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Infant With Right Leg Swelling, Tenderness, and Decreased Range of Motion

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The Case

A 9-day-old girl, delivered at 39 weeks' gestation via cesarean delivery for footling breech position, presented with right leg swelling, tenderness, and decreased range of motion since birth (Figure 1).

Prenatal and Birth Histories

- 23-year-old gravida 2 and para 1 white mother
- History of gestational diabetes mellitus controlled with diet
- Group B Streptococcus-negative; hepatitis B surface antigen-negative; human immunodeficiency virus -negative; gonorrhea and chlamydia-negative, Rubella immune, rapid plasma reagin nonreactive
- Apgar scores 8, 9, with no resuscitation required
- Birthweight: 4,366 g

Case Progression

The patient was afebrile, nipple-feeding well, and alert after delivery. However, patient's parents stated that her right leg was "swollen and not moving" since birth. Mother stated infant became very fussy during leg manipulation and diaper

Figure 1.

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changes. Parents say they spoke to the nurse at the birth hospital and were told this is a normal result of breech presentation. The parents deny any history of trauma. The mother denies the infant has experienced any vomiting, diarrhea, bloody stools, apnea, or seizure activity. Parents state they took the child to her pediatrician on day 7 after birth for her newborn check and told the pediatrician about the leg. They were instructed to go to the emergency department. Parents went to emergency department that evening, and the child was admitted for overnight observation and then transferred to Children's Hospital of Michigan NICU the next day to investigate child abuse.

Social History

Patient's parents were living with paternal grandmother. Patient's 30-year-old father stated that he has 3 other children (ages 6, 8, and 11 years), none of whom he has custody. Patient's father states he lost custody of his children because he was "young."

Vital Signs

- Heart rate: 170 beats/min
- Respiratory rate: 46 breaths/min
- Oxygen saturation: 98% on room air
- Temperature: 36.8°C
- Blood pressure: 95/61
- Weight: 3,933 g (weight-for-length at 47th percentile)

Physical Examination

- General: Well nourished, well developed, active, no signs of distress
- Eyes: Pupils equal and reactive to light, normal conjunctiva, bilateral red reflex present
- Head, ear, nose, and throat: Normocephalic, intact palate, atraumatic, anterior fontanelle soft, symmetric facies, patent nares, no neck mass or crepitus, no lymphadenopathy
- Respiratory: Clear to auscultation bilaterally, equal breath sounds
- Cardiovascular: Normal S1, S2, regular rate and rhythm; no murmurs or gallops
- Gastrointestinal: Abdomen soft, nontender, nondistended, no organomegaly, bowel sounds present in all four quadrants
- Genitourinary: Normal term female genitalia; patent anus
- Musculoskeletal: Flexed and internally rotated right extremity, spine appears normal
- Skin: No icterus, birthmarks, or other rashes
- Neurologic: Irritable but consoles with feeding, symmetric Moro, normal strength and tone

Laboratory Studies

- White blood cell count: $15.9 \times 10^3/\mu\text{L}$ ($15.9 \times 10^9/\text{L}$) with 33% neutrophils, 60% lymphocytes, 2% monocytes, 5% eosinophils
- Red blood cell count: $5.56 \times 10^3/\mu\text{L}$
- Hemoglobin: 19.8 g/dL
- Platelet count: $310 \times 10^3/\mu\text{L}$ ($310 \times 10^9/\text{L}$)
- Sodium: 133 mmol/L
- Potassium: 6.8 mmol/L
- Chloride: 102 mmol/L
- Bicarbonate: 22 mmol/L
- Calcium: 10.4 mg/dL
- Magnesium: 2.1 mg/dL
- Phosphorus: 6.1 mg/dL
- Alkaline phosphatase: 245 units/L
- Alanine aminotransferase: 54 units/L
- Aspartate aminotransferase: 5 units/L
- Total bilirubin: 11.5 mg/dL; Direct bilirubin: 0.2 mg/dL
- Blood glucose: 84 mg/dL
- Urinalysis: normal

Patient's father presented several still images of the child after birth with a flexed and slightly internally rotated right lower extremity. Upon further questioning, family history was remarkable for brain tumors in a sibling, "coffee spots in a paternal aunt," and skin tumors in the paternal grandmother. Social work was involved to interview parents and assess home environment.

The clinical workup included a head computed tomographic scan, full skeletal survey, and dedicated right lower leg radiographs. Pediatric ophthalmology examined the child for retinal hemorrhages. Pediatric orthopedic surgery team was consulted. Patient's leg was placed in a cast. Patient was observed overnight while child abuse was completely ruled out as a cause of injury. Patient was discharged from the hospital with instructions to follow up in the orthopedic surgery and genetics clinic.

Differential Diagnosis

- Physiological bowing
- Tibial fracture in a term infant
- Focal cartilaginous dysplasia
- Osteogenesis imperfecta
- Child abuse
- Congenital syphilis
- Renal osteodystrophy
- Osteomyelitis
- Congenital pseudarthrosis of the tibia

Actual Diagnosis

Congenital pseudarthrosis of the tibia (Figure 2)

The Experts

The radiographs of the infant show an isolated fracture of the lower one-third of the right tibia, bowing of the right fibula, and anterolateral bowing of the tibia. The skeletal survey did not show any other fractures and normal bone. The clinical presentation and radiographic features are consistent with a diagnosis of congenital pseudarthrosis (CPA) of the tibia. From 40% to 90% of patients with CPA have neurofibromatosis.

