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Concomitant rhomboid-shaped tibiae and fibulae, finger-like projections, and orthopedic management in a new variant of nievergelt syndrome: A case report

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ABSTRACT

INTRODUCTION: The rare Nievergelt syndrome (NS) is the most severe form of mesomelic dysplasia and is characterized by disproportionate shortness of the limbs. The aim of this case report was to describe the clinical and radiological features of a rare case of NS.
PRESENTATION OF CASE: Here we describe a female patient originally presenting with bilateral hand, lower leg, and foot deformities at the age of 10 years old. In addition to the characteristic features of NS, this patient presented with finger-like projections on her heels, bilateral hand anomalies, and atypical facial features. She underwent concomitant bilateral tibial lengthening and deformity correction using external fixators due to severe bilateral lower leg deformities with shortness. At 10 years of age, this patient was able to walk independently with significant improvement in her ambulation.
DISCUSSION: There is a clear gap in the literature regarding the orthopedic management of mesomelic limb deformities due to NS. No studies have been designed to illustrate surgical planning in the management of orthopedic deformities in this rare syndrome.
CONCLUSION: Limb lengthening and deformity correction using an external fixator can be considered as a salvage method or alternative to amputation for patients with severe mesomelic limb deformities due to NS.

1. Introduction

Mesomelic dysplasia consists of a constellation of hereditary congenital skeletal disorders characterized by disproportionate shortness of the limbs, predominantly involving the middle segments (mesomelia). This group of skeletal disorders presents with different patterns of inheritance, a broad variety of clinical manifestations (including skeletal and nonskeletal disorders), and specific radiological features [1–4]. With an autosomal dominant pattern of inheritance, Nievergelt syndrome (NS) is rare, but it is the most severe form of mesomelic dysplasia and was first described by Nievergelt in 1944. This disorder can be distinguished by distinctive triangular or rhomboid-shaped rudimentary bones of the lower legs and radioulnar dislocation [2,3,5]. A few NS case reports have been reported previously in the medical literature, but most of these were focused on the clinical and radiological characteristics of the disease [1,2,4–9]. However, the orthopedic management of the skeletal disorders relevant to NS remains controversial. For NS, the ideal surgical timing and the results of the deformity correction have not been clearly defined.

The purpose of this case report was to describe the clinical and radiological features of a rare case of NS, and to illustrate the surgical planning in the management of the orthopedic deformities of this rare syndrome. This patient and her family were informed that data concerning this case would be submitted for the publication. Both the patient and her family consented to this. This paper has been reported in line with the SCARE criteria [13].

2. Presentation of case

This female patient was the first child of healthy consanguineous parents (first cousins). She was born at 29 weeks of gestation via cesarean section due to the premature rupture of the fetal membranes. Her mother was gravida 1 para 1. The birth weight was 1580 g and her Apgar scores were 7 and 8 at one and five minutes,

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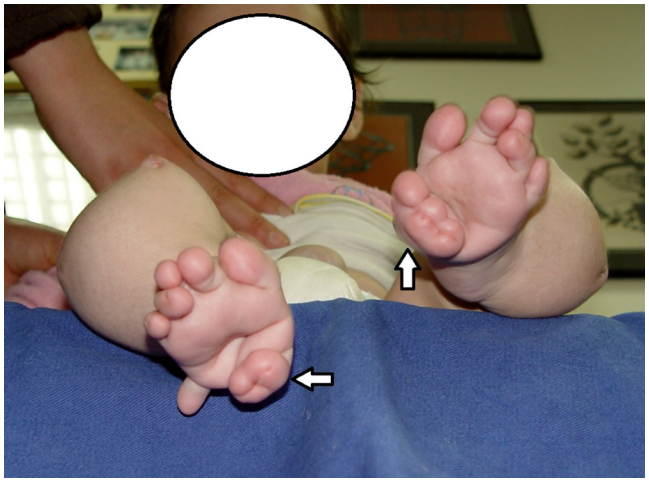

Fig. 1. Atypical facial features and finger-like projections on both heels (white arrows).

respectively. The patient was monitored in the neonatal intensive care unit for three weeks following delivery due to neonatal respiratory distress syndrome, hyperbilirubinemia, and congenital adrenal hypoplasia. Her abnormal lower limbs, with extreme shortness and thickness of the lower legs, and bilateral hand anomalies were recognized at birth, in addition to her atypical facial features, including low-set ears, short palpebral fissures, down-turned corners of the mouth, a long face, and retrognathia (Fig. 1). The karyotype was 46, XY. Her auditory brainstem responses were normal. The cranial magnetic resonance imaging, echocardiography, and abdominal ultrasonography showed no congenital anomalies. Moreover, there was no family history of skeletal disorders. Given the patient's characteristic skeletal deformities, a diagnosis of NS was established by genetic specialists at 3 months of age.

