

# Lesion in the Foot at Birth: A Sinister Diagnosis

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A 4-month-old male child was brought to the out-patient department of our tertiary-care hospital with a swelling in the right foot, which was noted since the day of birth. It was increasing in size and had bled profusely a month ago, which was managed with compression. On examination, a single non-pulsatile, well-defined reddish-purple lobulated mass of size 7.2 x 7.8 cm was noted in the distal half of the right foot, involving the plantar side of the fore foot and mid-foot. There was a healed ulcer on the ventral aspect of the lesion. The 3<sup>rd</sup>, 4<sup>th</sup> and 5<sup>th</sup> toes were engulfed by the mass and grossly disfigured [Figure 1]. The mass had a variegated consistency, being firm in some areas and hard in the rest. The blood investigations of the child were normal, including hemoglobin and platelet count.

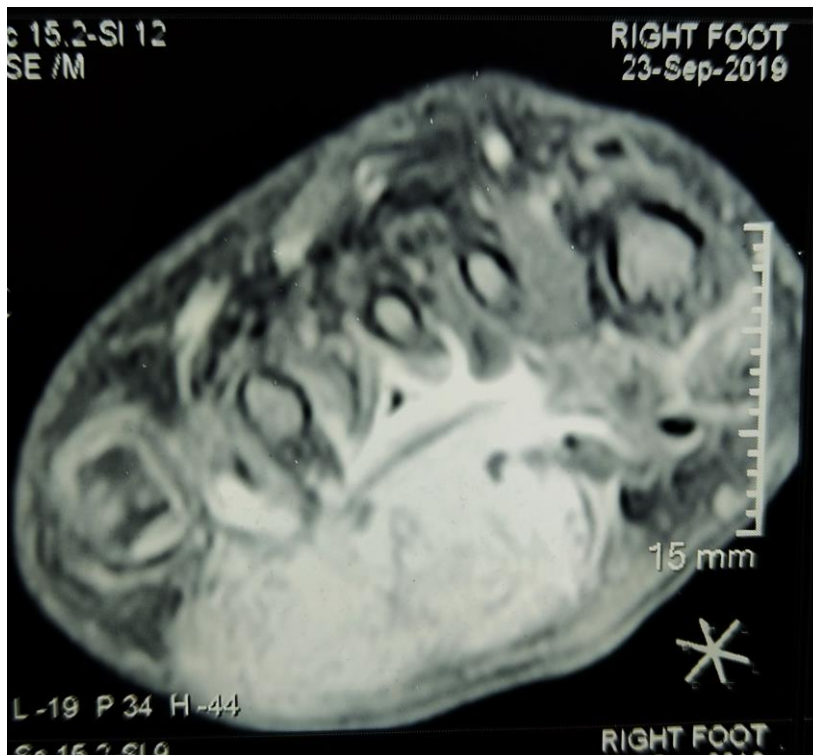


**Figure 1:** Reddish-purple mass in the distal half of the right foot

A plain X-ray of the foot suggested a soft tissue swelling with few subtle radio opacities within the lesion suggestive of phleboliths [Figure 2]. Ultrasound doppler revealed multiple arterial channels with high flows and dilated venous channels. Contrast enhanced MRI showed a highly vascular mass in the right forefoot encasing the 4<sup>th</sup> toe, extending to 3<sup>rd</sup>, 4<sup>th</sup> and 5<sup>th</sup> web spaces and 3<sup>rd</sup> and 4<sup>th</sup> metatarsal heads, along with the respective proximal phalanges [Figure 3]. Early arterial enhancement was noted.



**Figure 2:** Xray of the foot showing shadow of the lesions, leading to splaying of the digits. Subtle phleboliths are appreciable in the distal foot.



**Figure 3:** Contrast enhanced MRI- mass in the right forefoot on the plantar aspect of the foot extending dorsally to web spaces and metatarsal heads

**Question**

What is the most likely diagnosis?

