Case Report

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# Unusual case of congenital/infantile fibrosarcoma in a new born

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## ABSTRACT

Congenital infantile fibrosarcoma (CIFS) is a rare mesenchymal tumor that is primarily developed in the soft tissue of distal extremities and occasionally in unusual locations such as the lung and retroperitoneum. It occurs mainly in children below the age of 5 years. About 200 cases have been reported in the literature so far, very few of them in new-borns. The prognosis of this tumor is relatively good compared to adult forms. We report an unusual case of CIFS occurring in new–born mimicking an hemangioma and causing hemorrhage in the neonatal period. The tumor is located in the left arm and axilla and associated with a hand malformation. A shoulder amputation is performed after chemotherapy failure. The infant is now two-years old with no recurrence.

**Key words:** Chemotherapy, Childhood fibrosarcoma, surgery

## INTRODUCTION

Congenital or infantile fibrosarcoma (CIFS) is a rare soft-tissue neoplasm that represents less than 1% of all childhood cancers[1] and should be considered in the differential diagnosis of a large extremity mass presenting at birth. It accounts for approximately 5-10% of all sarcomas in children younger than 1 year old.[1] These tumors are notoriously misdiagnosed at birth as either hemangiomas or lymphatic malformations. The fusion transcript ETV6-NTRK3 resulting from the specific chromosomal translocation *t(12;15)(p13;q25)* is now a useful diagnostic tool for CIFS.

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Surgery with wide resection is the mainstay of treatment. However, if initial surgery cannot be done without mutilation or is impossible, preoperative chemotherapy should be given. Amputation should be reserved for chemoresistant patients in whom the involvement of neurovascular structures by the tumor make a limb-sparing aggressive excision impossible.

Although histologically similar to fibrosarcomas occurring in adults, the congenital lesions differ in their clinical behavior, metastases are rare, less than 10%, local recurrence is common, and the prognosis is good. Overall survival is given as high as 90% in different series.

The aim of our presentation is to report an unusual case of CIFS by its localization and rapid evolution. We recall the importance of early diagnosis in this tumor.

## CASE REPORT

We report the case of a new-born female from a normal pregnancy. Prenatal diagnosis birth weight was 3kg and the apgar 9/10.

Physical examination showed a large mass of the inner arm and axilla measuring 6cm. The tumor was bleeding and the new-born required a transfusion. Otherwise, there was a hypoplasia of the ipsilatéral hand [Figure 1]. Initial diagnosis was hemangioma.

There was no bone lesion in the humerus and the scapula. Radiography of the upper limb showed agenesis of hand bones.

Evolution was marked by the rapid increase in tumor volume. Tomodensitometry revealed a tissular mass measuring 75 × 65 × 84 mm. The mass infiltrated the vascular bundle and the skin outside [Figure 2].

A biopsy was performed to establish nature of this mass. Micoscopically, the tumor was composed of

sheets of spindle cells showing staghorn blood vessels (hemangiopericytoma-like vasculature) [Figures 3, 4]. The tumor cells were positive for vimentin, focally positive for smooth muscle actin and negative for EMA, CD34, S100, myogenin, HMB-45, ALK-1, and desmin immunostains. Because of the spindle nature of the neoplasm and the presence of prominent staghorn blood vessels, CIFS was strongly considered in the differential diagnosis. A conventional karyotype of the tumor could not be reported to support the presence of *t(12;15)* characteristic of CIFS.

Chemotherapy was the first attempt because of tumor's volume. The child received vincrystine and actinomycin (Act-D 1mg/m$^2$ day 1; vincristine 1.5mg/m$^2$ days 1, 7, 14). There was toxicity due to chemotherapy and no reduction of tumor's volume. A shoulder disarticulation was performed because limb-preservation surgery would be impossible as bundle of the compartment were invaded. The child is now 2 years old with no complications or recurrence.

## DISCUSSION

Infantile fibrosarcomas (CIFS) are soft tissue sarcomas that are diagnosed at or soon after birth.[2] Its etiology remains unknown. Fibrosarcoma constitutes approximately 10% of soft tissue sarcomas in children.[3] It is usually regarded as a tumor of borderline or low malignant potential, like most of the fibrohistiocytic tumors of this age. CIFS is a rare tumor and only a few cases are reported as congenital. We have reviewed 107 cases reported in the literature in children under one year of age until 2010.[1,2,4-10]

About 40% of CIFS are diagnosed at birth, and 60% before 3 months. Boys are more often affected than girls (sex-ratio = 3/1 to 4/1)[11]. CIFS affects chiefly the distal portions of the extremities, but there are unusual presentations as lung, retroperitoneum and heart.[5,6] In our presentation the tumor affects the proximal portion of the arm. Prenatal diagnosis is possible from 26 weeks of gestation by ultrasound and magnetic