When she was 2 years old, this patient was referred to our department due to her remarkable skeletal deformities and an inability to walk. Prior to admission, a below-knee amputation was suggested by the previous orthopedic surgeons; however, the parents refused that option. In order to determine whether or not a reconstruction could be performed, a detailed investigation was initiated by the senior author.

The physical examination revealed remarkable upper and lower limb deformities. The upper limb deformities included bilateral symmetrical oligosyndactyly, fusiform-shaped fingers, abnormal palmar-phalangeal creases, and cone-shaped fingernails on both hands, as well as a common triangular-shaped middle phalanx of the 3rd and 4th fingers on her right hand (Fig. 2). In the examination of the lower limbs, both striking and debilitating deformities drew our attention, which consisted of extreme shortness and thickness in both lower legs and cutaneous skin dimples above the anterior and lateral sides of the proximal tibia (Fig. 3). Additionally, her feet were in severe and fixed equinus positions of approximately 60°; however, each foot was composed of 5 toes that displayed normal anatomy. Interestingly, there were four finger-like projections on the right heel and five on the left heel, which had no connections to the metatarsal bones (Fig. 4).

The range of motion of the knee was limited on both sides (R: 0–110°, L: 0–95°), and there was no passive motion in the ankle or subtalar joint, with severe deformity. Otherwise, the hip joint range of motion was within normal limits on either side. This child displayed normal neurological and mental development for her age.

Among the various radiographic findings of this syndrome, the most impressive was the bilateral rhomboid-shaped fibula, which was accompanied by a rhomboid-shaped tibia (Fig. 5). Further X-rays of the patient's right hand demonstrated that the third

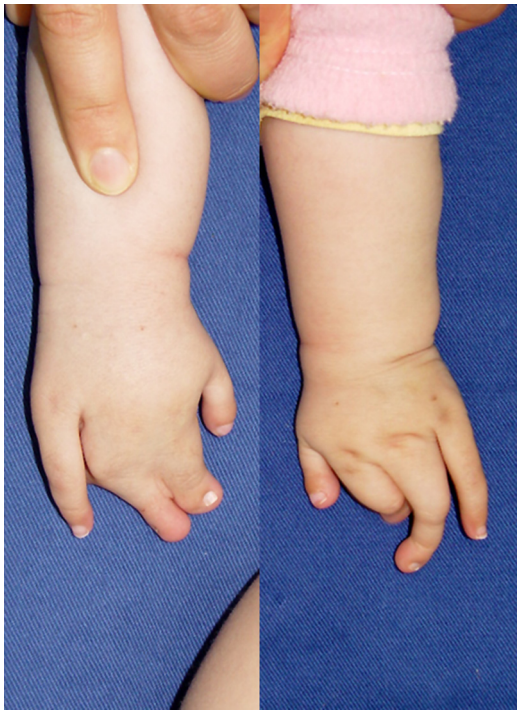

Fig. 2. Bilateral hand deformities including symmetrical oligosyndactyly, fusiform-shaped fingers, abnormal palmar-phalangeal creases, and cone-shaped fingernails.

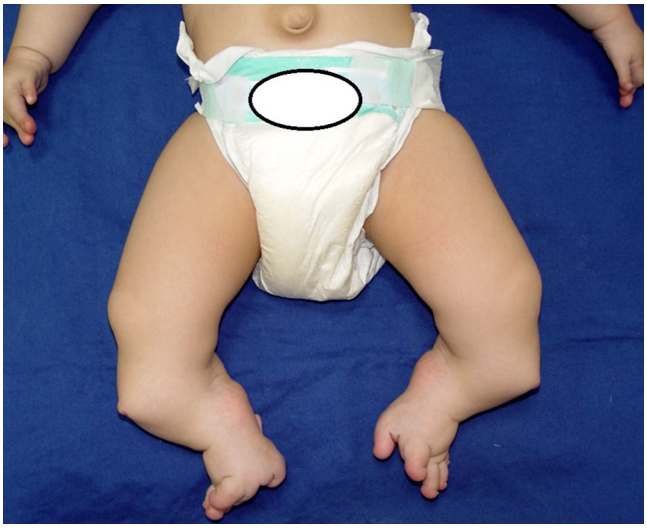

Fig. 3. Bilateral remarkable skeletal deformities consisting of short and thick legs with cutaneous skin dimples above the proximal tibiae.

