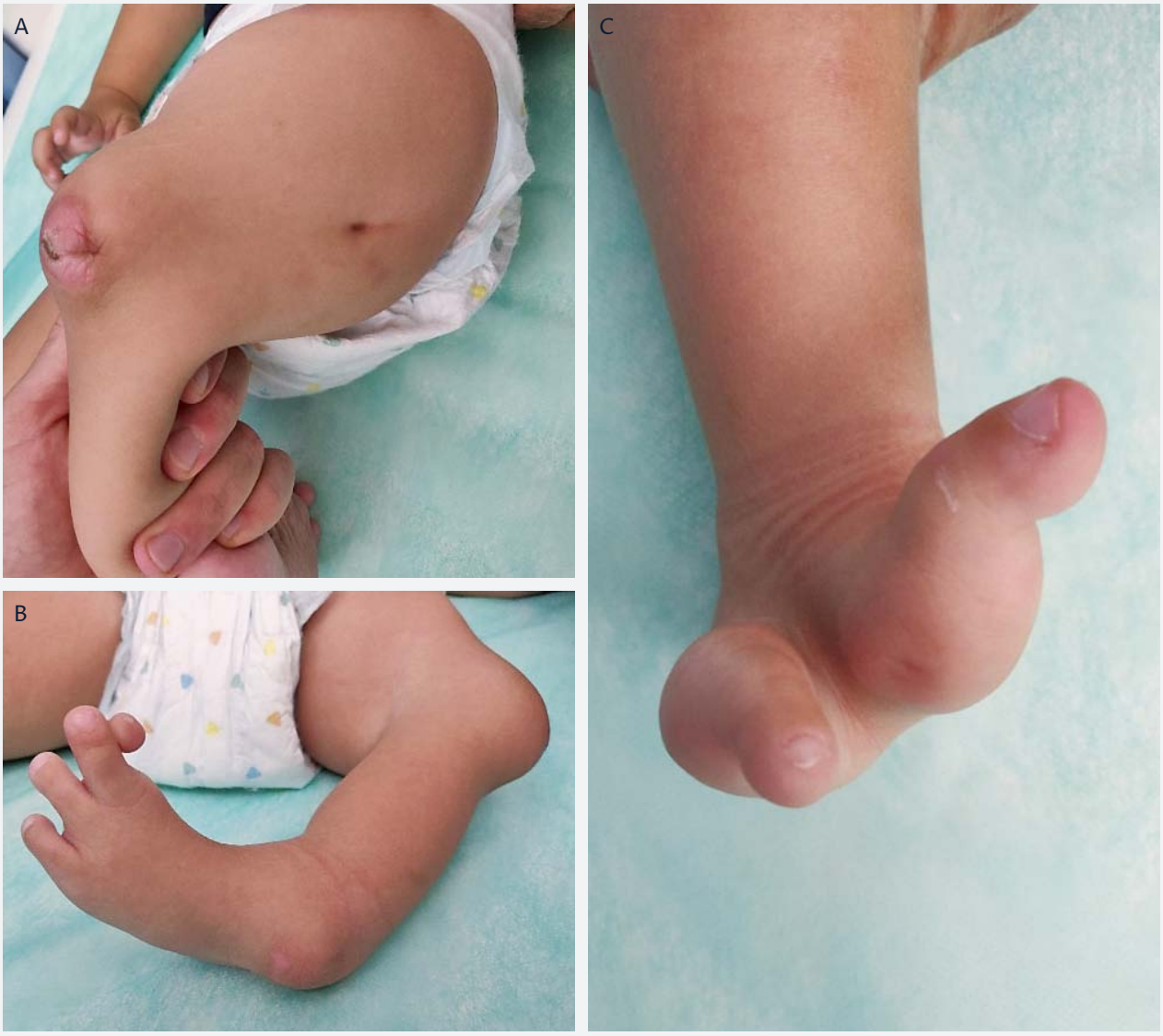

Figure 1A. Two skin dimples of the left leg: at the front of the knee and on the lateral side of the thigh
Figure 1B. Clubfoot deformity and oligodactyly on the left side
Figure 1C. Cleft foot anomaly on the right side