- a) Benign vascular lesion
- b) Congenital infantile fibrosarcoma

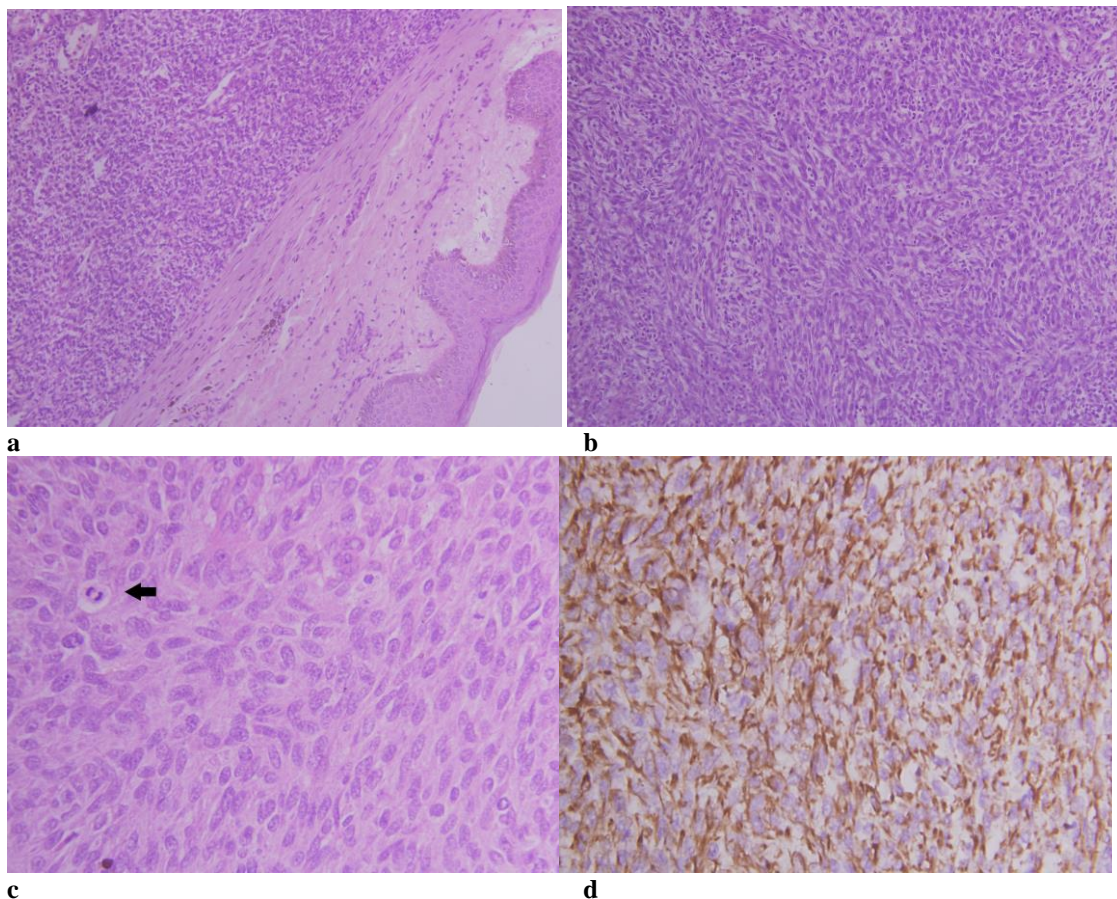
- c) Infantile myofibroma
- d) Spindle cell rhabdomyosarcoma

**Answer**

- b) Congenital infantile fibrosarcoma

**Discussion**

An excisional biopsy in the form of partial forefoot amputation was done and the wound was covered with a split thickness skin graft. On gross sectioning, a pale grey lesion was noted with focal areas of hemorrhage. Histological examination revealed a cellular mass composed of sheets of monomorphic spindle cells with nuclei showing plump oval to short spindled appearance, arranged in the form of vague bundles [Figure 4 a and b]. There were focal areas showing few lymphocytes admixed with scattered interspersed thin-walled blood vessels. Mitotic figures with minimal pleomorphism were identified [Figure 4c]. The cells were found to be immunoreactive for vimentin [Figure 4d] and negative for cytokeratin, epithelial membrane antigen (EMA), S100, desmin, myogenin and myo D1. A diagnosis of congenital infantile fibrosarcoma (CIF) was made. After ruling out regional and distant metastasis, a below knee amputation was done to achieve surgically clear margins. The child is disease free at 4 years follow up.



**Figure 4:** (a) Photomicrograph showing monomorphic cellular mass with multiple vascular spaces in the dermis- H&E, magnification = 40 x. (b) Sheets of monomorphic spindle cells plump oval to short spindled nuclei, arranged in the form of vague bundles with few infiltrating lymphocytes- H& E, magnification = 100 x. (c) Mitosis (black arrow)- H&E, magnification = 400 x. (d) Vimentin showing cytoplasmic positivity- IHC, magnification = 100 x.

Although congenital infantile fibrosarcoma is rare, it is one of the commonest soft tissue sarcomas in infants.<sup>1</sup> The distal extremities are the most frequent site of its occurrence, which is also associated with lesser risk of metastasis and mortality compared to its occurrence at axial sites such as the head and neck or trunk.<sup>2,3</sup> More than a third of affected infants present at birth and majority of the rest present during the first year of life.

The high degree of vascularity, erythematous-purple color, bleeding episodes and presentation in early infancy all may lead to the misdiagnosis as vascular lesions such as hemangiomas, kaposiform hemangioendothelioma, venous malformations, etc. both clinically as well radiologically, thus delaying the diagnosis and treatment.<sup>4,5</sup> Spindle cell rhabdomyosarcoma is another differential diagnosis but it presents in older children around 7 years of age, usually in the paratesticular and head and neck regions and is less commonly seen in extremities. Immunoreactivity to desmin and myogenin clinches the diagnosis. Infantile myofibroma has a similar age of presentation as CIF which may occur as well circumscribed solitary or multi centric masses which are more commonly seen in the head, neck and trunk regions. However, it is composed of mature spindle cells in whorled pattern as well as small immature round mesenchymal cells within a central vascular area.<sup>4</sup> Further, these cells are immunoreactive to desmin, vimentin and smooth muscle actin (SMA), thus differentiating it from CIF. The presence of chromosomal translocation t(12;15)(p13;q25) for an ETV6-NTRK3 fusion gene is specific for CIF.<sup>2</sup> In case of a dilemma despite histopathological examination, Fluorescent in-situ Hybridization (FISH) to detect the translocation can be done for confirmation.

While the first line of treatment is wide local excision of the tumor (often leading to amputations of the distal limbs), neoadjuvant chemotherapy is often used to limit the extent of major disfiguring or debilitating surgery in the head and neck or trunk regions.<sup>1,2,5</sup>

The stumbling block of CIF lies in its correct and timely diagnosis as it is easily confused with other benign conditions. Pediatricians and general practitioners have a huge role to play in advising biopsy for suspicious soft tissue masses in the newborns and neonates.

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