finger, which consisted of the fusion of two fingers, had two normal metacarpals, two malformed proximal phalanges connecting to the same triangular-shaped middle phalanx, and two normal distal phalanges. Moreover, the radiographs of the feet displayed rhomboid-shaped, deformed talus and calcaneus bones in addition to a tarsal synostosis on both sides (Fig. 6). The femur, pelvis, and hip joint showed normal anatomy. There were no other abnormalities of the extremities or internal organs. In the laboratory evaluation, the complete blood count, biochemical findings of the blood, urine analysis, and renal and liver function tests were all normal.

At 2 years of age, while considering the promising ambulatory capacity of this patient, with the favorable anatomical and functional status of her hip and knee joints, the following strategy was pursued. At the time of admission, we preemptively decided to

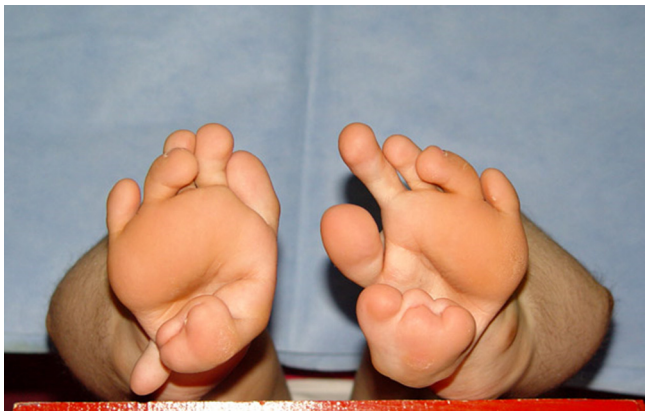

Fig. 4. Finger-like projections on both heels.

resect the bone spikes in the tibia due to the risk of skin ulcers. Then, the patient was followed up to monitor the natural process of her disease progression, and the variations in her ambulatory capacity over 4 years, thus obtaining a suitable skeletal maturity and muscle balance to employ the biological reconstruction and lengthening of both lower legs.

At 6 years of age, the similar skeletal deformities were more evident than at the time of admission. The hip joint range of motion was within normal limits on either side. However, the range of motion of the knee was not improved despite a meticulous physical rehabilitation program. The patient was able to walk on her knees without assistance and exhibited normal neurological and mental development for her age. Furthermore, a computed tomography (CT) angiography showed that the popliteal artery was divided into two branches above each knee (at the metaphysodiaphyseal portions of the distal femora), and that there were no fibular arteries leading to a poor vascular supply at the level of the lower legs (Fig. 7). And then, in a single-stage operation, we first reshaped the tibia and fibula to be more tubular in both lower legs, with a resection of the bone protuberances. Subsequently, bilateral tibial lengthening and deformity correction with a monofocal tibiofibular osteotomy using a circular external fixator was performed, in addition to a bilateral gradual closed correction of the equinus

