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Solitary infantile myofibromatosis of the skull

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CASE REPORT

SOLITARY INFANTILE MYOFIBROMATOSIS OF THE SKULL

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ABSTRACT

This is the report of a 6 months old boy presenting with a firm, solitary mass on the temporal region, associated with lysis of local bone. Investigations lead to a diagnosis of infantile myofibromatosis (IM). Wide local excision was performed. At one year follow-up, no recurrence was noted.

KEY WORDS: *Infantile myofibromatosis. Soft tissue tumors. Lesions.*

INTRODUCTION

First described by Stout in 1954, infantile myofibromatosis is a myofibroblastic lesion of infancy and childhood. It usually presents as a solitary or multicentric firm, nodular lesion involving the soft tissue, bone or viscera. Three classes of the disease are recognized and carry prognostic implications: solitary myofibromatosis, multiple myofibromatosis without visceral lesions and multiple myofibromatosis with visceral involvement.[1] The generalized form of the disease is associated with a high rate of early mortality, especially if visceral structures are involved. These tumors are locally aggressive and do not metastasize.

The solitary form is the most common, presenting as firm, discrete nodules in the skin, bone, muscles, viscera and rarely the central nervous system. In the multicentric form, the number of lesions may vary from 2-100. Some of these lesions may be highly vascular and resemble hemangiomas.[2] In the generalized form, the most common locations are the lung, heart, gastrointestinal tract and pancreas, and rarely the central nervous system. Infants with generalized visceral lesions have the worst prognosis. Death in these cases often occurs due to cardiopulmonary or gastrointestinal complications.

The natural history of infantile myofibromatosis is characterized by a period of rapid growth, subsequent stabilization and spontaneous regression, this is true especially for the solitary type. Radiologic evaluation of infantile myofibromatosis is important in determining the extent of the disease and in monitoring disease progression.[3] Although there are reports of spontaneous regression of IM due to rarity of the condition, biopsy is usually required for diagnostic purpose. The biopsy shows a characteristic myofibroblastic pattern with a specific immunohistochemistry. Following is the report of infantile myofibromatosis involving the skull.

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CASE REPORT

A 6 month-old developmentally normal male infant was admitted with a 2 months history of a progressively enlarging swelling in right temporal region. There was no history of weight loss, trauma or swelling elsewhere. On examination, swelling was found just above the right ear in temporal region, measuring 2.5 cm x 2.5 cm. It was firm and non-tender. Rest of the examination was normal. Investigations revealed a normal complete blood count, electrolytes and coagulation profile. Urinary vinyl mandellic acid (VMA) level was also normal. X-ray of skull showed a soft tissue mass over the right temporal region and CT scan showed an expansile lytic lesion in the same region (Figure 1). No calcification was seen and brain parenchyma was normal. Ultrasound of the abdomen was within normal limits and bone scan showed no further lesion. Wide local excision was performed.

Histopathology revealed a lesion composed of interlacing fascicles of spindle cells with elongated nuclei. No significant nuclear polymorphism was appreciated. Scattered mitosis (2-3/hpf) was seen with no evidence of necrosis. Focal areas showed myxoid background. Immunohistochemical studies revealed that the tissue was positive for vimentin and was diagnosed as infantile myofibromatosis.

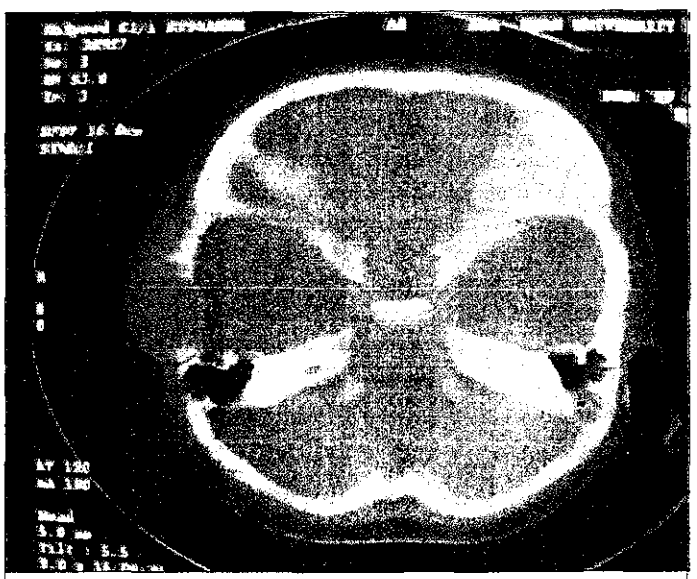

Figure 1: Infantile myofibromatosis of the skull : the tumor mass is seen eroding the right temporal bone.

The child made a quick postoperative recovery and was well on 8 weeks follow-up with no recurrence at the excised site and no new nodule elsewhere. The one year follow-up was uneventful.

Discussion

Fibromatosis represents a spectrum of non-neoplastic spindle cell tumors that may be locally aggressive but do not metastasize.[4] Infantile myofibromatosis (IM) is a rare mesenchymal tumor of infancy. To-date the etiology of IM is uncertain. All modes of inheritance, recessive, dominant and polygenic have been reported in literature.[3,5] The clinical presentation depends on the site of occurrence of the lesion. The solitary ones are usually subcutaneous and present with a lump on the involved site. This type was found to be the most common in a series of 170 cases reported by Wiswell.[3] Intracranial masses with extension into the skull bones and secondary brain compression have also been reported, defining them as locally aggressive tumors and a misdiagnosis of malignancy is frequently made with such a presentation.[6]

The X-ray usually shows a well-circumscribed lytic lesion with sclerotic margins. On CT scan, the tumor may have a variable appearance, simulating an aggressive lesion. MRI may show isointense lesions which enhance markedly after contrast injection.[2,7] In the present case also, there was a single well-circumscribed mass on the skull that appeared to be eroding the bone completely on CT scan; suggestive of an aggressive lesion and, therefore, a high index of suspicion is needed to differentiate it from more aggressive tumors like rhabdomyo sarcoma, histiocytosis and neuroblastoma.[3] The characteristic histological features differentiate it from other more sinister lesions and might be necessary to reach a final diagnosis, as was the situation in our case. Fascicles of spindle shaped cells which have staining characteristics intermediate between myoblasts and fibroblasts with central areas that are less well-differentiated and arranged in a hemangiopericytoma like pattern are focal in these tumors. Also the majority of these tumors show immunoreactivity for vimentin and smooth muscle desmin.[8]

Surgery is the mainstay of treatment, but spontaneous tumor regression has also been documented.[2,3] Hence surgical excision should be reserved for the confirmation of diagnosis or if vital structures are affected. Unfortunately, this is a rare tumor and diagnosis is mostly postoperative.

Multiple modalities other than surgery have been used like radiation and chemotherapy with varying success rates.[1] In this regard, the hypothesis presented by Toren in 1997, that masses appearing in the neonatal period with the histology of IM and with no life endangering features should be dealt conservatively, seems acceptable.[9]

Prognosis is excellent for solitary and multiple tumors without visceral involvement. Recurrence rate of 7-15% after regression or resection has been reported.[2] Clinical and histological features suggestive of recurrence are presentation at age greater than 5 years, extremity location of the disease, incomplete surgical resection, mitotic index of 5 or more/hpf and areas of necrosis and inflammation within the tumor.[10] Long-term follow-up is, therefore, required. The mortality rate from multicentric tumors with visceral involvement is upto 73%. IM should be kept in mind while dealing with newborns and infants presenting with solitary or multiple extraaxial subcutaneous nodules.

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