Denouement and Comment

Infantile Fibrosarcoma

Histopathological examination revealed a spindle cell neoplasm with positive staining for the smooth muscle marker actin. Fluorescence in situ hybridization studies demonstrated an *ETV6* gene rearrangement resulting from a translocation between chromosomes 12 and 15. These findings were consistent with infantile fibrosarcoma. The patient was subsequently treated with neoadjuvant chemotherapy (4 cycles of vincristine sulfate, actinomycin D, and cyclophosphamide) followed by surgical excision.

Infantile fibrosarcoma is the second most common soft tissue sarcoma in children younger than 1 year (incidence of 5 per million infants).[1] The vast majority of affected individuals present before the age of 2 years, with approximately one-third of cases being noted at birth.[2,3] There is no significant sex or racial predilection.

The typical clinical presentation of infantile fibrosarcoma is a round, dome-shaped, skin-colored, erythematous, or erythematous to blue tumor that is solid and fixed to the deep tissue planes; surface telangiectasia, bleeding, and/or ulceration may be observed. While usually slow growing, rapid increases in size can also occur.[4] The extremities, especially the distal aspects, are the most frequent site of involvement (up to 70% of patients), with the head, neck, and trunk being affected less commonly.[5]

Laboratory abnormalities are not usually associated with infantile fibrosarcoma, although mild coagulopathy has been reported in a few cases.[6] In addition, while radiographic investigation may be useful in differentiating soft tissue tumors from vascular anomalies, infantile fibrosarcoma does not exhibit any distinctive features.[7] For these reasons, a definitive diagnosis requires collection of a tissue specimen for histopathological and, if possible, cytogenetic analysis.

Fibrosarcoma has a much more favorable prognosis in infants than adults.[4] Despite local recurrence rates between 17% and 43%, distant metastases are reported in less than 10% of cases and the estimated 5-year overall survival ranges from 84% to 93%.[2,3] The primary tumor site appears to be an important prognostic factor, as axial involvement is associated with a higher risk of metastasis and mortality compared with disease limited to acral sites.[6]

In general, surgical resection remains the mainstay of treatment for infantile fibrosarcoma. However, chemotherapy also has a clearly defined role in minimizing extensive mutilating surgery, both in the neoadjuvant setting and when used alone. The most commonly used regimen is vincristine, actinomycin D, and cyclophosphamide, although other combinations of chemotherapeutic agents have also been effective.[8,9] Ultimately, the specific management plan for each patient is best decided on by a multidisciplinary team.

As illustrated by our case, the challenge of infantile fibrosarcoma lies in the diagnosis because this entity can be easily confused with other conditions.[4,6,10,11] Deep infantile hemangioma, noninvoluting congenital hemangioma, Kaposiform hemangioendothelioma, lymphatic/venous malformation, rhabdomyosarcoma, metastatic neuroblastoma, and infantile fibromatosis can all present in an identical manner and must be considered in the differential diagnosis.

Certain clues can help the physician decide if biopsy may be necessary for a soft tissue mass. Characteristics that should raise the suspicion of cancer include (1) full form at birth; (2) persistent growth beyond 6 to 9 months of age; (3) firm consistency; and (4) fixation to underlying fascia.[12] Of course, there are always exceptions, but keeping these features in mind, pediatricians can play an important role in early recognition of childhood malignancies such as infantile fibrosarcoma.

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