PST PEDIATRIC SURGERY IN TROPICS

Pediatric Surgery in Tropics
2025 (Jul-Sep) Volume 2 Issue 3
ISSN 3049-3404(Online)
DOI 10.70947/PST2025.22

Rare case of Congenital Infantile Fibrosarcoma of hand – A case report

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Keywords

Congenital infantile fibrosarcoma (CIFS), soft tissue tumor, Neoadjuvant chemotherapy, Tumor excision.

Abbreviations

1.CIFS : Congenital Infantile Fibrosarcoma
2.NTRK3 : Neurotrophin-3 receptor
3.USG : Ultrasonography
4.MRI : Magnetic Resonance Imaging
5.CT: Computed Tomography

Abstract

Introduction: Congenital infantile fibrosarcoma (CIFS) is a rare paediatric soft tissue tumour that presents most commonly at or after birth within the first year of life. It occurs most frequently over the extremity on the distal segments, followed by head, neck, and trunk. The usual presentation is a painless soft tissue mass with extensive local invasion and rare metastasis. The recommended treatment of CIFS is wide local excision. In recent times, preoperative chemotherapy is found to be useful in unresectable tumors by reducing tumor volume and hence reducing the morbidity of an extensive resection.

Aim: We present this case for its rarity and to share our experience on successful multidisciplinary management without compromising limb function and cosmesis.

Methods: Case: A 4-month-old female infant presented to us with a history of swelling over the dorsum of the left hand. It was noticed at birth and subsequently increased in size over the next month. On a clinical exam the size of the mass was noted to be around 4 x 5cms. Magnetic resonance imaging confirmed a soft tissue neoplasm. The pre-treatment work up was completed with ultrasound guided core needle biopsy, which showed features of infantile fibrosarcoma. Taking into consideration the extensive soft tissue involvement of the hand, the kid received 9 cycles of neoadjuvant chemotherapy. This was followed by near total excision of the tumour. The child continues to be on follow up for the past three years. No recurrence has been noted till date.

Conclusion: Congenital infantile fibrosarcoma is a rare, locally aggressive tumour of childhood with limited management options. It mandates a multidisciplinary approach for reducing the morbidity of an extensive resection in order to provide a good functional and cosmetic outcome.

Introduction

Congenital infantile fibrosarcoma (CIFS) is a rare pediatric malignant soft tissue neoplasm that most commonly presents as a mass in distal extremities. This variant of soft tissue sarcomas is diagnosed at or soon after birth with 90% of patients being diagnosed in infancy. The findings on radiologic imaging are generally non-specific. Nevertheless, tissue biopsy is confirmatory and considered the gold standard. Though the histologic features resemble fibrosarcoma in adults, it is considered a separate entity in kids as a result of its variant clinical behaviour with a more favourable prognosis [1, 2]. Metastasis is rare. However, it is noted to be locally aggressive with greater chances of local recurrence. This tumor is often mistaken at birth as hemangioma or lymphatic malformation [3, 4]. A distinctive feature is the specific translocation [t (12; 15)] leading to the gene fusion ETV6-NTRK3. [5] The primary treatment is surgical resection whereas chemotherapy is reserved for non-resectable tumors and recurrent ones. [6, 7]
Only a few cases involving the hand have been reported and a limited number have presented in the newborn period. We report a case of congenital infantile fibrosarcoma localized to the hand of a neonate, detailing the clinical presentation, diagnostic workup including imaging studies and histopathological findings, and the multidisciplinary approach for its management with a successful outcome.

Case report

Case report: A 4 months old female child, delivered at term, presented with a swelling on the dorsum of the left hand since birth [Fig. 1]. The swelling was gradually increasing in size. On physical examination, she was a healthy baby of 3 kg weight with an ill-defined mass involving the left hand, measuring about 3x4cms. It was not warm or tender and was firm in consistency. The skin over the swelling was tense and shiny. Her physical examination was otherwise unremarkable.