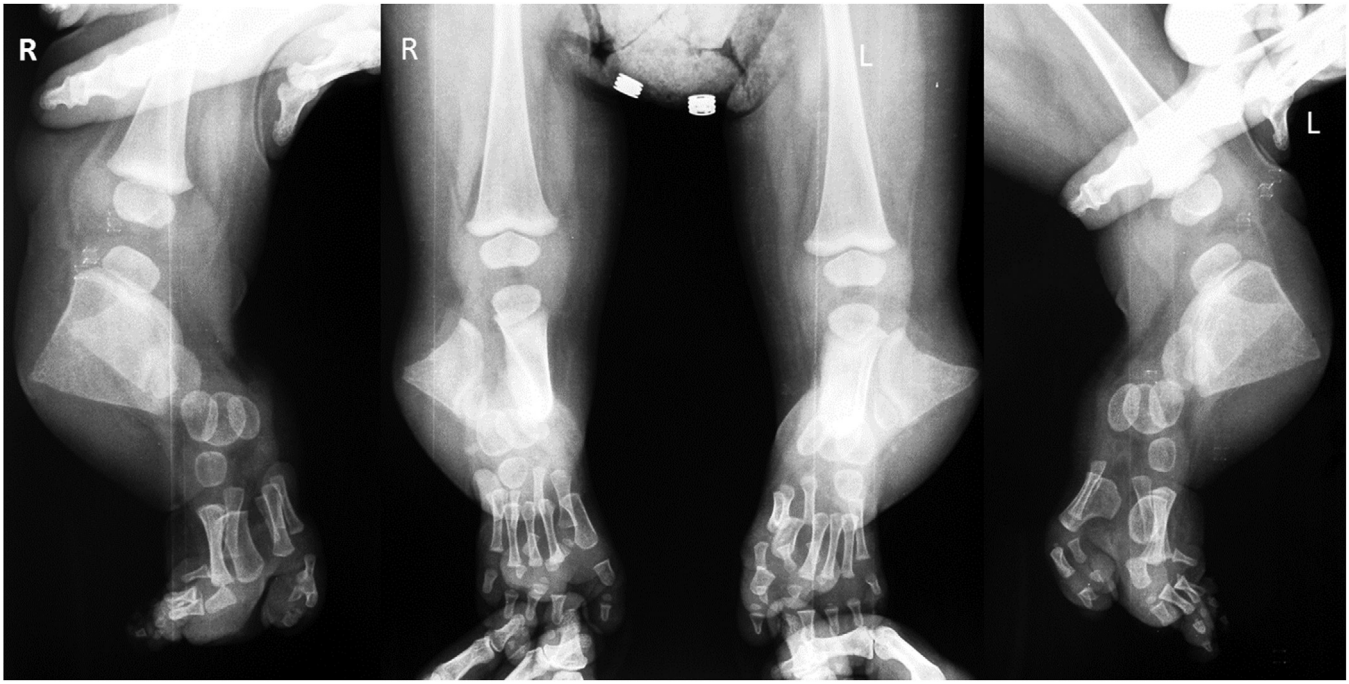

Fig. 5. Preoperative radiographs of lower limbs on admission to the hospital, showing bilateral rhomboid-shaped fibula accompanied by a rhomboid-shaped tibia.

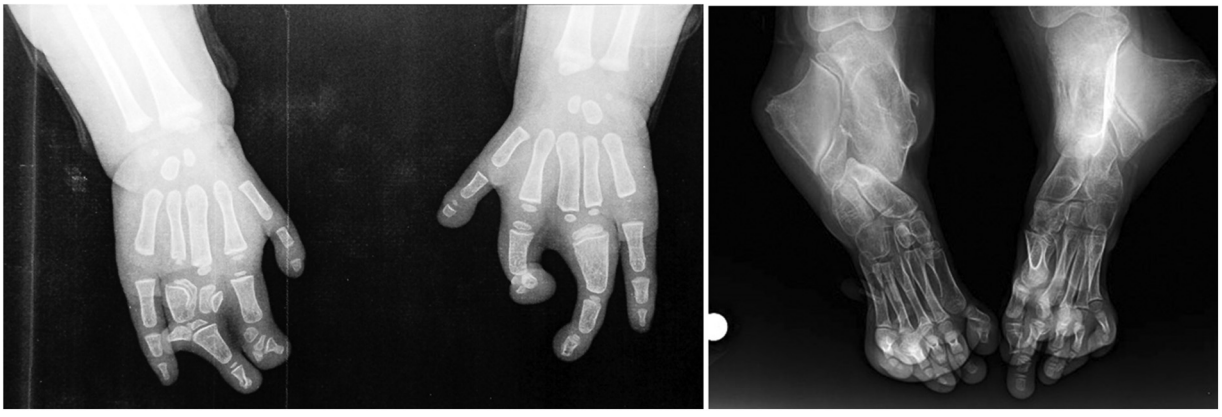

Fig. 6. Radiographs of the hands and feet.

