US, CT and MRI findings in a case of diffuse lymphangiomatosis and cystic hygroma

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KEYWORDS

Lymphangiomatosis;
Cystic hygroma;
Lymphangiomatosis;
US findings;
CT findings;
MRI findings.

Abstract Lymphangiomatosis is a rare disease with multifocal lymphatic proliferation that typically occurs during childhood and involves multiple parenchymal organs including the lung, liver, spleen, bone, and skin. Lymphangiomatosis may occur synchronously or metachronously with cystic hygroma of the neck. We present US, CT, and MRI findings of cystic hygroma of the neck associated with generalized lymphangiomatosis affecting bones and spleen in a 2-year-old girl.

Sommario La linfangiomatosi è una rara malattia caratterizzata da proliferazione multifocale del sistema linfatico che colpisce tipicamente i soggetti giovani e può interessare molti organi parenchimali tra cui il polmone, il fegato, la milza, le ossa e la pelle. La linfangiomatosi si può associare in sincronia o successivamente con l'igroma cistico del collo. Qui di seguito sono riportati i reperti ecografici, TC e di RM, di una bimba di 2 anni con igroma cistico del collo associato con linangiomatosi generalizzata delle ossa e della milza.

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Introduction

Generalized cystic lymphangiomatosis is a syndrome in which several organs are involved. It is a rare congenital malformation of the lymphatics. The disease is thought to originate from persistence of dilated lymphatics in the

14–20th week of intrauterine development. Up to 65% of involved patients are infants and children [1].

Cystic hygroma is one of the most common causes of neck mass in infants and young children. According to some authors, cystic hygroma, lymphangioma and cavernous lymphangioma are all variants of the same congenital disorder of lymphatic development [2].

Lymphangiomatosis occurring synchronously or metachronously with cystic hygroma of the neck is described in rare case reports [3–5]. In this report, we describe the US, CT, and MRI findings of cystic hygroma of the neck

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associated with generalized lymphangiomatosis affecting bones and spleen in a 2-year-old girl.

Case report

A 2-year-old girl with prenatal diagnosis of a cystic mass of the neck was monitored conservatively. US examination of the mass after birth revealed a multiloculated cystic structure compatible with cystic hygroma. The laboratory tests were unremarkable.

However, since physical examination revealed hepatosplenomegaly she underwent abdominal US which showed mild liver enlargement and marked splenomegaly, the latter extending into the pelvis and characterized by the presence of multiple anechoic cysts (Fig. 1). Because of these findings, splenic lymphangiomatosis was considered. The patient was subsequently submitted to CT scanning which confirmed the presence of an enlarged spleen with multiple well defined hypodense lesions. Additionally, multiple well defined hypodense lytic lesions in the lower thoracic and lumbar vertebral bodies, ribs, bilateral iliac wings and acetabular roofs, sacrum, right ischial and pubic bones were also detected. These lesions were not causing cortical disruption in the involved bones (Fig. 2).

MRI was then performed for further characterization of the bone lesions. Humeri, thoracic and lumbar vertebral bodies, ribs, both iliac bones and pubic bone and both femora were involved. The lesions in the bones were hypointense on T1-weighted images (WI) and hyperintense on T2-weighted images (WI). These lesions were not enhanced after administration of contrast agent (Fig. 3). Additionally, a 7×3.5 cm sized multiloculated T1 hypo and T2 hyperintense lesion compatible with cystic hygroma was seen in the left posterior cervical region. The mass showed minimal peripheral contrast enhancement on the T1 contrast enhanced series.

We interpreted the case as diffuse lymphangiomatosis with associated cystic hygroma according to the radiological findings.

Complete excision of cystic hygroma and total splenectomy was performed due to increased risk of complications in splenic lymphangiomatosis (bleeding, rupture, consumptive coagulopathy, hypersplenism). Histopathologic analysis confirmed cystic hygroma in the neck mass and diffuse lymphangiomatosis in the splenectomy specimen (Fig. 4).

Fig. 1 Coronal sonogram shows multiple anechoic cysts in the splenic parenchyma.

Fig. 2 Contrast enhanced CT shows multiple non-enhanced well defined hypodense lesions. Some small round hypodense lesions in the vertebral body are also evident.

Discussion

Generalized cystic lymphangiomatosis was first described by Rodenberger in 1828. It is a term used for diffuse involvement of soft tissue, viscera (especially lungs and spleen) or bone by lymphangiomatous proliferation. Histologically, generalized cystic lymphangiomatosis is a benign malformation of the lymphatic vessels that is composed of endothelium-lined cystic spaces containing eosinophilic homogeneous material or chyle. On the basis of histologic analysis, three types of lymphangiomas have been described: simple lymphangiomas, cavernous lymphangiomas, and cystic hygromas. Histologic classification may be

Fig. 3 Coronal T2-WI shows enlarged spleen with multiple hyperintense cysts. In the left side of the neck, there is a multiloculated hyperintense lesion with multiple hypointense septal structures. Note also small hyperintense cystic foci in the thoracic and lumbar part of the spine, and in the ribs of the left hemithorax.

