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about Neurofibromatosis

Congenital Pseudarthrosis of the Tibia/Fibula in Neurofibromatosis Type 1 (NF1)

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Although Pseudarthrosis can affect other bones such as the radius, ulna, and clavicle, this article will focus on the more commonly affected tibia and fibula in the case of NF1.

What is Congenital Pseudarthrosis?

Congenital pseudoarthrosis (CPT) is a term that describes a state of environment where the bone is prone to spontaneous fracture and heals poorly with routine treatment and remains at increased risk for re-fracture even after it heals. Even though the term “congenital” means present at birth, many times the bones at risk are bowed and deformed with no formal fracture at birth. However, the structural changes make the bones fragile and put them at increased risk for fracture with minimal or no trauma.

Once the bone is fractured, pseudoarthrosis – a state of non-union establishes itself. Although bones such as the radius, ulna, and clavicle could be affected, tibia and fibula are affected the most.

Congenital Pseudarthrosis of tibia/fibula and Neurofibromatosis Type 1

While approximately 5-6% of children with neurofibromatosis type 1 (NF1) have congenital pseudoarthrosis of tibia/fibula (CPT), up to 55% of children with CPT could have NF 1.

Although it is not fully understood, poor vascularity, soft tissue environment, and local biology leading to increased bone resorption at the fracture site in NF1 patients could be considered among many factors behind congenital pseudoarthrosis. Patients with NF1 may have lower than normal 25-Hydroxy Vitamin D levels.

What are the signs/symptoms?

Often the first sign of Pseudarthrosis is a deformity of the leg, such as tibial bowing, or a confirmed fracture that has occurred with minimal trauma. Anterolateral bowing is commonly present within the first year of life, with fracture most commonly occurring by the time the child is 2-2.5 years old.

How is Pseudarthrosis diagnosed?

X-rays are the primary way of diagnosing Pseudarthrosis along with a physical exam by your doctor.

Provided by NF Midwest www.nfmidwest.org

*This resource should not be used as a substitute for professional care or advice.
Consult with a qualified healthcare professional.*

What is the treatment for Pseudarthrosis?

Bracing the leg early can be preventative against fractures and aids in stability as your child becomes mobile. Commonly a KAFO (knee-ankle-foot orthoses) or an AFO (ankle-foot orthoses) is fitted by a specialist and then worn most hours of the day, or as your doctor sees fit. Surgical interventions are the common long-term treatment in Pseudarthrosis, especially if the bone has already fractured. Rod placement, internal or external fixation, & bone grafting are options, as well as amputation. Seeking out an orthopedic surgeon who has experience in this disease will be important as you explore the right treatment plan for your child. Advances in medical treatments, as well as surgical techniques have really improved the outcomes of what has been considered a difficult disease to treat.

Are there long-term complications?

Bone refracture can be minimized with proper care but does remain a risk especially among children. It is thought once a child reaches full skeletal maturity (full growth) the risk for fracturing drops significantly. Limb length discrepancy and ankle deviations can also be a common complication. Your doctor will keep a close eye on this and may use shoe lifts to help compensate the shorter limb. Surgical interventions can also be discussed as a long-term solution. Pain related to fractures or surgical recovery can be managed by your surgical team.

Living with Pseudarthrosis

There is no absolute cure for Pseudarthrosis, but with proper care and treatments your child can live a relatively normal life. Some physical limitations may be present and some benefit from physical therapy to help improve their physical quality of life. Finding the right provider to care for you is important. Wearing a brace is a common practice, especially in children, to help prevent additional fracture(s). The future only looks brighter as advances in medical technology, orthotics, prosthetics, & surgical interventions improve. The outcomes for people living with Pseudarthrosis are more optimistic now than ever before.