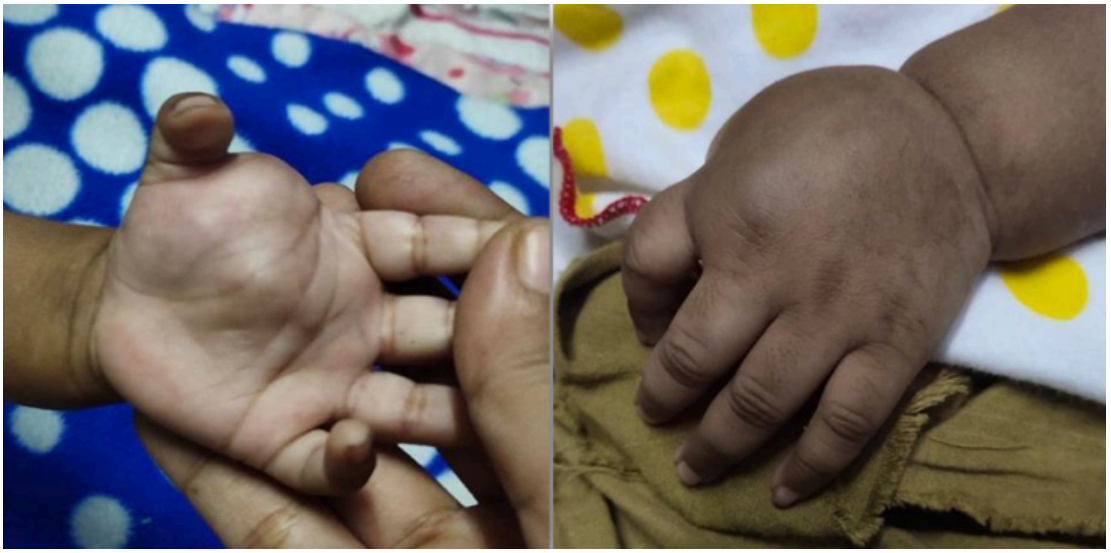

FIG. 1: Clinical presentation picture showing the mass over volar and dorsal aspect

A plain radiograph of the left hand revealed a soft tissue mass. Magnetic resonance imaging (MRI) of the left hand showed a well-defined enhancing

lesion involving the intramuscular plane of both palmar and dorsal aspect of hand with significant widening and bony remodeling changes, suggestive of slow growing soft tissue neoplasm. [Fig. 2] There was no evidence of systemic or distant spread from the primary lesion.

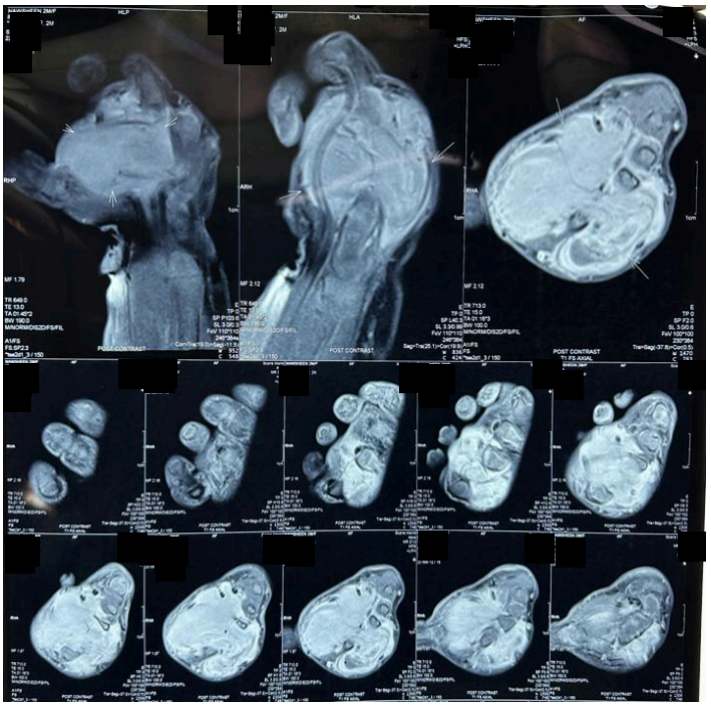

FIG.2: MRI picture of the left hand

Histopathology of a core needle biopsy from the mass revealed the features of an infiltrating neoplasm composed of round to spindle-shaped cells with ovoid nuclei and foamy cytoplasm [Fig. 3]. Immunohistochemistry of the slides proved to be positive for vimentin and negative for MyoD, Myogenin, Cytokeratin, Neuron Specific Enolase and CD99.