PATIENT PRESENTATION

We present the case of a 28-day-old boy, who was referred to our hospital for clinical and radiological evaluation and treatment of congenital deformities of both lower limbs and neural tube defect. The child was born at 38 weeks of gestation by spontaneous vaginal delivery (measures taken at birth: 3480 g, 49 cm; Apgar scores 10/10). He is the third child of a non-consanguineous couple; a 36-year-old father and 31-year-old mother. During the first trimester, mother used dydrogesterone because of threatened miscarriage. Other medications included paracetamol and diazepam during sixth month of pregnancy due to lower back pain. Mother had no history of diabetes, hypertension or using any teratogenic drugs. There was no history of congenital anomalies in the family and early post-natal period was uneventful.

Clinical findings

Multiple deformities of the child's lower extremities were observed. The most obvious was hypoplasia of the left leg. The left thigh and lower leg were shorter and bowed compared to the right leg, and two skin dimples were present, one on the outer side of the left thigh and other at the anterior part of the left knee joint (Figure 1A). There was a clubfoot deformity of the left foot

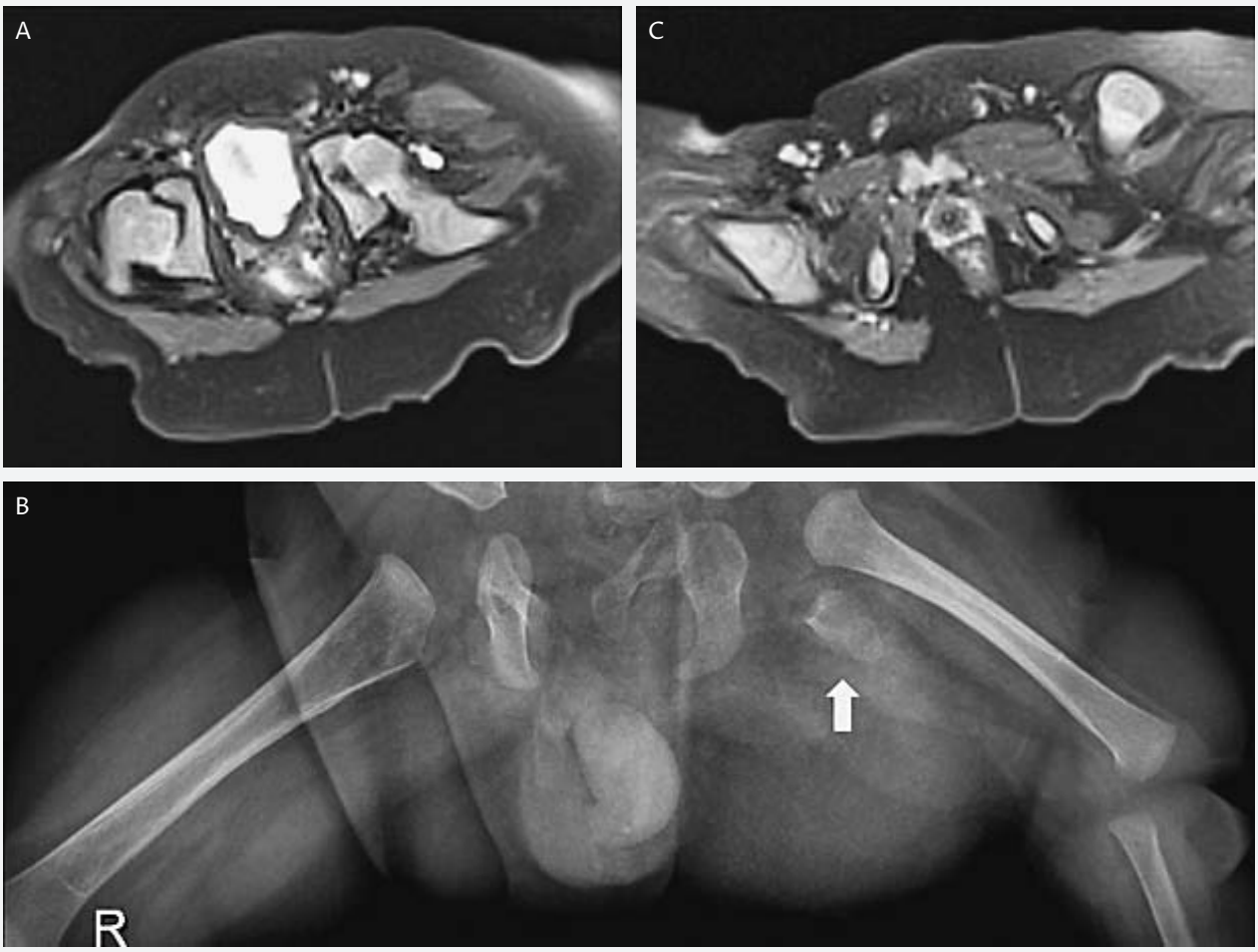

Figure 2A. MR of the hips when the child was 1 month old
Figure 2B. X ray image: partially duplicated femur on the left side
Figure 2C. MR of the hips when the child was 6 month old

