

POSTAXIAL HYPOPLASIA OF THE LOWER EXTREMITY IN CHILDREN – CASE REPORT

T. Marcovici¹, I. Sabau¹, I. Simedrea¹, P. Tepeneu¹, O. Marginean¹, C. Daescu¹, L. Tunea¹, D.Chiru¹, S.Olariu², M.Puiu¹

¹“Victor Babes” University of Medicine and Pharmacy, Timisoara, Romania

²“Louis Turcanu” Children’s Emergency Hospital, Timisoara, Romania

Abstract

Background: Postaxial hypoplasia of the lower extremity is a rare, congenital disorder characterized by partial or total absence of the fibula. It occurs in about 7 to 20 per million living birth. Males are twice affected as females. It has variable expression, ranging from mild to severe deformity and associated anomalies of the foot. **Material and methods:** We present a 14 months old male toddler, product of a non-consanguineous marriage, born at term, after an uncomplicated pregnancy. The patient presents mild shortening of the left lower extremity and foot deformity: syndactyly of the first and second toe and absence of the fifth toe. Evaluation was made by history data, clinical and genetic examination, laboratory and imaging studies. **Results:** No familial incidence of malformations, congenital infections or teratogenic factors were noticed. Completely absent left fibula and associated anomalies: tibiotalar valgus, tarsal coalition with 3-ray foot aspect; limb-length discrepancy of 2,5 cm were diagnosed. Observation and nonoperative management are appropriate in this case. Special shoes are prescribed to enable the child to gain maximal function. **Conclusions:** The diagnosis was made by the absence of the left fibula with associated skeletal anomalies. The functional, social and psychological state of the child will be considered. The patient must be monitored throughout his growth.

Key words: postaxial hypoplasia, lower extremity, rare disorder, child

Introduction

Postaxial hypoplasia of the lower extremity (fibular hemimelia) is rare and has variable expression, ranging from mild to severe deformity. It is a congenital disorder characterized by partial or total absence of the fibula. Agenesis of the fibula is the most common manifestation of a spectrum of dysplasias of the limbs, a reduction malformation affecting the long bones. Congenital absence of the fibula was first described by Goller in 1698. O’Rahilly and Frantz’s classification would be used to describe true congenital absence of the fibula and corresponding portion of the foot terminal as complete paraxial fibular hemimelia.¹ A constellation of lower-extremity features accompanies fibular hemimelia. Shortening of the limb is the most common sign and the

anomaly is mainly unilateral.¹ Generally, reported cases involve normal pregnancy with no familial incidence.¹ Theories of origin correspond to the sixth and seventh week of embryological formation with genetic versus teratogenic factors.¹ In type II deformity fibula is completely or almost completely absent. An affected extremity typically displays a valgus foot and ankle, shortening of the leg, anterior bowing of the tibia and knee, tarsal coalition and radiographic absence of one to several lateral rays.² Males are twice affected as females. It is estimated to occur in about 7 to 20 million living births.

Case report

We present an 14 months old male toddler admitted in the First Pediatric Clinic of “Louis Turcanu” Children’s Emergency Hospital for recurrent wheezing. He is second in birth order, product of a non-consanguineous marriage. The child was born at term, delivered vaginally, after an uncomplicated, normal pregnancy. No familial incidence of fibular hemimelia, no congenital infections were present. Mother wasn’t exposed to teratogenic factors. At birth he was noticed to have a left foot deformity (syndactyly of the first and second toe, an absent fifth toe) associated with a discreet shortening of the left limb.

Clinical findings: A leg-length discrepancy of 2,5 cm was associated with calcaneovalgus deformity in the left foot and skeletal anomalies: syndactyly of the first and second toe, the absence of the fifth toe; genu valgus with lateral axis displacement; a skin dimple in the left midtibial area. (Fig.1)

X-ray series (long-leg standing; pelvis/hip; tibia/fibula; ankle/foot) were done. The long-leg series noticed left fibula agenesis. (Fig.2) The ankle/foot series noticed: tibiotalar valgus and tarsal coalition; a 3-ray foot; metatarsals associated with three phalanges, two of which are fused to form only three toes; limb-length discrepancy. (Fig.3) Ultrasonography examinations (transfontanelar/cardiac/abdominal) noticed normal aspects. Laboratory studies pointed out normal data. Orthopedic, genetic, ophthalmologic, cardiology and neurologic evaluation was done and no other abnormalities were noticed.