Fig. 4 In the cut section parallel to the long axis of a $12 \times 7 \times 5$ cm sized spleen, numerous cysts of varying sizes filled with gelatinous material were seen.

difficult because of the wide morphologic variation of potential lesions.

Splenic lymphangiomatosis is rare, and no large series of cases have been reported in the literature. Only a small number of case reports are available, predominantly in adults with isolated splenic lymphangiomatosis characterized by pain in the left upper quadrant and splenomegaly. Diffuse lymphangiomatosis with splenic involvement has been reported even less frequently than the focal form, and it was reported most frequently in children [6]. Splenic lymphangiomas tend to occur in subcapsular locations, reflecting the anatomic distribution of splenic lymphatics. Thin-walled subcapsular or parenchymal cysts have been described as well as global splenic enlargement by a diffusely infiltrating lesion. Mural or septal calcifications may be present [7].

True splenic cysts, pyogenic abscesses, parasitic cysts, infarction, peliosis, hemangioma, lymphoma, and cystic metastasis should be considered in the differential diagnosis for splenic lymphangiomas. In lymphangiomatosis, the bones are the most commonly affected sites. The disease was originally described as affecting a single bone or multiple contiguous bones with no visceral involvement [8]. However, both noncontiguous bony and visceral involvement have been described later [9]. The diagnosis of lymphangiomatosis can be made on the basis of a combination of clinical, radiological and histopathological findings [10].

The overlap between "lymphangiomatosis of the bone" and "generalized lymphangiomatosis" can be appreciated from the presented series. Histologically, both processes are characterized by a variable proportion of lymphangiomatous and hemangiomatous proliferation. This finding suggests that there is a spectrum of disease with primary soft tissue/visceral involvement at one end and primary bone involvement at the other [11].

Bone involvement consists of multiple lytic lesions most commonly in the skull, ribs, pelvis, femur, humerus, and vertebrae, but practically any bone can be affected. As

opposed to visceral lesions, bone lesions tend to remain stable and show no progression and can even regress [12].

Cystic hygromas are present as soft, doughy, compressible masses situated mostly in the posterior triangle of the neck. It is generally considered to be a hamartoma of the lymphatic system and not a neoplasm. Cystic hygromas, cavernous lymphangiomas, and capillary lymphangiomas are congenital lymphatic malformations. These malformations may occur because a portion of the lymphatic network fails to reestablish communication with the venous system and is sequestered early in embryogenesis. Early malformations involving the primitive jugular, subclavian and axillary sacs are believed to cause the formation of larger cystic hygromas. Cystic hygromas occur in soft areolar tissues with wide fascial planes.

The treatment of cystic hygromas is surgical resection. Lesions that extend over several anatomic areas are difficult to resect and are prone to recurrence. Such patients need close follow-up and may require multiple surgical procedures. Percutaneous sclerotherapy is gaining wider acceptance for more advanced lesions that infiltrate deep fascial planes.

CT and sonographic features in splenic lymphangiomatosis consist of multiple cysts of various sizes, ranging from a few millimeters to several centimeters in diameter. On sonograms, these cystic lesions appear as well defined hypoechoic masses with occasional internal septations and intralocular echogenic debris. On CT scans, lymphangiomas appear as single or multiple thin-walled low attenuation masses with sharp margins that are typically subcapsular in location. No significant contrast enhancement is typically seen [13].

On T1-weighted MR images, the cystic lesions appear hypointense compared to the surrounding viscera. However, high T1 signal intensity may result from internal bleeding or the presence of large amounts of intracystic proteinaceous content. T2-WI demonstrates multiloculated hyperintense areas that correspond to dilated lymphatics. The intervening septa appear as hypointense bands, corresponding to the presence of fibrous connective tissue.

Because of the high contrast resolution, MR imaging may prove useful in the detection of solid elements within the cystic lumen in the very rare cases in which malignant degeneration may be present [13].

In cystic lymphangiomatosis, MRI characterizes the lesions as endothelium-lined cysts by virtue of a very high signal on T2-WI and absence of any central enhancement. The disorder most commonly confused with cystic angiomatosis is Langerhans cell histiocytosis. Although the plain film findings may be very similar, CT and MRI findings are completely different. Unlike cystic lymphangiomatosis, histiocytic lesions will frequently show a more irregular contour with some evidence of bone destruction; periosteal reaction and soft tissue lesions may be seen.

Cystic hygromas are anechoic masses that may occasionally contain internal septations or internal debris. The classic appearance on CT is a sharply demarcated low attenuation mass with imperceptible walls. Large lesions may be multilobular and may displace neighboring structures. On MRI, cystic hygroma shows low signal on T1-WI and high signal on T2-WI. There is no perceptible wall or enhancement following administration of contrast agent [14].

In conclusion, we have described the radiological imaging findings in a rare case of generalized cystic lymphangiomas involving bones and spleen, associated with cystic hygroma in the neck. In the pathogenesis of lymphangiomatosis and cystic hygroma, there is an underlying failure in the development of the lymphatic system. The extent of this developmental failure can vary and explain the coexistence of cystic hygroma with diffuse lymphangiomas. It should be kept in mind that, although very rare, the viscera and bones should be searched for diffuse lymphangiomas in patients with cystic hygroma.

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