and only three developed toes with a rudimentary one attached to the first toe (Figure 1B). Right thigh and lower leg clinically appeared normal. The right foot was cleft in the midline so the first and the fifth toe were separated (Figure 1C). There were no signs of venous stasis, arterial insufficiency or legs' edema. Clinical examination of the lumbosacral region revealed a single tail-shaped lump surrounded by two red spots. The lump had a vesicle on the top and measured 1x1 cm in size. There was no facial dysmorphism, and the upper extremities and the trunk were normal.

Radiological findings

A babygram obtained at first month of life as a part of multidisciplinary diagnostic procedure showed hypoplastic left femur, absent left tibia, bowed fibula and the left hip was highly suspicious of a subluxation. Magnetic resonance imaging (MRI) of the pelvis taken when the child was 1 month, showed some anomalies of the left hip anatomy which were uninterpreted (Figure 2A). The percutaneous overhead traction was applied for one week, followed by hip-spica immobilization for the next 4 weeks and hip abduction orthosis afterwards. During the hip treatment course, when the child was 6 months old, we observed that the left femoral head was dislocating again, which directed us to perform additional imaging. The pelvic and lower legs' radiographs then revealed the secondary ossification center in the left inguinal region, positioned medially to the hypoplastic left femoral head (Figure 2B). Radiographic findings of the right thigh were normal. Second MRI finally clearly revealed an accessory bone segment, located near rectus abdominis muscle origin, just below the sartorius muscle (Figure 2C). The bone segment was separated