Pseudarthrosis or “false joint” is a pathologic entity that may be an isolated congenital condition or may be associated with neurofibromatosis-1 [NF-1] (40%–90% of cases), fibrous dysplasia (15% of cases), or osteogenesis imperfecta. It also could be a complication at a fracture site. There is one case report of the association of CPA with vascular malformation. CPA of long bones results from a basic defect in the mesoderm, with presence of abnormal bone structure prone to bowing deformities, pathologic fracture, and poor healing.

The tibia is the most commonly affected bone, with an incidence ranging from 1 in 140,000 to 1 in 250,000 live births. Other long bones such as fibula, ulna, radius, and femur, also may be affected. Unlike the infant in the

vignette, most CPA are not present at birth, but the underlying disease process and deformation of the tibia usually are present at birth; it often is merely a matter of time before a first fracture occurs. The pseudarthrosis or site of nonunion is typically situated at the middle third and distal third of the tibia. The left tibia is affected slightly more than the right, and it is uncommon for the condition to be bilateral. The condition commonly is sporadic, although hereditary cases have been described.

The diagnosis of anterolateral bowing of the tibia often can be made at birth. The deformity is evident from the apical prominence laterally in the leg, with the foot inverted or at least medially displaced in relation to the lower part of the leg. If neonatal fracture has occurred, motion at the pseudarthrosis site will be evident. Because the vast majority of anterolateral deformities are unilateral, the shortening and angulation are easily noted when the affected leg is compared with the normal leg. If cutaneous signs of neurofibromatosis are present, the diagnosis is readily apparent.

Congenital pseudarthrosis of long bones occurs in about 3.6% to 5% of patients who have NF-I, whereas 40% to 90% of patients who have congenital long bone pseudarthrosis have NF-I. Solitary pseudarthrosis involving the tibia in a child may be the first clinical manifestation of NF-I. Because café au lait spots, axillary freckling, or other cutaneous manifestations of NF-1 may not be present in infancy but may appear later, the inability to diagnose NF-1 at birth, when the anterolateral bowing is first observed, does not mean that the patient may not eventually demonstrate NF-1.

Neurofibromatosis-1 is diagnosed when any two of the following seven features are present:

- *Café au lait macules* (≥ 6) are the hallmark of neurofibromatosis and are present in almost 100% of patients. They are present at birth and increase in size, number, and pigmentation, especially during the first few years after birth.
- *Axillary or inguinal freckling* that usually appears between ages 3 and 5 years with a frequency of $>80\%$ by 6 years of age.
- *Lisch nodules on the iris* (≥ 2). The prevalence of Lisch nodules increases with age, from only 5% of children younger than age 3 years, to 42% among children age 3 to 4 years, and virtually 100% of adults age 21 years or older.
- *Neurofibromas* (≥ 2) or one plexiform neurofibroma. These lesions characteristically appear during adolescence or pregnancy.

Figure 2.

- *Distinctive osseous lesion*, such as sphenoid dysplasia (which may cause pulsating exophthalmos) or cortical thinning of long bones (eg, of the tibia) with or without pseudarthrosis.
- *Optic gliomas* present in approximately 15% of patients.
- *First-degree relative* who had NF-1 and whose diagnosis was based on the aforementioned criteria.

The treatment for CPA of the tibia is difficult and has poor results. Regardless of the treatment method used (internal and external fixation, distraction osteogenesis, bone grafting with or without microvascular transfer of bone, electrical stimulation [either direct current by implantation or via pulsed electromagnetic fields]), there is general pessimism regarding the quality and longevity of any union that may be obtained, and the ultimate future function of the leg is uncertain. Amputation is recommended if shortening of >2 or 3 inches (5 to 7.5 cm) is anticipated, a history of multiple failed surgical procedures, and stiffness and decreased function of a limb that would be more useful after an amputation and fitting with a prosthesis.

Child abuse should be considered in nonambulatory children who have lower-extremity long-bone fractures. No fracture pattern or types are pathognomonic for child abuse; the fractures that suggest intentional injury include femur fractures in nonambulatory children, distal femoral metaphyseal corner fractures, posterior rib fractures, scapular spinous process fractures, and proximal humeral fractures. Multiple fractures in various stages of healing are suggestive of abuse; nevertheless, underlying conditions need to be considered.

The differential diagnosis includes conditions that increase susceptibility to fractures, such as osteopenia and osteogenesis imperfecta, metabolic and nutritional disorders (eg, scurvy, rickets), renal osteodystrophy, osteomyelitis, congenital syphilis, and neoplasia.

A skeletal survey is essential in every suspected case of child abuse to reveal other fractures in different stages of healing. Retinal hemorrhages are an important marker of abusive head trauma. In the child in this vignette, retinal examination results were normal. The clinical presentation and radiographic features in this infant are not typical for nonaccidental trauma or other metabolic or neoplastic bone disease.

American Board of Pediatrics Neonatal-Perinatal Content Specifications

- Know the clinical features and prognosis of birth injuries, such as fractures, lacerations, and facial palsies.
- Know the clinical features, diagnosis, management and outcome of neuromuscular disorders, including neurofibromatosis, tuberous sclerosis, Sturge-Weber syndrome, etc.

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