Soft Tissue Ankle Mass in a 15-month-old Girl

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History and Physical Examination

A 15-month old girl was brought by her parents for evaluation of a left ankle mass. The parents reported the mass was first noticed by family members approximately 2 months after the girl's birth. They believed the mass had not caused any pain to the patient, but that it had increased in size, enough to begin interfering with shoe wear. The patient was the product of a full-term, normal vaginal delivery and had had no other medical problems. Family history was negative for any neoplastic conditions.

On physical examination, the patient was an active 15-month-old girl in no apparent distress. An obvious mass was seen at the anteromedial aspect of her left ankle. No additional masses were visible or palpable. There was no palpable lymphadenopathy. The patient had full active and passive ROM of her lower extremity without visible discomfort. Distal pulses were present and

normal in the lower extremity. There was a soft, somewhat mobile mass, approximately 3 cm x 2 cm, over the medial aspect of left ankle. There were no overlying skin changes. The mass did not seem to be tender, and the patient appeared to have full, intact sensation throughout her foot and ankle.

Before presentation, the patient had MRI while under sedation (Figs. 1–3). The MR images showed a lesion in the subcutaneous tissue of the medial ankle.

Based on the history, physical examination, and imaging studies, what is the differential diagnosis at this point?

Imaging Interpretation

MR images of the left ankle showed a subcutaneous lesion in the area of concern that was primarily hyperintense on T1-weighted images (Fig. 1), with linear areas of hypointensity. The lesion was hypointense on T2-weighted images (Fig. 2). There was minimal enhancement of the lesion after administration of gadolinium (Fig. 3).

Differential Diagnosis

Lipoma
Infantile fibromatosis
Fibrous hamartoma of infancy
Lipoblastoma
Lipofibromatosis

An open biopsy was performed and biopsy specimens were evaluated histologically (Figs. 4–6).

Based on the history, physical examination, imaging studies, and histologic appearance, what is the diagnosis and recommended treatment?

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Fig. 1A–C (A) Sagittal, (B) axial, and (C) coronal view T1-weighted MR images show enhancement (arrows).

Fig. 2A–C (A) Sagittal, (B) axial, and (C) coronal view T2-weighted MR images show less enhancement (arrows).

Fig. 3A–C (A) Sagittal, (B) axial, and (C) coronal view contrast-enhanced MR images show minimal enhancement (arrows).

Fig. 4 Microscopic examination shows an admixture of mature adipose tissue and spindle cell component (Stain, hematoxylin and eosin; original magnification, $\times 4$).

Histology Interpretation

The biopsy revealed a white-yellow, homogeneous, fatty-appearing lesion. Microscopy revealed a mixture of mature adipose tissue and fibroblastic component (Fig. 4), with

collections of small, vacuolated cells near the interface between fibroblastic elements and mature adipocytes (Fig. 5). Immunohistochemistry showed positive staining for smooth muscle actin in the spindle cells (Fig. 6) and negative staining for S100, CD34, and desmin.

Diagnosis

Lipofibromatosis.

Discussion and Treatment

The diagnosis of lipofibromatosis was made by the characteristic pathology findings of collections of small vacuolated cells near the interface between the fibroblastic component and mature adipocytes. Lipofibromatosis is a rare pediatric tumor described by Fetsch et al. in 2000 [5]. Since then, several case reports have been published. Previously, the lesion had been designated as infantile fibromatosis of nondesmoid type. However, Fetsch et al. differentiated between infantile fibromatosis and lipofibromatosis by

Fig. 5 Higher-power microscopy shows the bland nature of the spindle cells (Stain, hematoxylin and eosin; original magnification, $\times 10$).

Fig. 6 Smooth muscle actin immunohistochemical stain confirms the myofibroblastic differentiation of the spindle cells (Original magnification, $\times 10$).

noting that the former had a more “sheet-like” pattern of growth and did not contain fat as an integral element. In addition, the anatomic location differs between the lesions, with fibromatosis more likely involving the head and neck [5]. Lipofibromatosis typically presents as an ill-defined, slowly growing, painless mass most commonly manifesting in the hands and feet. It rarely occurs in the thigh, trunk, or head, with a male to female prevalence of 2:1. It is described as occurring exclusively in children from infancy to early second decade [7].

MRI features of this lesion include high signal on T1 and T2 imaging that shows loss of signal with fat suppression, indicating that the lesion contains fat, which is consistent with this case [1, 2, 11, 12]. Contrast enhancement on MRI has been described as heterogenous, primarily in the internal septations. The lesion did not contain nonfatty nodular areas, as can be seen with a liposarcoma [1, 11].

On gross examination, the tumor displays yellowish or whitish-tan lesions with the fatty component clearly evident. Usually, the lesion measures 1 to 3 cm and rarely exceeds 5 cm [5].

The histologic features of lipofibromatosis are composed of alternating streaks of mature adipose tissue and a fibrous spindle cell component. Mitotic activity is low, and nuclear atypia is absent. Immunohistochemical staining is often positive for CD34, BCL2, S100 protein, actins, CD99, and epithelial membrane antigen but is variable and nonspecific, as seen in this case. The tumor has a high rate of recurrence when incompletely excised; however, it has no metastatic potential [7].

Initial pathologic interpretation of this case suggested the lesion resembled fibrous hamartoma of infancy attributable to the admixture of mature adipose tissue and fibroblastic component. However, the nodular primitive fibromyxoid component commonly seen in this entity was not present. Furthermore, fibrous hamartoma of infancy more commonly occurs in the axillary fold, upper arm, shoulder, thigh, groin, back, and forearm. Rarely does it occur in the hands and feet, as does lipofibromatosis [6].

Infantile fibromatosis was another entity in the differential diagnosis. This lesion commonly involves the muscles of the head, neck, and proximal extremities, similar to fibrous hamartoma of infancy. It is also a benign soft tissue tumor that presents during the first 2 years of life [3, 4, 8]. Histologic interpretation can be classified as diffuse or desmoid types. As noted above, the fatty component of infantile fibromatosis is not as prevalent as in lipofibromatosis [13].

Lipoblastoma is a lipomatous soft tissue tumor of infancy that commonly involves the extremities, similar to lipofibromatosis. Patients present with a slowly growing soft tissue mass that is well circumscribed in the subcutaneous tissue. Although a lipoblastoma has internal septations seen on MR images, it also contains areas of high signal on T1- and T2-weighted images. It also differs histologically from lipofibromatosis in its presence of immature adipose tissue and lobulation, occasional myxoid areas, and absence of a fibroblastic component [9, 10].

The MRI appearance of this lesion also was suggestive of a lipoma. A lipoma is typically hyperintense on T1-weighted and hypointense on T2-weighted images. There may be thin septations present in lipomas but typically are not as prevalent as with our patient. In addition, histologically, lipomas are comprised of lobules of mature adipocytes with thin septations but no fibroblastic component. The vacuolated cells seen in a lipoblastoma are not present in lipomas.

The primary treatment of lipofibromatosis is excision of the mass, as was performed in our patient. There is a low incidence of local recurrence; recurrence is typically the result of incomplete excision [7]. Although lipofibromatosis is a rare and easily treatable tumor, it should be considered in the clinical and radiographic differential of

benign, fatty tumors of the distal extremities of children. Histologic examination should confirm the diagnosis in most cases. Our patient underwent excision of the lesion without complication. Her wounds were completely healed at the 1-month postoperative visit. At the last clinical followup at 1 year, the patient had no pain and no discernible recurrence of her lesion.

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