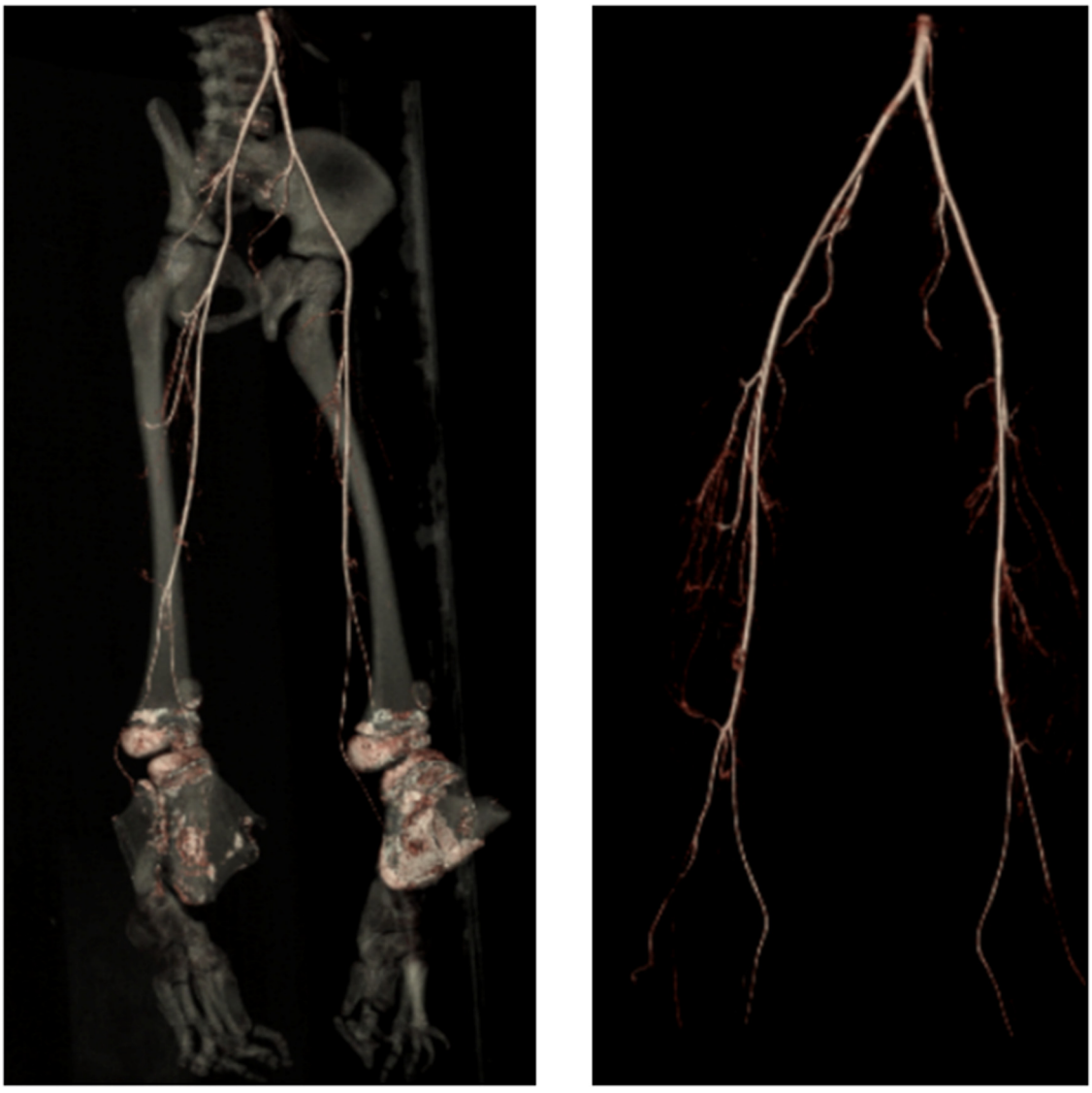

Fig. 7. CT angiography depicting a poor vascular supply at the level of the lower legs.

