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Case Report

Unrecognized Proximal Focal Femoral Deficiency Associated with Ectrodactyly. The Importance of Antenatal Skeletal Screening. A Case Report and Review

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Abstract

Antenatal care (ANC) aims to promote maternal health, detect fetal anomalies, and prepare families and clinicians for potential complications. Prenatal ultrasonography is important diagnostic tool to identifying skeletal abnormalities, but diagnostic accuracy varies according to operator skills, equipment quality, and organizational differences in screening programs.

We report a case of proximal focal femoral deficiency (PFFD) in a newborn whose prenatal scans were reported as normal. Postnatal examination revealed right limb shortening and left-hand ectrodactyly. Imaging confirmed PFFD, and systemic screening was normal. The case highlights the potential for missed diagnoses during ANC and underscores the need for standardized anomaly scans regardless of maternal risk classification.

Keywords: *Proximal focal femoral deficiency; skeletal dysplasia; prenatal diagnosis; antenatal care; limb length discrepancy.*

Introduction

Antenatal care (ANC) aims to promote maternal health, detect fetal anomalies, and prepare families and clinicians for potential complications.Antenatal care (ANC) encompasses counseling, optimization of maternal health, and structured screening for fetal anomalies. Prenatal ultrasonography is the cornerstone of anomaly detection, yet its diagnostic accuracy is influenced by operator expertise, image acquisition quality, and variability in national or institutional screening guidelines. Among detectable fetal disorders, skeletal dysplasias represent a vast and heterogeneous group, with more than 450 defined conditions affecting bone and cartilage development.

Prenatal identification of skeletal dysplasias relies on systematic sonographic assessment, including long bone measurements, evaluation of bone morphology and mineralization, thoracic size, and associated anomalies. Short long bones are particularly challenging, as they may reflect constitutional variation, chromosomal abnormalities, or underlying skeletal dysplasia. Operator dependency and heterogeneous screening protocols contribute substantially to variation in detection rates.

Proximal focal femoral deficiency (PFFD) is a rare congenital limb malformation resulting from disruption of early embryonic development of the proximal femur, acetabulum, and surrounding soft tissues between the fourth and eighth gestational weeks. The condition presents on a spectrum, from mild femoral shortening with preserved joint morphology to severe cases with near-complete femoral absence. Radiographically, PFFD is characterized by a short, tapered femoral shaft, delayed or absent femoral head ossification, coxa vara, and acetabular dysplasia. Multiple classification systems—Aitken, Gillespie, Bonafé, and Paley—aid in assessing severity, reconstructive feasibility, and long-term prognosis.

In addition to PFFD, congenital limb anomalies such as ectrodactyly may coexist. Ectrodactyly, or split-hand/split-foot malformation, is characterized by absence or hypoplasia of central rays, resulting in a median cleft of the hand or foot. It may occur as an isolated anomaly or within multisystem genetic syndromes. Embryologically, ectrodactyly arises from disruption of the apical ectodermal ridge (AER) during digital patterning between the fourth and seventh gestational weeks. Given its potential syndromic associations and variable functional impact, comprehensive evaluation is recommended.

The coexistence of PFFD and ectrodactyly, as in this case, raises consideration for broader limb malformation patterns, although systemic evaluation may remain normal. This report describes a newborn with right-sided PFFD and left-hand ectrodactyly not identified during routine prenatal care, highlighting limitations in anomaly detection and the need for standardized, expert-performed screening.

Case Presentation

A four-day-old girl was brought to the Orthopaedic Clinic at the Halibet National Referral Centre in Asmara, Eritrea, by her mother, who had noticed a discrepancy between her lower limbs and an abnormally shaped left hand.

Maternal History

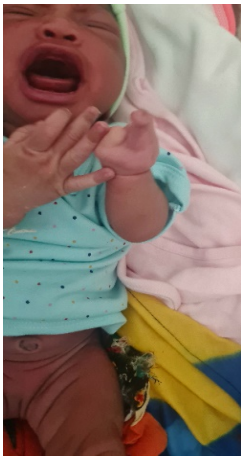
The 34-year-old mother had an unremarkable medical history, reported no teratogenic exposures or radiation, and had three previous uncomplicated pregnancies.

