Infantile rhabdomyofibrosarcoma: A potentially underdiagnosed aggressive tumor

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Abstract

Infantile Rhabdomyofibrosarcoma (IRMFS) is a rare clinicopathological entity that resembles infantile fibrosarcoma (IFS) but has ultrastructural and immunohistochemical evidence of rhabdomyoblastic differentiation. We report a 2 years and 6 months old boy who presented with a slowly progressive large soft-tissue mass in left axillary region. After complete excision, histopathology report revealed diagnosis of IFS. Review of the histopathology with immunohistochemistry (positive for desmin) revealed diagnosis of IRMFS. He was treated with aggressive adjuvant chemotherapy. He was in complete remission 6 months after completion of chemotherapy. In view of poor prognosis and aggressive treatment approaches for IRMFS, it must be differentiated from IFS to avoid under treatment.

Key words: Immunohistochemistry, infantile fibrosarcoma, infantile rhabdomyofibrosarcoma

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Introduction

Infantile Rhabdomyofibrosarcoma (IRMFS) is a rare clinicopathological entity that resembles infantile fibrosarcoma (IFS) but has ultrastructural and immunohistochemical (IHC) evidence of rhabdomyoblastic differentiation. Despite close histological resemblance, the prognosis and treatment of this tumor is different from that of IFS. Three cases of IRMFS were first reported by Lundgren et al. in 1993, and since then only three more cases have been added to the literature.[1-4] We report here a case of IRMFS, previously diagnosed as IFS, but subsequently found positive for desmin. Although rare, IRMFS must be differentiated from IFS to decrease the chances of undertreatment.

Case Report

A 2 years and 6-month-old male presented with a large soft-tissue mass in the left axillary region. The mass was painless and had slowly progressed in size over 10 months. He did not have any constitutional symptoms. On examination, the mass measured 15 cm × 15 cm. It was firm, bosselated, non-tender, free from the overlying skin, but adherent to the underlying tissue.

Computed tomography (CT) scan revealed large homogenously enhancing mass without any calcification or underlying bone involvement. During excision surgery, the mass was found to be well encapsulated and arising from the serratus anterior and adherent to the pectoralis, scapula, and subscapularis. Histopathology revealed a tumor composed of spindle cells, arranged in fascicles and whorls, having pale eosinophilic cytoplasm, hyperchromatic pleomorphic oval and spindle nuclei, with 8-10 mitoses/10 high power fields [Figure 1]. There was no evidence of hemorrhage or necrosis. The morphological diagnosis was reported as IFS. Histopathology review and immunohistochemistry revealed diagnosis of IRMFS with strong positivity for desmin, smooth muscle actin and vimentin, and negativity for myogenin-D1, myoglobin and cluster of differentiation (CD) 34 [Figure 2]. There was no evidence of metastases to the lung, bones or bone marrow. In view of an aggressive...
nature of the tumor, he underwent wide local excision and received adjuvant chemotherapy with weekly vincristine with 3 weekly actinomycin-D and cyclophosphamide for 1 year as per rhabdomyosarcoma (RMS) protocol. CT scan after treatment completion did not reveal any tumor. At the time of submission of this article, he was in remission about 2 years after completion of chemotherapy.

**Discussion**

Although, termed as infantile RMFS, this entity has not been diagnosed in infants. The median age was 24 months (range 13-48 months) with male: female ratio of 5:2. There was no specific site of predilection. IRMFS occupies an intermediate position between IFS and spindle cell RMS in its clinical presentation, behavior, morphology, IHC and ultrastructural features. The cells express vimentin, smooth muscle actin, and desmin but not myoglobin, myoD1 or myogenin (MyoD1 and myogenin not tested in earlier reports).[1]

Though karyotyping could not be carried out in our patient, the present case is in concordance with the previous studies in other characteristics [Table 1].

The treatment for IFS is primarily surgery and usually no adjuvant treatment is needed whereas RMS needs adjuvant chemotherapy with or without radiotherapy. Hence, overlooking the diagnosis of IRMFS might increase the chances of local recurrence or metastatic disease. Five out of six reported patients either were relapsed or developed metastases. Only one patient was in remission 25 months after multimodality treatment. Hence, aggressive multimodality

**Table 1: Clinical and IHC profile of the cases of IRMFS reported**

| Authors | Case no. | Age (months) | Sex | Site | IHC | Treatment | Outcome |
|---------|---------|--------------|------|------|-----|-----------|---------|
| Lundgren et al.[1] | 1 | 24 | F | Thigh | Vimentin | + | WLE+CT | Metastasis (P+L); death 1 year |
| Mentzel et al.[2] | 2 | 13 | F | Back | Desmin | + | WLE+CT | Recurrence; metastasis (P); death 2 years |
| Miki et al.[3] | 3 | 36 | M | Prostate | SMA | + | PE+CT | Recurrence; metastasis (P); alive 6 months |
| Rao et al.[4] | 4 | 48 | M | Intra-thoracic | Myo-D | * | E+CT+RT | Recurrence; metastasis (P+T); death 3 years |
| Present case | 5 | 15 | F | Buttock | Myogenin | − | CT+E+Intraoperative RT | 25 month event free |
| | 6 | 18 | M | Forearm | S-100 | − | E+CT+WLE | Recurrence; alive 6 months |
| | 7 | 30 | M | Extra-thoracic | Cytogenetics | −19/−22 | E+WLE+CT | 24 months event free |

IRMFS: Infantile rhabdomyofibrosarcoma; IHC: Immunohistochemistry; SMA: Smooth muscle actin; WLE: Wide local excision; CT: Chemotherapy; PE: Partial excision; E: Excision; RT: Radiotherapy; +: Positive; −: Negative; *Information not available/test not done
treatment is needed for these patients. Our patient did not receive radiotherapy considering complete resection at diagnosis and young age of the patient.

**Conclusion**

Infantile RMFS is a potentially under-diagnosed entity and should be kept in mind when a child is tested positive for IFS. Early recognition of this condition and a multidisciplinary approach can help in improving the prognosis of these patients.

**References**

1. Lundgren L, Angervall L, Stenman G, Kindblom LG. Infantile rhabdomyofibrosarcoma: A high-grade sarcoma distinguishable from infantile fibrosarcoma and rhabdomyosarcoma. Hum Pathol 1993;24:785-95.
2. Mentzel T, Mentzel HJ, Katenkamp D. Infantile rhabdomyofibrosarcoma. An aggressive tumor in the spectrum of spindle cell tumors in childhood. Pathologe 1996;17:296-300.
3. Miki H, Kobayashi S, Kushida Y, Sasaki M, Haba R, Hirakawa E, et al. A case of infantile rhabdomyofibrosarcoma with immunohistochemical, electronmicroscopical, and genetic analyses. Hum Pathol 1999;30:1519-22.
4. Rao SI, Uppin SG, Ratnakar KS, Sundaram C, Senthil RP. Infantile rhabdomyofibrosarcoma: A distinct variant or a missing link between fibrosarcoma and rhabdomyosarcoma? Indian J Cancer 2006;43:39-42.

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