Multicentric neonatal myofibromatosis

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ABSTRACT
Infantile myofibromatosis is a rare, benign proliferative myofibroblastic tumor which occurs mostly in infants and young children. It presents as either solitary or multiple nodules arising from soft tissues, bones, or visceral organs. Accurate diagnosis and differentiation from more aggressive tumors are important because of the variations in the benign clinical course, conservative treatment, and possible spontaneous regression of infantile myofibromatosis. We present a case of multiple infantile myofibromatosis of soft tissues without involvement of visceral organs diagnosed at the neonatal age.

Key words: newborn, infantile myofibromatosis, congenital myofibromatosis.

INTRODUCTION
Infantile myofibromatosis is a mesenchymal neoplasm of infancy and early childhood. Although its incidence is extremely rare, it is the most common fibrous tumor of infancy, typically presenting as single or multicentric nodular masses of soft tissues, bones, or visceral organs.1 First described by Stout in 1954, various terms such as congenital multiple fibromatosis, diffuse congenital fibromatosis, multiple mesenchymal hamartomas, multiple vascular leiomyomas of the newborn, benign mesenchymomas, and generalized hamartomatosis have been used to describe the same entity. Chung and Enzinger used the term “infantile myofibromatosis” for the first time after careful review of 61 cases.2 We present a case of multicentric infantile myofibromatosis involving soft tissues, diagnosed at the neonatal age and we review clinical manifestations, pathologic and immunohistochemical features, and prognosis of this entity.

CASE REPORT
Our patient was a newborn female, delivered at 36 weeks gestation by vaginal delivery after an uncomplicated bichorial, biamniotic pregnancy (the somatic examination of her twin brother is without characteristic). There was no consanguinity between the parents. Birth weight was 2600 gr. Physical examination revealed a right axillary hemangiomatous appearance mass measuring 3 cm of diameter, the second mass was on the level of the neck measuring 4 cm of diameter (Fig. 1). These masses were painless with mobile consistency. There were also multiple subcutaneous firm nodules in abdominal and back wall fixed to the skin and two stellar plane lesions on the left thigh and ankle.

The laboratory assessment showed a low rate of the platelets (65000/mm3). On ultrasound, these masses had limited parietal development without blood flow. Abdominal and transfontanelar ultrasound and skeletal X ray radiographies were normal.

Excisional biopsy with histological examination of two cutaneous masses was performed. On macroscopic examination, the two masses were firm, well circumscribed and had a white-gray surface. They measured 4 and 1.5 cm in greatest diameter. Histologic examination showed a proliferation of ovoid or spindle shaped cells without cytologic atypia, arranged in short fascicles (Fig. 2). These tumors had a central hemangiopericytoma-like vascular pattern. The greatest mass had area of coagulative necrosis. Immunohistochemical study showed positivity of cells for smooth muscle actin (Fig. 3) and negativity for Desmin and PS100.

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The evolution was marked by progressive expansion of the masses, stabilization then regression of their sizes (6 months retreat). Axillary mass was necrosed and removed by surgery. The patient did not receive chemotherapy.

**DISCUSSION**

Infantile myofibromatosis is a rare mesenchymal tumor of infancy and early childhood, usually in the first 2 years of life (89%), although an adult counterpart has been described. Three clinical forms were described by Chung and Enzinger including the solitary form characterized by a single nodular lesion, multicentric form without visceral involvement like in our patient, and multicentric form with visceral involvement. These lesions are more common in males. They are most frequently located in the soft tissues (skin, muscle, and subcutaneous tissue) of the head and neck, followed by the trunk, then extremities. Among the patients with multicentric form, 35% involved not only multiple soft tissues, but also bone and visceral organs, typically the lung, heart, gastro intestinal tract, and rarely, the central nervous system. The exact etiology of infantile myofibromatosis is still obscure. Most cases are sporadic and familial aggregation has been observed in some cases, but the exact pattern of inheritance is unknown. Some authors suggested an autosomal recessive inheritance, while others suggested an autosomal dominant inheritance with variable penetration. Differential diagnosis can be difficult based solely on clinical and radiological observations; therefore, excisional biopsy and histological examinations are required to make the precise diagnosis as in our case. The use of fine-needle aspiration biopsy has been described but is not well established. Microscopically lesions are characterized by a central hemangiopericytoma-like vascular proliferation surrounded by fascicles of spindle-shaped cells with myofibroblastic features. Myofibroblasts are mesenchymal cells with both features of smooth muscle cells and fibroblasts. Immuno staining for smooth muscle actin and vimentin, with negative staining for desmin and S-100 protein support the myofibroblastic differentiation of infantile myofibromatosis.

Differential diagnosis includes the group of pediatric sarcoma, neurofibromatosis, desmoid tumors, fibrous hamartoma of infancy, nodular fasciitis, hyaline juvenile fibromatosis and juvenile haemangioma. The prognosis of infantile myofibromatosis depends on the distribution of lesions. Solitary or multicentric forms of infantile myofibromatosis are often benign and sometimes regress spontaneously if only confined to skin or bone. In rare cases, solitary lesions may be locally aggressive with slow, continuous, destructive proliferation. Excision of solitary lesions is usually performed for diagnosis and is typically curative. The recurrence rate is low (10%) and usually successfully treated with re excision. Conservative “wait and see” approach is the treatment of choice for multicentric forms without life-endangering pressure on vital organs. Patients with visceral involvement usually have poor prognosis and high rates of early death. Several treatment modalities have been proposed in this forms, including low dose chemotherapy.

**CONCLUSION**

Infantile myofibromatosis should be considered in the differential diagnosis in any child who presents with either a solitary or multiple tumours, particularly those occurring in the first 2 years of life. Because this lesion can involve visceral organs, for patients presenting with solitary or multiple tumors a careful and complete evaluation is necessary.

**REFERENCES**