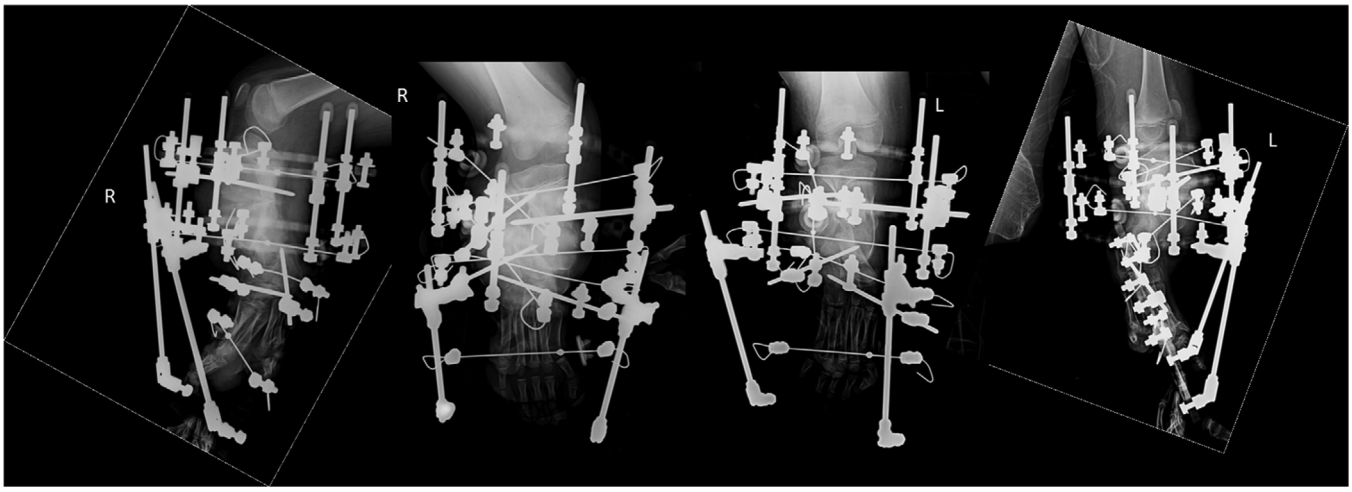

Fig. 8. At 6 years of age, early postoperative radiographs of the patient display bilateral tibial lengthening and deformity correction with a monofocal tibiofibular osteotomy using a circular external fixator.

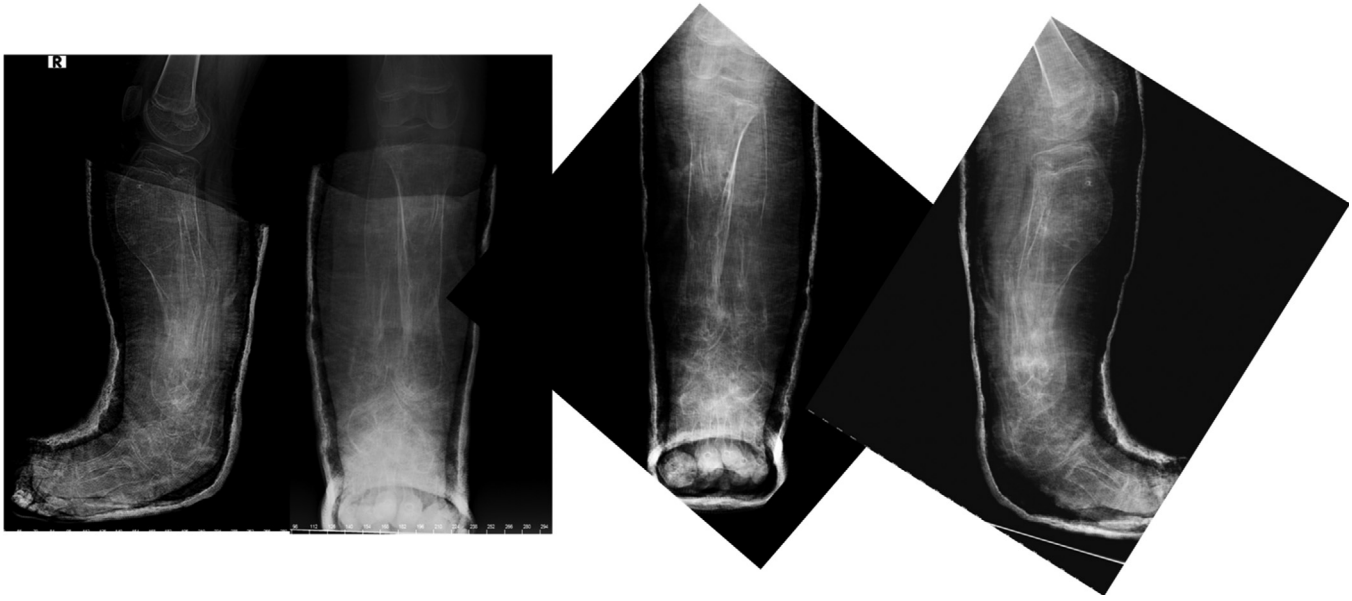

Fig. 9. The control radiographs after removing external fixators showing a total of 5 cm of lengthening and an acceptable deformity correction in both lower legs in addition to the equinus contractures to approximately 30° on each side.

