Ulnar Pseudarthrosis in a Child with Type 1 Neurofibromatosis

A 9-year-old boy was evaluated for a malformation of his right forearm present from birth (Figure 1). Type 1 neurofibromatosis was confirmed by genetic testing. The mother and maternal grandmother were also affected by neurofibromatosis with only skin involvement. Initially the forearm malformation was attributed to hyperlaxity but a radiograph showed evidence of dysmorphism of the ulna, with bone deficiency along the stem (Figure 2).

In patients with congenital nonunion of long bones, 50%-80% have an underlying diagnosis of type 1 neurofibromatosis. The prevalence of nonunion in patients with type 1 neurofibromatosis is 5%.[1] The formation of pseudarthrosis is a result of the nonunion between bone fragments of a long bone fracture and the formation of a false joint; the site most commonly affected is the tibia. This mechanism can occur in type 1 neurofibromatosis following bone dysplasia that can cause alterations in the repair processes.[2] Pseudarthrosis of the forearm is rare and usually involves the ulna. Radiologic findings may include cystic lesions, bone deformity, and cortical thinning; anecdotal cases of osteolytic reaction are described. Surgical treatment is recommended in the event of a fracture to prevent recurrence and includes bone fixation and/or debridement of the nonunion fibrous tissue between the bone segments.[3] ■

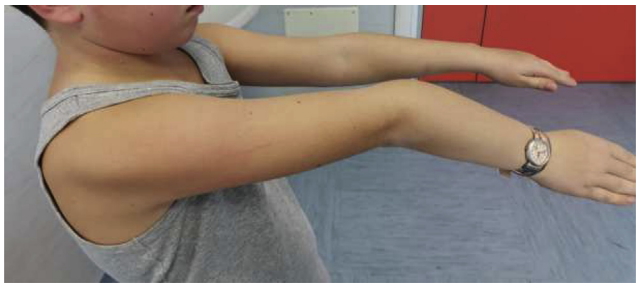

Figure 1. Right forearm with evidence of malformation and varus deviation of the right upper limb.

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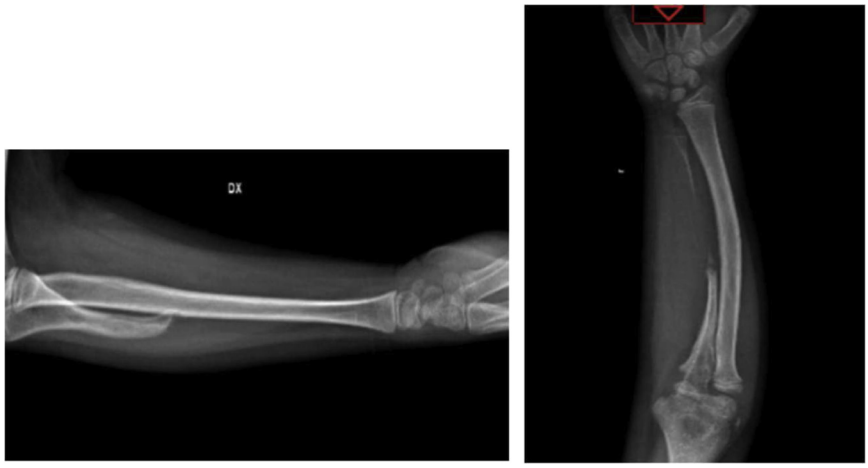

Figure 2. Dysmorphism of the ulna, with bone deficiency along the stem, fragmented in the middle third shaft with loss of distal trophism.

The authors declare no conflicts of interest.

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Ecthyma Gangrenosum in an Infant with Interleukin-1 Receptor-Associated Kinase 4 Deficiency

A 1-year-old boy was transferred to our hospital due to severe septic shock that led to cardiac arrest. The patient reportedly had fluid-refractory septic shock that required catecholamine administration. On admission, a characteristic skin rash covered the patient's entire body (**Figure 1**). His right buttock and thigh were reddish–purple and swollen (**Figure 2**). An incision was made in the right thigh, and the patient was diagnosed with necrotizing fasciitis. Surgical debridement was performed within the first 5 hours following his arrival. Wound and blood cultures revealed a *Pseudomonas aeruginosa* infection. The patient underwent continuous renal-replacement therapy with an endotoxin adsorption filter, empiric antimicrobial therapy (meropenem and ciprofloxacin), and necrotic tissue excision. Based on the drug susceptibility test results, the patient's treatment was de-escalated to piperacillin monotherapy. On the 31st hospitalization day, the infant died of intra-abdominal hemorrhage due to liver failure-associated coagulopathy. Rapid screening via flow cytometric analysis of monocytic intracellular tumor necrosis factor-α production in response to lipopolysaccharide suggested a Toll-like receptor signaling pathway deficiency. The postmortem sequence analysis of the *IRAK4* gene revealed a homozygous frameshift mutation (c.167_172insA).

The skin rash covering the entire body was ecthyma gangrenosum (**Figure 1**), caused by *P aeruginosa* bacteremia. Ecthyma gangrenosum is characterized by a central necrotic ulcer, sharp edges, and an erythematous halo. First described in 1897, ecthyma gangrenosum is a distinct

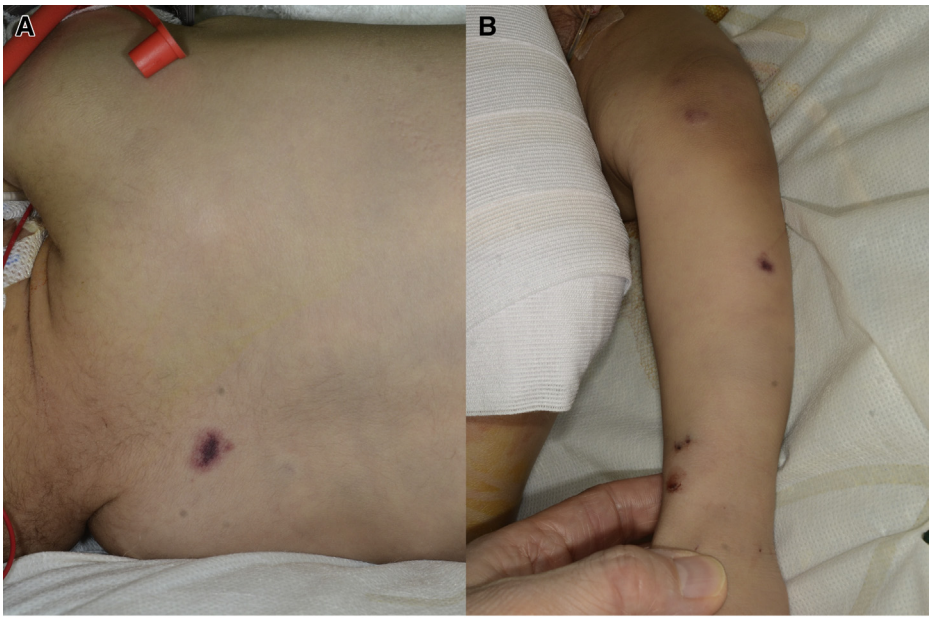

Figure 1. **A,** Ecthyma gangrenosum in the back, measuring 10 mm in diameter. **B,** Ecthyma gangrenosum in the left foot.