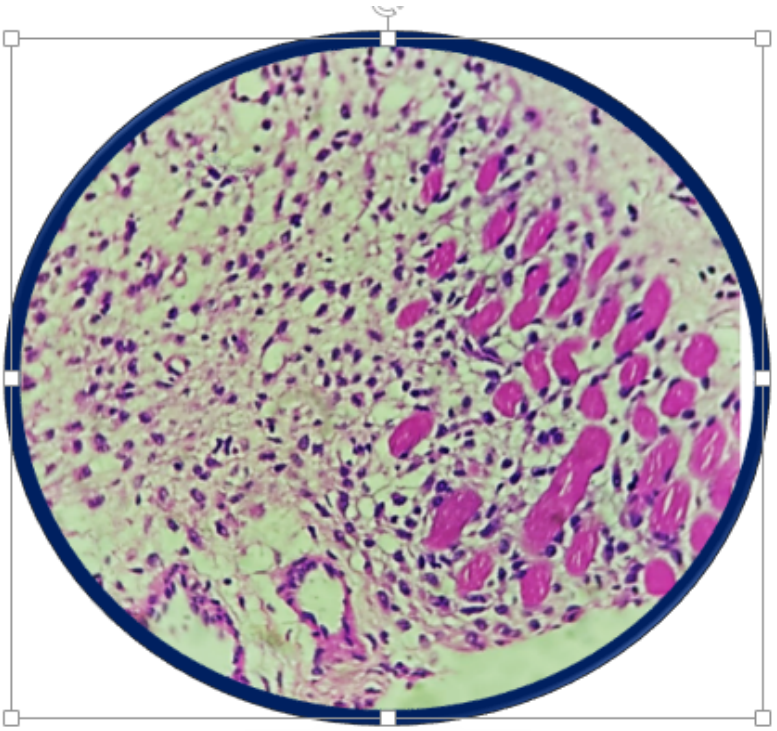

Figure 3: Histopathology pictures

In addition, the cells were positive for NTRK3-ETV6 gene fusion on polymerase chain reaction.

Based on these findings on the pathological examination, a diagnosis of CIFS was made. The tumour at presentation was considered inoperable as a consequence of its location and ill-defined margins between the tumour and rest of the hand. A multidisciplinary team consisting of a Paediatric Surgeon, Paediatric Plastic Surgeon and a Paediatric Oncologist decided to start the kid on neoadjuvant chemotherapy, ensuing a tumour board discussion.

The baby received Vincristine and Dactinomycin as neoadjuvant chemotherapy based on an European Paediatric Soft Tissue Sarcoma Study Group (EpSSG) protocol [9]. After completing 9 cycles of chemotherapy, an MRI was repeated which showed significant reduction in size of tumor by approximately 75%. After meticulous planning a surgical removal of the tumour was considered with focus on preserving

both the structural integrity and functional capacity of the delicate hand anatomy and to provide good cosmesis. After meticulous dissection, piecemeal excision of the mass was done, safeguarding the important tendons, ligaments and nerves of the involved area of the hand [FIG. 4].

