ORIGINAL ARTICLE

# Infantile myofibromatosis: A series of 28 cases

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***Background:*** Infantile myofibromatosis (IM) is a rare disorder of fibroblastic/myofibroblastic proliferation in children.

***Objectives:*** We sought to document common and unusual characteristics of patients with IM.

***Methods:*** This was a retrospective study of 28 children diagnosed with histopathologically confirmed IM between 1992 and 2012. Epidemiologic, clinical, and treatment data were reviewed.

***Results:*** IM was more frequent in boys (60.8%). Skin lesions were congenital in 64.3% of cases. The solitary form accounted for 50% of cases. Most nodules were painless, arising in cutaneous or subcutaneous tissue. The multicentric form accounted for 39% of cases; the skin, subcutaneous tissue, or muscle was involved in 97.8% of cases, and the bones in 50% of cases. The generalized form had a mortality rate of 33% (one-third of cases). Multicentric and generalized forms regressed spontaneously; severe local complications were observed, and late recurrent nodules developed in a few cases.

***Limitations:*** The retrospective review and the ascertainment of patients (from the departments of obstetrics and pediatrics) may have introduced bias in the analysis of severity of the different forms of IM.

***Conclusion:*** The diagnosis of IM must be confirmed histopathologically because the clinical presentation can be misleading. The prognosis is usually good, although local morbidity can occur. The generalized and multicentric forms merit long-term follow-up. (J Am Acad Dermatol http://dx.doi.org/10.1016/j.jaad.2014.03.035.)

***Key words:*** good prognosis; infantile myofibromatosis; long-term follow-up; pediatrics; systemic complications; tumoral lesions.

Infantile myofibromatosis (IM), a rare disorder of fibroblastic/myofibroblastic proliferation, is one of the most common benign fibrous tumors of infancy.[1] The incidence is not known. Most IM lesions occur in neonates or infants <24 months of age, with few reports of adult onset.[2-6] Based on the involved cell, the myopericyte located in the vessel wall, and its adjacent stroma, IM is now considered to be part of a spectrum of tumors with perivascular myoid differentiation.[2,7] The term myopericytoma was recently proposed as a generic name for this group of tumors in adults.[6]

*Abbreviations used:*

CT: computed tomography
ECG: electrocardiography
IM: infantile myofibromatosis

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Funding sources: None.
Conflicts of interest: None declared.
Accepted for publication March 19, 2014.

Reprints not available from the authors.
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Published online May 31, 2014.
0190-9622/$36.00



http://dx.doi.org/10.1016/j.jaad.2014.03.035

IM usually presents as firm, flesh-colored or purple nodules in the skin and subcutaneous tissue.

Three clinical types are recognized. The most frequent is the solitary form, with a single cutaneous nodule. The multicentric form involves the skin, subcutaneous tissues, muscles, and bone. The course is generally benign, with no metastases and regression of the tumor over a period of 12 to 18 months. The generalized form is defined by visceral involvement in addition to skin lesions. The prognosis is often poor, with death caused by visceral dysfunction. Results of surgery and chemotherapy have been disappointing, but spontaneous regression may occur.

We report epidemiologic and clinical data from a series of 28 pediatric IM cases to characterize this rare and poorly understood disease and share our experience of its management.

**CAPSULE SUMMARY**

- Although it is rare, infantile myofibromatosis is one of the most common benign fibrous tumors of infancy.
- We observed 28 infants with infantile myofibromatosis whose prognosis was good in most cases. The multicentric forms frequently regressed spontaneously.
- While solitary infantile myofibromatosis may be cured surgically, multicentric/generalized forms require ongoing clinical evaluation.

## METHODS

The database of a large municipal hospital was searched for all cases of IM with clinical follow-up of $\geq 1$ year. A dermatopathologist (S.F.) reviewed the histopathologic diagnosis for each patient.

Recorded information included sex, age at diagnosis, family background, characteristics of the skin lesions, involvement of subcutaneous tissues, bone, and viscera (all patients had abdominal ultrasound imaging and chest and skeletal radiographs) and both treatment and outcome.

## RESULTS

The clinical data on 25 of the 28 IM cases, 14 solitary and 11 multicentric, are summarized in Table I. Three cases of generalized IM are reported separately. Clinical presentations are shown in Fig 1.

### Solitary form

None of 14 patients with the solitary form had a family history of IM. The mean age of onset was 3.1 years (range, 0-4 years), and the mean age at diagnosis was 3.8 years (range, 0-5 years). The lesions were infiltrated nodules in 12 patients and sclerodermoid in 2 patients. They were fixed in 11 of 14 and mobile in 3 of 14 cases. Six were blue to purple, and 1 was yellowish in color; telangiectasia was noted in 2 patients. Five nodules were located on the head and neck, 6 on the trunk, and 3 on the limbs. All were painless (Fig 1, *A* and *B*).

Ultrasonography performed in 9 of 14 cases before surgery revealed a round, well defined hypoechogenic lesion in the subcutaneous tissue. Treatment consisted of complete excision of the nodule; none recurred. One patient carried a chromosome 6q deletion associated with dysmorphic features and mental retardation.

### Multicentric form

Five of 11 patients with the multicentric form belonged to the same family (autosomal dominant inheritance). Nine cases were congenital. The mean age of onset was 27 days (range, 0-6 months), and the mean age at diagnosis was 2.9 months (range, 0-6 months). The number of lesions varied from 2 to 30 in each case. The nodules were either mobile or fixed, and some had a prominent vascular component clinically resembling a hemangioma. In 6 cases, some lesions appeared atrophic. All were painless. Nodules were found on every part of the body, but most frequently on the head, neck, and trunk (Fig 1, *C*).

Skeletal radiography revealed bone involvement in 6 of 11 patients, evidenced by well-defined lytic lesions without sclerotic margins. Bone erosion caused by extension of an overlying cranial nodule was seen in 2 cases. Abdominal, cardiac, and transfontanellar ultrasonography, electrocardiography (ECG), and cerebral and thoracoabdominal computed tomographic (CT) imaging were normal in all 11 patients.

The nodules regressed spontaneously after the initial diagnosis in 7 of 11 patients during the first 2 years of life, sometimes leaving residual calcification and atrophic scars. Three patients had local complications because of pressure on the surrounding organs: (1) on the brachial plexus, which was treated by partial surgical excision because of reduced mobility of the hand; (2) on the vertebral canal, which was asymptomatic and detected by CT imaging; and (3) on the larynx causing stridor, which regressed spontaneously. One patient had a recurrence in the thyroid gland several months after the initial lesions had resolved. The patient with