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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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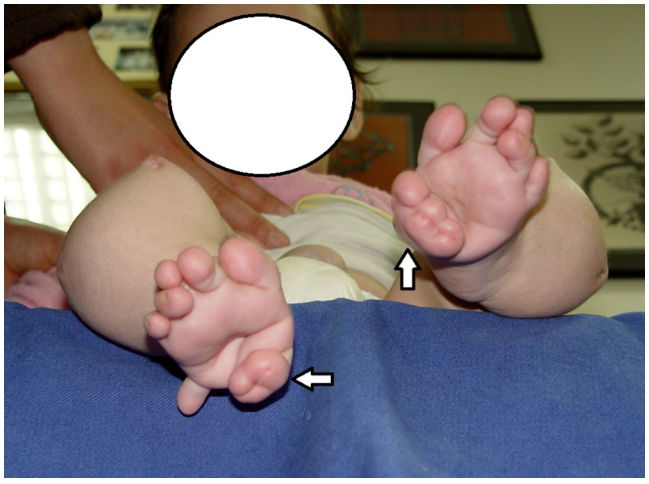

Fig. 1. Atypical facial features and finger-like projections on both heels (white arrows).

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

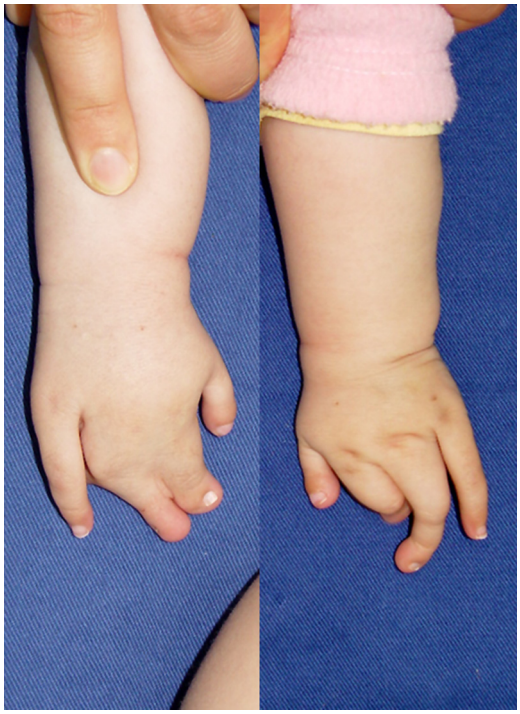

Fig. 2. Bilateral hand deformities including symmetrical oligosyndactyly, fusiform-shaped fingers, abnormal palmar-phalangeal creases, and cone-shaped fingernails.

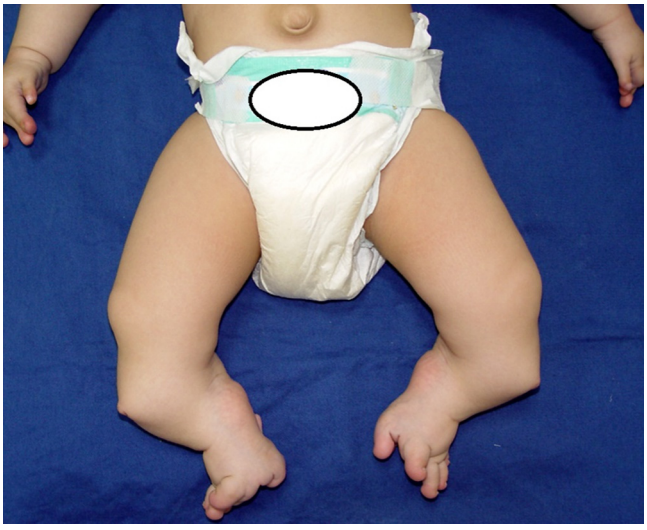

Fig. 3. Bilateral remarkable skeletal deformities consisting of short and thick legs with cutaneous skin dimples above the proximal tibiae.

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