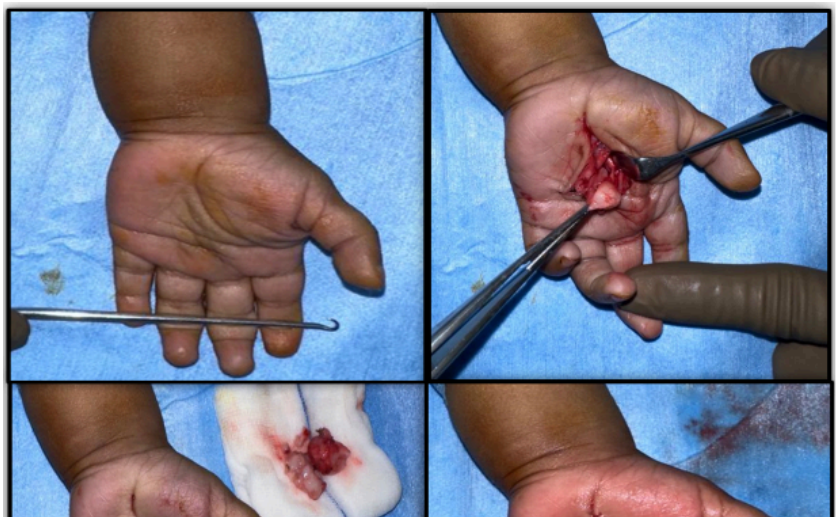

Fig 4 : Intraoperative pictures showing the incision and final outcome
Histopathology of excised mass showed hypercellular areas of spindle cells arranged in sheets and vague fascicles with areas of hyalinization and lymphoid aggregates without increased mitosis or necrosis confirming the diagnosis. The kid continues to be on follow with us. Three years post excision, he is disease free with no sign of clinical or radiological recurrence. [Fig 5]

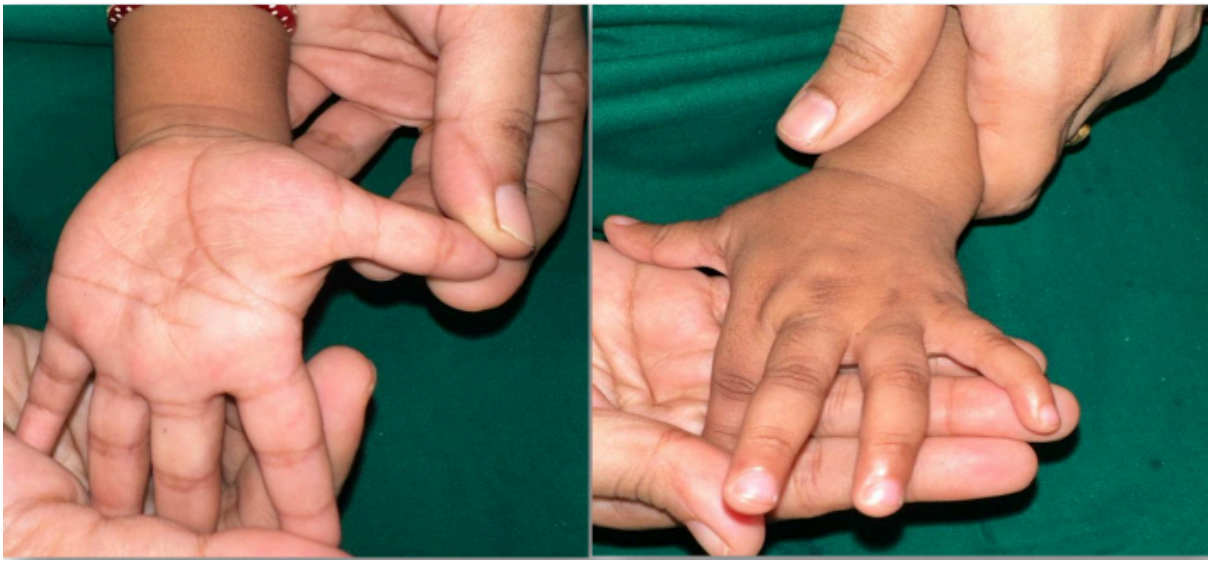

Figure 5 : Postoperative pictures on follow up

Discussion

Congenital infantile fibrosarcoma (CIFS) is a rare malignant soft tissue tumor that usually occurs at birth or in the neonatal period, mostly presenting within the infantile period [7]. They originate from primitive mesenchymal tissue characterised by proliferation of fibroblasts. CIFS occurs most frequently on an extremity followed by head, neck, and the trunk. When involving an extremity, it more commonly involves the upper rather than lower and distal rather than the proximal end [8]. It is usually located in the subcutaneous or deep soft tissue, and is characterised by rapid growth, reaching a large size within a few weeks or months. It is generally poorly circumscribed and infiltrates subcutaneous fat, muscle, fascia, and tendon tissues.