**Figure 1: Gross morphology of the infantile fibrosarcoma**

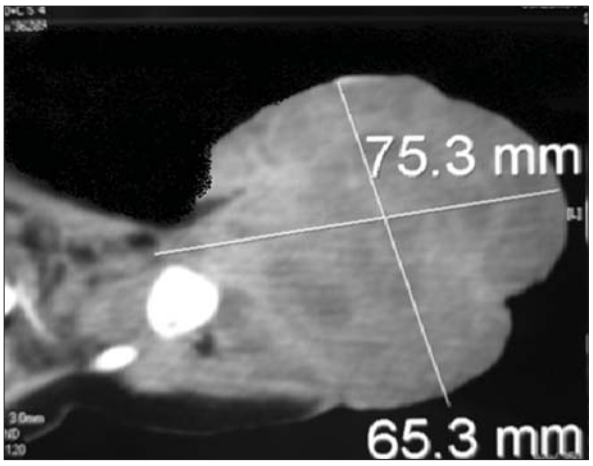



**Figure 2: CT scan appearance of the tumor**

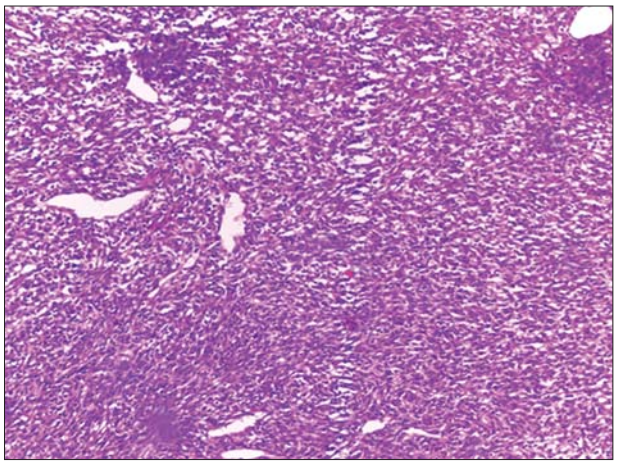


**Figure 3: Spindle cell neoplasm with a staghorn blood vessel ( 200×)**

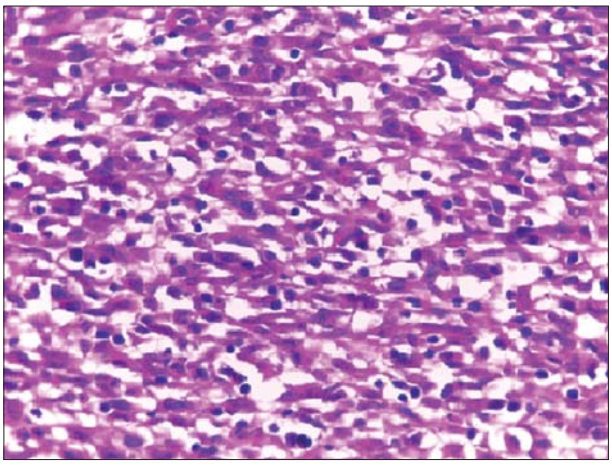


**Figure 4: Spindle cell neoplasm with lymphocytic infiltrate (400×)**

resonance imaging (RMI).[8] At birth, the tumor is sometimes confused with hemangioma and can cause fetal anemia and requires transfusion as in our patient.[8] Our presentation is the first showing an association of CIFS with a hand malformation, we have found no publication of association with a limb malformation. We have not been able to explain this association.

The typical histologic appearance of infantile fibrosarcoma is a solid, densely cellular proliferation of spindle cells in interlacing bundles and sharply intersecting fascicles with a focal herringbone pattern as in our patient. Histologic variants include myxoid, small round cell, and whorled patterns. Immunohistochemistry reveals positivity with vimentin. S100 protein is negative. In some cases there is focal positivity with desmin, smooth muscle actin and cytokeratin.[9] CIFS has been found to contain a novel recurrent reciprocal translocation *t(12;15)(p13;q25)* resulting in the gene fusion ETV6-NTRK3 (ETS variant gene 6; neurotrophic tyrosine kinase receptor type 3). This translocation is found in more than 70% of cases.[11] Some authors suggest a correlation between the presence of this translocation and chemosensitivity.[5]

The primary therapy of CIFS is surgery. Despite rapid growth and large size, the majority can be cured by wide local excision or amputation. Chemotherapy may shrink the tumor to facilitate complete resection. The chance of developing metastatic spread is rather small (5 – 8.3%).[3] Chemotherapy can also be used to treat local recurrences and metastases. In literature, we found some cases of CIFS treated successfully only with chemotherapy.[11] Specific drug sensitivities are not clearly defined but Vincristine and Actinomycin D are the most commonly used drugs. In our patient, we opted for primary chemotherapy to prevent metastasis, especially after finding a rapid increase in tumor volume in a few days. We decided to perform an amputation of the upper limb. We think that radical treatment is the only therapeutic option of proximal CIFS of limb, healing and functional adaptation are fast at this age. The 5-year survival rate for CIFS is high and has been reported between 84% and 93%.[10] Our patient is now two years old with no recurrence.

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