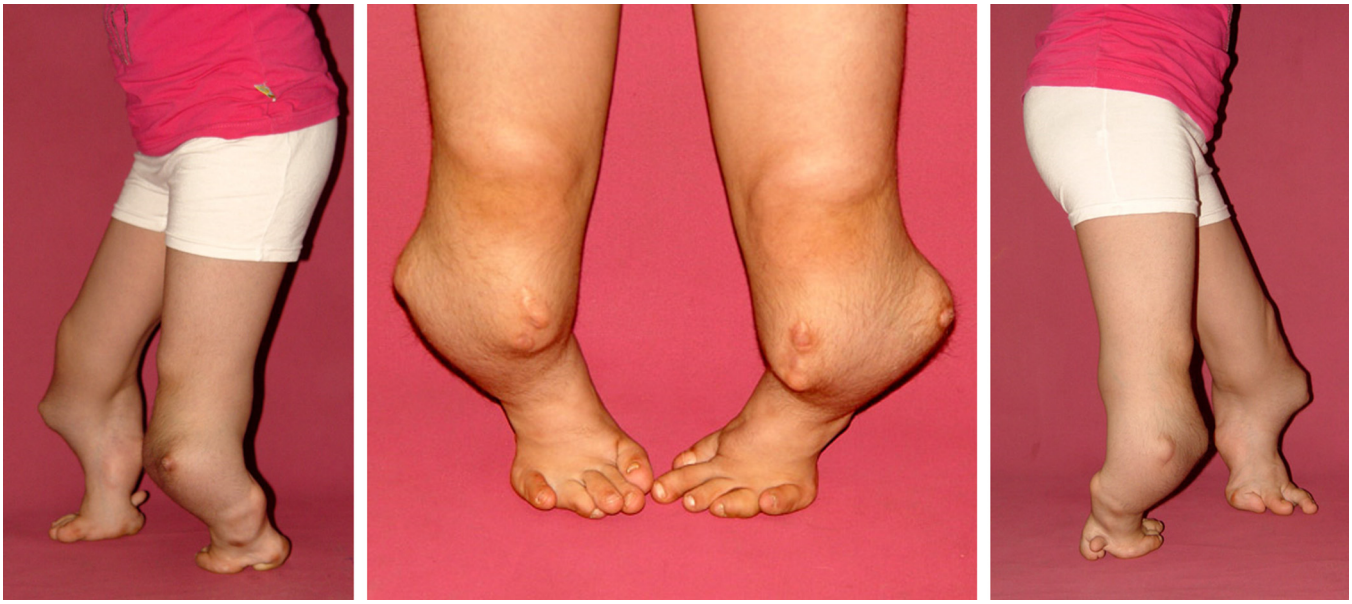

Fig. 10. Clinical photographs of the patients at the final follow-up visit.

contracture (Fig. 8). No intraoperative complications were encountered. Because of the poor bone quality, the parents conducted the lengthening by means of a one-quarter turn twice each day (0.5 mm/day) on a distractor inserted into the external fixator. Meanwhile, a bilateral gradual correction of the equinus contracture was initiated at a rate of 4 mm per day (1°/day), with medial and lateral hinges connecting the tibial ring. At the second month follow-up, due to the early fusion of the posterior cortex of the left tibia, we performed a second osteotomy. This was the only complication encountered during the treatment period. After surgery, the patient was followed up monthly for the first 6 months and every three months afterwards by plain radiography.

We obtained a total of 5 cm of lengthening and an acceptable deformity correction in both lower legs at the 4-month follow-up. Likewise, the treatment reduced the equinus contractures to approximately 30° on each side. The patient underwent no soft tissue or tendon lengthening procedure. After achieving complete fusion, the external fixators were removed 6 months after the operation (Fig. 9). At 10 years of age, this patient was able to walk independently with a significant improvement in her ambulatory status (Fig. 10). There were no deteriorations in her deformities.