Clinically, it presents as fast growing soft tissue swelling that is difficult to distinguish from hemangioma, lymphatic malformation and fibrosarcoma. Other differential diagnoses include infantile fibromatosis/myofibromatosis, malignant fibrous histiocytoma, malignant peripheral nerve sheath tumor and rhabdomyosarcoma. X-ray shows soft tissue swelling that may obliterate normal fat planes and deform or destroy the adjacent bone. USG shows a solid heterogeneous echogenic mass with some cystic areas and Doppler sonography may show a highly vascular pattern similar to hemangioma. MRI findings of CIFS include a mixed cystic and solid tumor of heterogeneous density which typically has inhomogeneous enhancement. MRI helps to delineate the extent of a lesion and involvement of adjacent structures like neurovascular bundle, muscles, joints or bones. MRI is particularly useful in showing the disruption of the soft tissue planes. Limitations of MRI include the inability to differentiate residual or recurrent disease from postoperative edema, inflammation and haemorrhage [11]. CT is superior to MRI in the evaluation of osseous involvement and therefore should be performed when osseous invasion is suspected.

Histology is characterised by sheets of solidly packed spindle shaped cells arranged in bundles and fascicles resembling a herringbone pattern. Other features include well oriented fibroblasts, scattered round cells and chronic inflammatory cells like lymphocytes. Multinucleated giant cells are rare whereas mitotic figures are common and interspersed with areas of increased vascularity. At Immunohistochemistry, CIFS are usually vimentin positive, desmin negative, and S100 negative. [12] CIFS is highly associated with a t (12; 15) (p13; q25) translocation, which creates a fusion between ETV6 (TEL) and NTRK3 (neurotrophin-3 receptor) genes, thus providing a useful diagnostic marker. This translocation is detected by reverse transcriptase-polymerase chain reaction using frozen or paraffin-embedded tissue. [5, 10] Though this is used to confirm the diagnosis of CIFS, ETV6/NTRK3, fusion negative cases have been described in literature.

While CIFS shares histological similarities with fibrosarcoma in adults, it exhibits distinct clinical behaviour. Unlike the adult variant, metastases are rare though local recurrence is common. Despite these differences, the prognosis of CIFS is notably favourable, with a 5-year survival rate of 84 to 90%. [7, 8]

A wide range of treatment modalities are described for Congenital Infantile Fibrosarcoma (CIFS), ranging from wait and watch, to a combination of surgical intervention, chemotherapy, or both. Surgical options include subtotal resection, gross total resection, and wide resection. These may involve potentially disfiguring surgeries or even amputations.

When the tumor's extent or size renders surgical therapy challenging, such as involvement of the neurovascular bundles or loss of limb function, Neoadjuvant chemotherapy needs due consideration. It helps in reduction of tumor size and hence facilitates a more conservative surgical strategy as in our case, thereby avoiding mutilating surgeries. This approach underscores the importance of good functional outcomes for patients. Recent advancements include dual neoadjuvant therapy involving vincristine-actinomycin-D. [6, 7]

This case testifies the therapeutic challenges in the treatment of CIFS due to diffuse soft tissue involvement of the extremities, involving a poorly circumscribed tumour, infiltrating subcutaneous fat, muscle, fascia, neurovascular bundle and tendon tissues. Wherein initial surgical excision preserving functionality of hand could not be accomplished and the child was started on neoadjuvant chemotherapy with Vincristine and Actinomycin D. This resulted in reduction of the tumor bulk and aided with the planning and execution of surgical removal of the tumour. Furthermore, our case underscores the significance of multidisciplinary team involvement in navigating complex scenarios, from diagnosis till completion of therapy. Their combined efforts ensured optimal tumor management and enhanced the overall quality of care for the patient. We, however, agree that a longer follow-up period (the child is currently recurrence-free for three years) will be necessary to declare the child cured.

Conclusion

CIFS is a rare pediatric soft-tissue tumor which requires a high index of suspicion and careful evaluation for diagnosis. Management requires involvement of a multi-disciplinary team. Neoadjuvant chemotherapy should be considered for large tumors with significant involvement of vital structures, thereby compromising functionality. Efforts should be directed towards limb conservation, thereby avoiding radical surgeries.

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Received on 23rd March 2025	Published on 1st july 2025
Acknowledgements None	Conflicts of interest None
Source of funds None	Ethical clearance Obtained
Citation Sihag A, Kamraj SG, Sridhar S et al. PST July 2025 V:2 I:2 p	https://doi.org 10/70947/PST2025.21
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