Diffuse Lipomatosis of the Chest Wall: Report of a Neonatal Case

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Diffuse lipomatosis is a very rare condition, the nature of which, neoplastic or hamartomatous, has not yet been defined. We report the case of a small child with a right thoracic lesion of neonatal onset and extremely rapid growth, who successfully underwent complete surgical resection. The differential diagnoses and proper surgical treatment are discussed.


Lipomatosis is a rare condition consisting of diffuse, nonlocalized overgrowth of adipose tissue [1-3]; various forms are nosologically distinguished, differentiated by anatomic location, with overlapping features. Most lipomatoses affect middle-aged patients; multiple symmetric lipomatosis (Madelung disease or Launois-Bensaude syndrome), mediastinoabdominal lipomatosis, steroid lipomatosis, and adiposis dolorosa (Dercum disease).

In the pediatric age, the most frequently reported form is congenital infiltrating lipomatosis of the face, in which the adipose proliferation is accompanied by proliferation of fibrovascular tissue and nerves [4]. The Bannayan-Zonana syndrome (lipomatosis, angiomatosis, and macroencephaly) is a congenital hamartomatous disorder with autosomal dominant inheritance and variable expression. Lipomatosis of nerve can present at birth or in early childhood and is frequently associated with macrodactyly.

Diffuse lipomatosis (DL) usually occurs in patients under 2 years of age but may also occur in adolescents and adults. It usually affects the trunk or a limb, but also the head and neck. The adipose overgrowth involves subcutaneous tissue and muscle but not nerves. The DL tends to recur, often repeatedly over many years [5-7]. It may attain a large size, thus causing compression of vital structures or impaired function. Because of its rapid growth and large size, it can clinically simulate liposarcoma. On histologic grounds the lesion, which lacks a true capsule and can infiltrate striated muscles, is composed entirely of mature fat. No lipoblasts are detected, nor have cytologic abnormalities, pleomorphism, or atypia been reported. No specific karyotypic aberration has so far been described in DL.

A 1-month-old girl was referred to our institution for an upper right thoracic mass, which had recently been noted by her parents. The patient was in good clinical condition, showing no respiratory distress or other symptoms. Family and personal histories were unremarkable. Prenatal ultrasound imaging was negative. Ultrasound scan showed a solid mass in the right hemithorax with echogenicity similar to that of subcutaneous fat. Follow-up ultrasound scans revealed very rapid enlargement of the mass. At 3 months of age, magnetic resonance imaging showed a large mass (7.8 x 8.2 x 3.6 cm) in the right hemithorax, infiltrating the pectoralis muscles and in contact with the right mammary gland and the axillary vessels. On T1-weighted images, signal intensity was the same as that of subcutaneous fat (Fig 1), while on T2-weighted fat-suppressed images the signal was hypointense. These findings were typical of fat content, the extremely rapid growth of the lesion was of particular concern and prompted a tru-cut biopsy approach in order to rule out malignancy.

The biopsy was carried out with a 16G needle and produced 5 core biopsies; a part of the tissue was used for cytogenetic analysis. The histologic picture, identical in all of the biopsies, consisted entirely of mature fat. No lipoblasts or signs of malignancy of any histotype were found. This was in keeping with the cytogenetic analysis, which revealed no particular aberrations. Nevertheless, given the mostly subcutaneous site of the mass, a sampling error could not be ruled out.

As the mass continued to grow extremely rapidly, with progressive infiltration of the mammary gland, an attempt at complete removal was undertaken (Fig 2). The surgical approach involved an S-incision; the mass showed adhesions to the muscular fascia and infiltration...
of the posterior aspect of the pectoralis major muscle. A macroscopically radical resection was performed, while an attempt was made to preserve the bud of the mammary gland. The surgical specimen consisted of a huge mass (18 × 15 × 7.5 cm), weighing 622 g, adherent to a skin strip (8 × 0.5 to 0.9 cm). The specimen was extensively sampled and processed according to standard pathology procedures. On microscopic examination, the overall picture was that of a normal fat tissue overgrowth. Gross pathologic features and histology (Fig 3), in accordance with cytogenetic analysis, were consistent with the final diagnosis of DL.

Comment

Our case presented several diagnostic and managerial challenges. Fine-needle and tru-cut biopsies have been advocated in children because of the minimal invasiveness of the procedure; although an incisional biopsy of a soft tissue mass is considered the best choice in that it minimizes the risk of sampling error, reduces the diagnostic difficulties linked to the histological diversity of soft tissue neoplasms, provides enough fresh material for cytogenetic studies or electron microscopy.

In the present case, preoperative magnetic resonance imaging of the lesion indicated the following: (A) the adipose nature of the proliferation; and (b) the subcutaneous and intramuscular location, together with irregular delimitation.

The differential diagnosis, in this context, included lipoblastoma or lipoblastomatosis, liposarcoma (myxoid), and intramuscular lipoma; given the age of our patient, lipoblastoma (which, however, does not usually show such rapid growth), or its diffuse form (lipoblastomatosis), was the first entity considered. Lipoblastoma is a rare benign tumor almost exclusively...
Fig 4. Postoperative results. The radical surgical approach achieved good aesthetic results while sparing the mammary gland.

Affecting infants and young children (under 8 years of age). It consists of mature and immature adipose tissue in variable proportions. The more immature forms are made up of primitive mesenchymal cells, lipoblasts, and capillaries in a myxoid background. The more mature forms (more frequent in older children) are very much like lipoma. Lipoblastoma shows consistent abnormalities of chromosome 8q11-13, resulting in rearrangement of the PLAG1 gene.

Liposarcoma in children is extremely rare (mostly in the 10 to 15 year age group) and most often myxoid in type. In the most differentiated lesions, a population of small spindle cells and small lipoblasts (predominantly univacuolated) is set in a myxoid matrix containing numerous thin-walled capillaries. Diagnostic multivacuolated lipoblasts are sometimes only found in the peripheral subcapsular parts of the tumor. Cytogenetically, myxoid liposarcoma is characterized by the recurrent translocation t(12;16)(q13;p11) or rarely, by the variant t(12;22)(q13;q12).

Intramuscular lipoma is a variant of lipoma, most often poorly circumscribed and infiltrative. It usually occurs in adult patients as a slow-growing mass. The most common cytogenetic abnormalities are aberrations involving 12q13-15, loss of material from 13q, and aberrations involving 6p21-23.

In our case, the typical clinical settings (young age, diffuseness of the process, and rapid growth) were all clues to the diagnosis of DL, which was consistent with the microscopic picture (poorly circumscribed, fully mature adipose tissue) and the absence of cytogenetic aberrations.

Strict follow-up was warranted because of the frequency of relapse reported in the literature. After 1 year, the patient is in good clinical condition, with no signs of recurrence. Moreover, the radical surgical approach achieved good aesthetic results while sparing the mammary gland (Fig 4).

Although rare, DL should be considered in the differential diagnosis when a rapidly growing lipomatous mass is found in a neonate as early surgical treatment can be crucial to obtaining complete removal, thus reducing the risk of recurrence and its associated morbidity.

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References


Congenital Tracheobiliary Fistula in an Adolescent Patient

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Congenital tracheobiliary fistula is a rare malformation that allows communication between the respiratory system and hepatobiliary tract. We describe a male adolescent patient who was admitted with a destroyed lung caused by repetitive bile pneumonitis with a congenital tracheobiliary fistula. Left pneumonectomy was performed, and the fistula tract was successfully divided. (Ann Thorac Surg 2015;99:328–31) © 2015 by The Society of Thoracic Surgeons

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