3. Discussion

NS is mainly associated with mesomelic disproportion of the limbs. The primary phenotypic characteristics, first described by Nievergelt in 1944, include a rhomboid-shaped tibia and overgrown fibula accompanied by bony protuberances and skin

dimples. Other musculoskeletal findings documented in the literature include an equinovarus deformity of the foot, synostoses of the tarsal bones, metatarsi, foot phalanges, and radioulnar joint, and dislocation of the radial head [9,10]. Hess et al. discovered that NS is inherited with an autosomal dominant pattern [6]. However, sporadic cases without a family history of skeletal disorders have been reported in the literature [7,11], as seen in our case.

We have described the case of a now 10-year-old girl with impressive lower leg deformities and the characteristic features of NS. In our case, the concomitance of rhomboid-shaped fibulae and tibiae drew our attention. Previously, Vasil et al. presented an unusual form of NS comprised of hypoplastic, malformed lower leg bones, symmetrical upper limb anomalies, and noted triangular shaped-fibulae, which were more severely involved than the tibiae. The concomitance of rhomboid-shaped fibulae and tibiae distinguished our case from that case [8]. In addition, Tuysuz et al. described a case similar to ours, with short and triangular-shaped tibiae and fibulae, but no upper limb abnormalities except for the hands [5].

Regarding the involvement of the upper limb, with the exception of the hand anomalies documented in the Case Presentation section, no upper limb deformities were present in our case. Although rare when compared to the lower limb, upper limb deformities could be caused by this syndrome. Petrella et al. presented a case of NS with miscellaneous upper limb deformities that were manifested by dislocated radial heads, the proximal widening of the ulnae, and short forearms, as well as severe hand anomalies including post-axial hexa-syndactyly and hypoplastic thumbs [7].

To our knowledge, the distinctive features of this patient, including the finger-like projections on her heels and vascular status recognized via CT angiography, have yet to be reported in the literature. By means of these projections, the patient was able to walk, despite the slight equinus deformities of her feet. The vascular status of the lower extremity has not been investigated previously in NS. As far as we could determine, the present case is the first reported case of NS in whom the vascular status of the lower extremities has been documented.

According to our review, there is a clear gap in the literature regarding the orthopedic management of mesomelic limb deformities due to NS. However, none of these studies were designed to illustrate the surgical planning of the management of the orthopedic deformities of this rare syndrome [2,4–8,12]. The limb lengthening and deformity corrections using an external fixator yielded successful functional (ambulatory) and cosmetic outcomes in the orthopedic treatment of our case, despite her equinus contractures. The equinus deformities could be corrected completely with an additional hindfoot or ankle osteotomy. However, considering the poor vascular supply according to the CT angiography and her ability to walk with the finger-like projections on her heels, the senior author made a follow-up decision to forgo an additional surgical intervention. In our opinion, an early diagnosis, detailed investigation for the assessment of walking ability, and a well-planned surgical approach played crucial roles in correcting this patient's deformities and yielding a satisfactory ambulatory capacity.

4. Conclusion

An early diagnosis, detailed walking ability determination, and well-planned surgical approach are crucial for correcting deformities and increasing the ambulatory capacity of patients with NS. Limb lengthening and deformity correction using an external fixator can be considered as a salvage method and as an alternative to amputation. In addition, a preoperative evaluation of the vascular status could prevent several postoperative complications. Physicians should keep these finger-like projections and concomitant rhomboid-shaped tibiae and fibulae in mind as rare features of such a rare syndrome.

Conflict of interest

The presence or absence of the conflict of interest (COI) should be declared for the individual authors like "Tuna PEHLİVANOĞLU, Mehmet DEMİREL, Yavuz SAĞLAM, Halil İbrahim BALCI, Hayati DURMAZ" declare that they have no conflict of interest.

Sources of funding

None.

Ethical approval

Advocate Health Care Institutional Review Board does not require review for case reports.

Consent

Written informed consent was obtained from the patient's parents for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

Author contribution

(1) Tuna Pehlivanoğlu, MD: drafting of the article; (2) Mehmet Demirel, MD: equal contribution as lead author, drafting of the article, critical revision of the article for important intellectual clinical content; (3) Turgut Akgül, MD: revision of the article for important intellectual clinical content; (4) Yavuz Sağlam, MD: revision of the clinical and intellectual content of the article; (5) Hayati Durmaz, MD: intellectual content of the article.

Guarantor

Hayati DURMAZ, MD

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