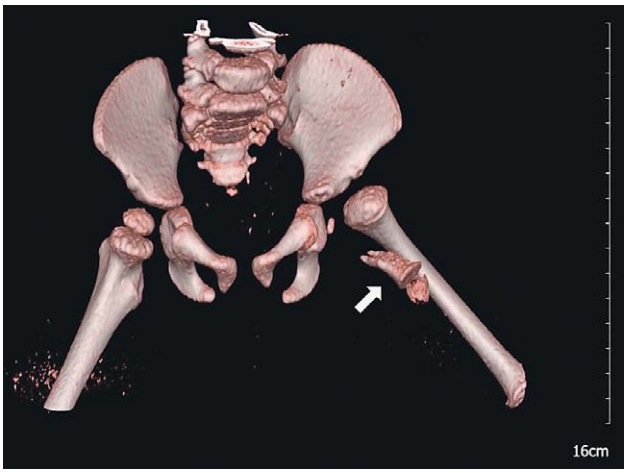

Figure 3. 3D CT of the hips and partially duplicated hypoplastic left femur

from the surrounding bony structures, measuring 21 x 16 mm in size. This rudimentary bone segment was also articulating with the acetabulum preventing the main but hypoplastic left femoral head from being fully positioned within the acetabulum thus causing the subluxation. Iliac and sacral bones of the pelvis on the left side showed delayed maturation and a shallow V-shaped acetabulum oriented ventrally. There was agenesis of gluteus medius muscle, hypoplastic vastus lateralis muscle, and oval shaped iliopsoas muscle on the left side. The rest of the left quadriceps femoris muscle and the left patella were missing. The 3D CT taken when the child was 16 months showed dysplastic left acetabulum and caudomedially to the dysplastic left hip the duplicated femur which was anteriorly oriented measuring 23 mm in length with an altered epiphyseal nucleus of 12 mm (Figure 3). For comparison, transverse diameter of proximal epiphyses of the left main femur and right femur were 6 mm and 13 mm respectively.

The right tibia was hypoplastic but developed in full length, unlike the left lower leg where the tibia was completely absent and the fibula was bowed. This is clearly seen on radiographs taken at the age of 21 months (Figure 4).

The ultrasound examination showed no abnormal findings of the brain, heart and abdominal organs. There were also no urogenital and the digestive system anomalies. Brain MRI revealed no hydrocephalus or any other anomalies. MRI of the neuroaxis revealed congenital dermal sinus which communicated with the spinal canal at the level of

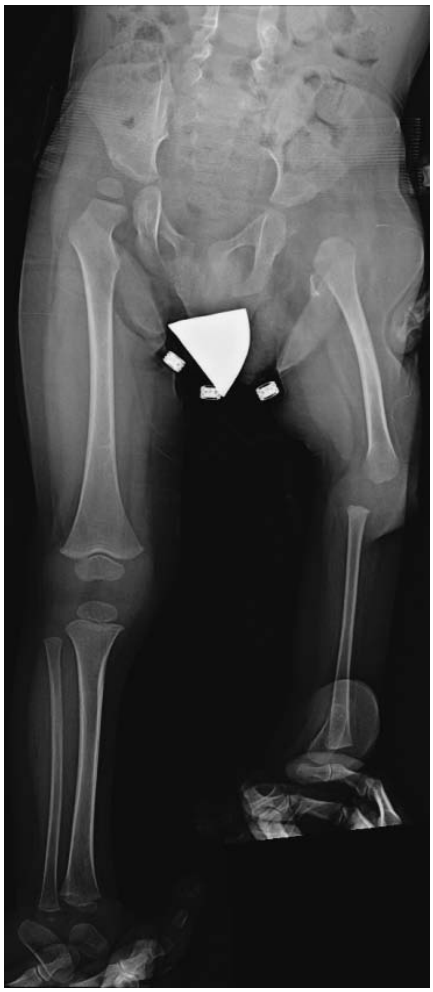

Figure 4. X ray image: anomalies of the lower extremities

L4-L5 and presence of the spina bifida at L3 level extending caudally. There was evidence of tethered cord syndrome with conus medullaris ending at L3 level. The tethered spinal cord was released, and resection of the dermal sinus tract was performed. Chromosomal microarray (CMA) testing was performed, but did not reveal any findings of clinical significance. Complexity of the anomalies induced us to find a solution in highly specialized centers of which only one accepted the challenge.

Two-stage surgery was performed addressing the left lower extremity deformities first. Removal of accessory femur along with hip stabilization procedure and through-knee amputation were performed to centralize the femoral head within acetabulum and allow for proper hip joint articulation. Closure of the left cleft foot was performed in the same surgery. The second stage included right hip stabilization as well as right foot reconstruction procedures. Following sur-

gery, the child underwent prosthetic limb fitting and rehabilitation and is able to ambulate independently.