Neonatal Examination

Clinical evaluation revealed:

- Shortening of the right thigh, with the right ankle positioned at the same level as the left knee. (Pictur:1)
- Left-hand ectrodactyly.(picture:2)
- No additional external abnormalities.

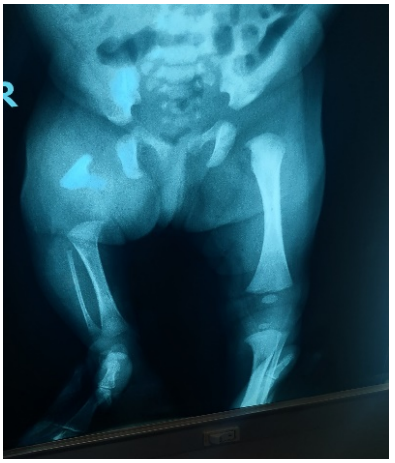
Picture 1



Picture 2

Imaging and Systemic Evaluation

Pelvic and femoral radiographs(picture :3) demonstrated findings consistent with proximal focal femoral deficiency. Systemic evaluations, including abdominal, cardiac, and central nervous system ultrasounds, were normal.



Management

The family was informed of the diagnosis and prognosis. Surgical intervention was not appropriate at this early stage. A staged, long-term rehabilitation plan and orthopedic follow-up were recommended.

Discussion

Missed Fetal Anomalies in Antenatal Care

This case underscores several factors contributing to missed fetal anomalies during ANC. Differences in resource availability, technician expertise, and institutional practices lead to inconsistent prenatal detection rates. Moreover, categorizing pregnancies into "low" and "high" risk based primarily on previous obstetric history may be misleading.

Systematic adherence to standardized anomaly-scan protocols should be maintained for all pregnancies, regardless of maternal history. Failure to do so may result in missed anomalies and compromised perinatal planning and counseling.

Skeletal Dysplasias and PFFD

Skeletal dysplasias form a diverse nosologic group with wide variability in presentation, prognosis, and genetic basis [1–4,8–13]. Prenatal diagnosis relies heavily on a structured ultrasonographic approach assessing long bone length, shape, mineralization, fetal profile, thoracic dimensions, and associated malformations [5,14,15].

PFFD involves deficiency of the proximal femur with varying degrees of hip and knee involvement. Classification systems such as Aitken, Bonafé, and Paley guide therapy by describing the anatomic severity and reconstructive feasibility [7–9,16].

Coexisting Ectrodactyly

Ectrodactyly, or split-hand/split-foot malformation, is a rare congenital anomaly that may co-occur with PFFD. It results from disruption of the apical ectodermal ridge during early limb development, leading to absence or hypoplasia of central digital rays. Functional impairment varies and management is individualized, including reconstructive surgery, prosthetics, and therapy.

Management Strategies

Management depends primarily on limb-length discrepancy (LLD), joint morphology, and functional prognosis. Treatment options range from conservative measures to complex limb-lengthening procedures.(table: 1)

Treatment options	**LLD (mm)**
Conservative management: no treatment, insole, or shoe raise	0-20
Consider epiphysiodesis of the contralateral limb at the appropriate age, *unless the child is already short* — in such cases consider lengthening of the short limb after specialist consultation. After skeletal maturity, **acute shortening of the long limb** is also an option.	20-50
Offer limb lengthening of the short limb; consider **epiphysiodesis of the long limb** as part of combined strategy	>50

Table 1. Treatment options according to limb-length discrepancy (LLD)

Conclusion

Proximal focal femoral deficiency is a rare but clinically significant congenital anomaly with long-term functional implications. The coexistence of ectrodactyly highlights the spectrum of congenital limb malformations. This case emphasizes the risk of missed prenatal diagnoses even in low-risk pregnancies and underscores the importance of standardized, expert-performed anomaly scans for optimal family counseling and long-term management.

Conflict of interests: No conflict to declare.

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