Congenital Infantile Fibrosarcoma: A Clinical Mimicker and Histopathological Surprise

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Abstract

Congenital infantile fibrosarcoma is defined as fibrosarcoma in children below 2 years of age. It is a rare soft-tissue malignancy with only 300 cases reported in the world literature. Herein is presented a case of a 10-month-old infant with a soft-tissue swelling over his right sole. The swelling was present since birth with sudden increase in size and change in color in the overlying skin. With a clinical diagnosis of a vascular tumor, excision was performed. Microscopy showed a cellular tumor with spindle-shaped cells arranged predominantly in sheets. Atypical mitosis was seen. Scattered lymphocytes were also seen. The tumor cells were positive for vimentin. A final diagnosis of congenital infantile fibrosarcoma was made. Clinically, it can be confused with a tumor of vascular origin. Compared to its adult counterpart, the clinical course of infantile fibrosarcoma is a favorable one. It is intended to describe this case due to its rarity and to increase awareness about this entity amongst clinicians and pathologists to prevent misdiagnosis.

Keywords: Aggressive, Congenital, Infantile fibrosarcoma.

Introduction

Pediatric soft tissue sarcomas are rare and constitute only 7% of all childhood tumors. Of these, congenital infantile fibrosarcoma is a rare soft-tissue malignancy and represents less than 1% of all childhood cancers. However, it is the most common soft-tissue sarcoma in children under 1 year of age. Approximately 300 cases have been reported in world literature till date. While 50% of these tumors are present at birth, the remainder develop during the first 2 years of life. Compared to its adult counterpart, the clinical course of infantile fibrosarcoma is a favorable one. Clinically, they tend to mimic vascular or lymphatic tumors and thus, clinical and pathological awareness of this entity is essential to prevent misdiagnosis.

Case Report

A 10-month-old infant presented with a swelling over his right sole. The swelling was present since birth measuring 2×2×1 cm initially. It suddenly increased in size in the past 2 months to the present size of 4×4×3 cm. This increase in size was associated with a change in color in the overlying skin. On examination, the swelling was non-tender, mobile and soft-to-firm in consistency. X-ray revealed a soft tissue swelling with no involvement of the underlying metatarsal and tarsal bones (Fig. 1). Excision of the mass was performed with an initial diagnosis of a vascular tumor. The mass was sent for histopathological examination.

Grossly, the tumor was poorly circumscribed and soft-to-firm in consistency. Cut surface was greyish white with areas of hemorrhage. Microscopy showed a cellular tumor with cells arranged predominantly in sheets. Focally herringbone pattern could be identified. The individual tumor cells were spindle-shaped with moderate amount of eosinophilic cytoplasm and a spindle-shaped hyperchromatic nucleus. There was mild degree of nuclear pleomorphism with presence of atypical mitosis (Fig. 2). Chronic inflammatory cells, particularly lymphocytes, were seen scattered in between these tumor cells (Fig. 3). Numerous congested blood vessels were noted. On performing immunohistochemistry, the tumor cells were positive for vimentin (Fig. 3) and negative for CK, EMA, SMA, desmin and CD 34. Based on the clinical findings, morphology and immunohistochemistry, a final diagnosis of congenital infantile fibrosarcoma was given.
Discussion

Congenital infantile fibrosarcoma is a relatively rare tumor first described by Stout in 1962.\(^5\) About 50% of these are diagnosed at birth with boys being affected more than girls (male:female ratio=3:1).\(^6\) This tumor mainly involves the lower extremities (72%), of which foot is the most common site. It can also involve the head and neck region particularly orbit, lung, heart, retroperitoneum and mesentery.\(^7,8\)

Clinically, congenital infantile fibrosarcoma can mimic a wide spectrum of neoplasms. On one hand, it has to be differentiated from lesions like infantile fibromatosis/ myofibromatosis and congenital hemangioma, which have a benign course, and on the other hand from the more aggressive neoplasms like congenital hemangiopericytoma and rhabdomyosarcoma.\(^9\) Histopathology and immunohistochemistry play a crucial role in the exact typing of the tumor and thereby deciding the future treatment modality.

Infantile fibrosarcoma typically has a densely cellular histological appearance. Herringbone pattern can be appreciated only focally in comparison to its adult counterpart where this is the predominant pattern in most instances. The cells show mild nuclear pleomorphism and are mitotically active. Scattered chronic inflammatory cells, particularly lymphocytes, are another common, sometimes a striking feature. Immunohistochemistry reveals positivity with vimentin and negativity for muscle markers like SMA and desmin. Based on this characteristic, morphology and deep-seated location of this tumor it can be distinguished from infantile fibromatosis. Desmin and SMA negativity helps in excluding embryonal rhabdomyosarcoma and infantile hemangiopericytoma respectively.
Congenital infantile fibrosarcoma 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) in more than 70% of cases.\textsuperscript{10} The primary therapy of congenital infantile fibrosarcoma is surgery. Some oncologists also recommend a pre-surgical chemotherapy regime to shrink the tumor and facilitate complete resection. Majority of the patients can be cured by wide local excision. Amputation is performed only in the rare cases of chemo resistance. The chance of developing metastatic spread is rather rare (5-8%).\textsuperscript{10} Regional lymph node spread is rare but hematogenous spread can occur with commonly to the lungs, bone marrow or abdominal organs. The reported 5-year survival rate for congenital infantile fibrosarcoma is high and has been reported between 84 and 93%.\textsuperscript{11}

**Conflict of Interest:** Nil

**References**