DISCUSSION

Classification of tibial hemimelia often proves difficult as the condition is usually associated with a wide variety of skeletal and extraskeletal anomalies. Several classification systems for tibial hemimelia are proposed in literature to guide the treatment of such patients. Most commonly used classification system in literature for tibial hemimelia is Jones classification, which describes 4 types of tibial aplasia and dysplasia ranging from most deficient to least deficient (11). Jones classification of tibial hemimelia, which was later modified by *Kalamchi and Dawe*, was based on plain radiography findings (12). Modern diagnostic imaging techniques, such as MRI and ultrasonography, lead to a change in classification systems and treatment approaches. Newer classification systems took into account the whole lower extremity in order to define the full extent of the malformation and limb impairment. Weber introduced a classification system that emphasizes the importance of cartilaginous anlage in planning operative procedures, while also assigning a score to describe functional ability of the limb (13). Latest classification system by Paley also considers presence or absence of associated duplications and deficiencies adding a plus (+) or minus (–) modifier to each type. Associated anomalies reported in literature include hip dysplasia and hip dislocation, cleft palate, lobster claw deformity, hand syndactyly, polydactyly, bifurcated femur, club foot, spinal anomalies and other (9-14, 10-15). Clinical applicability of classification system depends not only on ease of use and extensiveness, especially in tibial hemimelia characterized by a wide spectrum of pathological findings, but also on its ability to guide treatment and define prognosis. Therefore, we believe that a classification system used should primarily aim to provide patient assessment and treatment guidelines and allow for a comparison of treatment outcomes. Our patient presented with tibial hemimelia along with partially duplicated femur on the left side, as well as spinal dysraphism. Although neural tube defects are known to be associated with tibial hemimelia, to our knowledge, this is the first reported case of tibial hemimelia associated with partially duplicated proximal femur. The deformity presented as a complete tibial aplasia on the left side and was classified as Paley type 5C, which was confirmed by absence of patella and quadriceps muscle on MRI (11-16), while on the right side it manifested as a cleft foot. Recommended surgical management depends mainly on severity of deformity and usually includes reconstructive options and amputation followed by the use of a suitable prosthetic fitting, with knee disarticulation being the gold standard in case of total absence of the tibia. Current surgical techniques allow for reconstructive options with satisfactory results even in patients with more severe types of deformity. Although amputations are less demanding and more reliable procedures with good functional and patient-reported outcomes that can be performed by most surgeons, limb-salvage techniques should be considered in patients who refuse amputation, who have no access to suitable prosthetic care and in those who have bilateral involvement with higher grade of limb deficiency, especially in those with active quadriceps function and present patella.

CONCLUSION

We report this case because of its complexity and rarity. It highlights the importance of thorough and recurring clinical and diagnostic examinations in patients with tibial hemimelia due to a broad spectrum of possible accompanying anomalies. It also emphasizes the value of modern imaging modalities such as MRI in defining the severity of deficiency and presence of accompanying anomalies.

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SAŽETAK

Tibijalna hemimelija udružena sa djelomično udvostručenim femurom: prikaz slučaja

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Tibijalna hemimelija je rijetki kongenitalni poremećaj sa raznolikom kliničkom slikom te može zahvaćati jedan ili oba donja ekstremiteta. Cilj ovog istraživanja je doprinijeti spoznajama o mogućim kliničkim slikama ovog rijetkog stanja. Prikazujemo slučaj muškog novorođenčeta sa tibijalnom hemimelijom koji je bio upućen u našu kliniku radi daljnje procjene i liječenja. Prema našim spoznajama, ovo je prvi opis slučaja tibijalne hemimelije sa udvostručenjem femura u medicinskoj literaturi. Za cjelovitu procjenu pacijenata sa tibijalnom hemimelijom potreban je multidisciplinarni pristup te temeljita i opetovana dijagnostika, a sve u svrhu točne i detaljne klasifikacije poremećaja kako bi se omogućilo pravodobno i adekvatno liječenje.

Ključne riječi: TIBIJALNA HEMIMELIJA; UDVOSTRUČENJE FEMURA; UVRNUTO STOPALO; UROĐENE ABNORMALNOSTI EKSTREMITETA