Figure 1. Clinical aspect of the left lower limb.



Figure 2. Long-leg standing X-ray aspect.



Figure 3. Ankle-foot X-ray series.

By the Achterman and Kalamchi classification system, is a type **II** of fibular hemimelia with completely absent fibula in this case.³ Fibula hemimelia is associated with deficiencies of the lateral aspect of the foot, calcaneovalgus deformity and associated skeletal anomalies: syndactyly of the first and second toe, the absence of the fifth toe, tarsal coalition.

By Stanitski D.F. and Stanitski C.L. system, the patient is classified as having a type **IIISc3** fibular hemimelia, where **III** is the complete absence of the fibula;

S is the spherical, ball-and-socket ankle; **c** is the presence of the tarsal coalition and **3** is the 3-ray foot.³

We considered to be a sporadic incidence of postaxial hypoplasia of the lower extremity in this case.

Therapy: Because the deformities are mild, observation and non-operative management is appropriate in this case. Special shoes are prescribed to enable the child to gain maximal function. Because the discrepancy between lower extremities may progress with growth, in the future procedures in the left foot may include resection of talar coalitions and fusions.

Follow-up: This patient must be monitored throughout his growth. The clinician must inform the family of what they might confront with. The limb-length discrepancy is one of the most difficult to address.³

Outcome and prognosis: Genu valgum associated with postaxial hypoplasia of the left lower extremity is progressive and it can adversely affect alignment of the lower limb in this case. Special shoes will facilitate an increase in weight bearing activity with minimal discomfort.⁴ The patient and his parents are at risk of emotional problems due to limb's cosmetic troubles.³

Discussions

Presentations of postaxial hypoplasia of the lower extremity (fibular hemimelia) vary widely, ranging from

what appears to be an absent fifth toe in a newborn or a minimal difference in limb lengths, to severe fibular deformities that are immediately apparent. The ipsilateral tibia may be hypoplastic, bowed or normal.^{5,6} Postaxial hypoplasia of the lower extremity can be frequently associated with femoral deficiency, deformities of the lateral aspect of the foot, or is part of a malformation syndrome.^{3,6} The most associated anomalies are skeletal and includes: syndactyly, brachydactyly, clinodactyly of fingers and toes and facial dysmorphism. The pediatrician must look for associated abnormalities, including problems with alignment and stability, because the clinical appearance may evolve with growth and development.³

Table I. Clinical findings of postaxial hypoplasia of the lower extremity.³

<ul style="list-style-type: none"> ▪ Fibular anomaly (shortening/complete absence) ▪ Femoral hypoplasia ▪ Lateral patellar subluxation ▪ Genu valgus with lateral mechanical axis displacement 	<ul style="list-style-type: none"> ▪ Coxa vara ▪ Tibial deformities (shortening/bowing) ▪ Ankle anomalies (valgus/ball-and-socket aspect) ▪ Foot deformities (absent tarsal bones/tarsal coalition/absent foot rays)
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Postaxial hypoplasia of the lower extremity is usually sporadic, with a negligible recurrence risk for the patient's siblings.^{7,8} In a small percentage of cases, a familial incidence (autosomal recessive) has been reported.^{7,8}

In type II deformity patients have unilateral absence of the fibula, anterior bowing of the tibia, foot deformity with absent rays and marked shortening of the leg.^{9,10,11} These aspects are observed in about 35% of the cases.

Prognosis and rehabilitation are mainly dependent of the limb malformation's severity and the possibility of orthopedic correction.¹²

Conclusions

Postaxial hypoplasia of the left lower extremity was diagnosed by the absence of the fibula with associated skeletal anomalies in the foot. The functional, social and psychological status of the patient will be considered. The ultimate goal of the treatment is to enable the child to gain maximal function by achieving adequate lower-extremity alignment, length and stability. The patient must be monitored throughout his growth.

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Correspondence to:

Marcovici Tamara
Iosif Nemoianu Street, No.2
Timisoara 300011
Romania
E-mail: t_marcovici@yahoo.com