International Journal of Radiology Research
www.radiologyjournal.in
E-ISSN: 2663-4562, P-ISSN: 2663-4554
Received: 16-07-2024, Accepted: 15-08-2024, Published: 30-08-2024
Volume 6, Issue 2, 2024, Page No. 22-25

Generalized cystic lymphangiomatosis: A rare entity revisited

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Abstract
Generalized cystic lymphangiomatosis (GCL) is an exceedingly rare disorder characterized by widespread proliferation of lymphatic vessels throughout the body [1, 2]. This disorder is believed to be because of maldevelopment of the lymphatic system during intrauterine period [2].
Despite its rarity, GCL presents a diagnostic and therapeutic challenge due to its variable clinical presentation and radiological features. In this article, we present the case of a pediatric patient diagnosed with generalized cystic lymphangiomatosis, focusing on the radiologic findings observed during the diagnostic process [3]. A comprehensive review of the clinical manifestations, imaging findings, diagnostic approaches, and management strategies for GCL based on a synthesis of existing literature and our clinical experience [4].

Keywords: Generalised cystic lymphangiomatosis, lymph cysts, lymphatic anomaly, pediatric bone cysts, multifocal cysts

Introduction
A 5-years 10 months-old male child presented with complaints of abdominal pain for 2 days. Physical examination revealed splenomegaly. Patient was referred to our department for imaging diagnosis for ultrasound abdomen, CT and MRI abdomen-chest.

Case presentation
Past operative history of mediastinal cyst excision at the age of 4 days after birth was given. On ultrasound of chest done at the time of birth (scan done in outside hospital) showed a large anechoic mediastinal cystic lesion with multiple internal septations was seen (Fig 1). On chest radiograph at birth showed a large mediastinal lesion seen in mediastinum and left hemithorax (Fig 2).
On physical examination patient revealed splenomegaly. Rest of the abdomen was soft and non-tender.
Ultrasound abdomen was performed on PHILIPS AFFINITY 70 machine, which showed splenomegaly (measuring 14.8 cm) with multiple variable sized subcentimetric anechoic cysts throughout splenic parenchyma (Fig 3).
Followed by CT abdomen with chest was performed on PHILIPS INGENUITY 128 machine (Protocol: A preliminary AP topogram of the Thorax and abdomen was obtained. Volume scans were performed starting from apices of lungs down to the level of pubic symphysis employing 0.625 mm sections). The CT scan showed numerous hypodense cysts was present in the spleen (Fig 4D, E) and multifocal intraosseous lytic lesions were present in ribs, spine and humeri (Fig 4A, B, C).
MRI chest and abdomen was performed on PHILIPS INGENIA ELLITION 3 Tesla machine (Protocol: Multiplanar sequences of abdomen and chest in T1 (TR-10, TE-6.5), T2 (TR-2725, TE-4000), FLAIR (IR-1650 ms) and axial DWI (b value- 500, 1000) and corresponding ADC sequences were performed), it showed multiple T2W hyperintense cysts of varying sizes scattered throughout the parenchyma of spleen (Fig 5). Few of the cysts appear hyperintense on T1W images which is suggestive of haemorrhage within the cysts. The cysts showed no restricted diffusion on DWI (Fig 5D, E).
Bilateral humeri, bilateral scapula, bilateral ribs, within all the vertebral bodies, pelvic bones, proximal shafts of bilateral femurs, left side of mandible showed Multifocal varying sizes T2W hyperintense intraosseous cysts.
No cysts were revealed in MRI chest (Fig 5).
A single T2W hyperintense cyst with imperceptible wall was seen anterior to the pancreas.
No surgical procedure was considered in our patient.

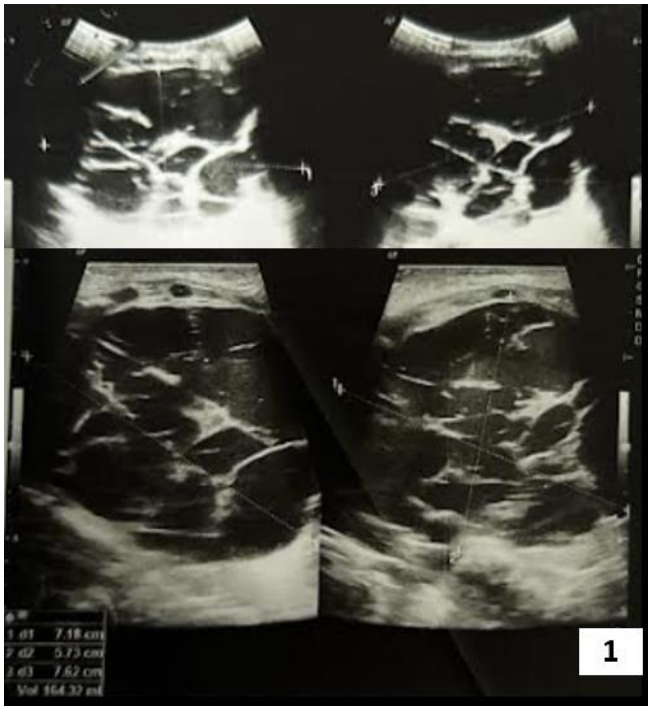

Fig 1: Ultrasound of the chest at the age of 4 days showing a large anechoic thin walled cystic lesion with multiple internal septations.

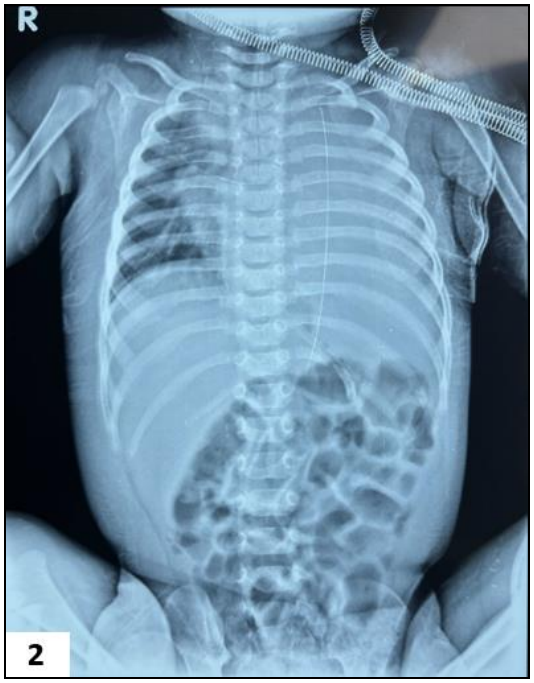

Fig 2: Chest radiograph of 4 days old neonate showing a homogenously radio-opaque lesion involving mediastinum and left hemithorax. It is silhouetting left lung with volume loss and mild mediastinal and tracheal shift to right.

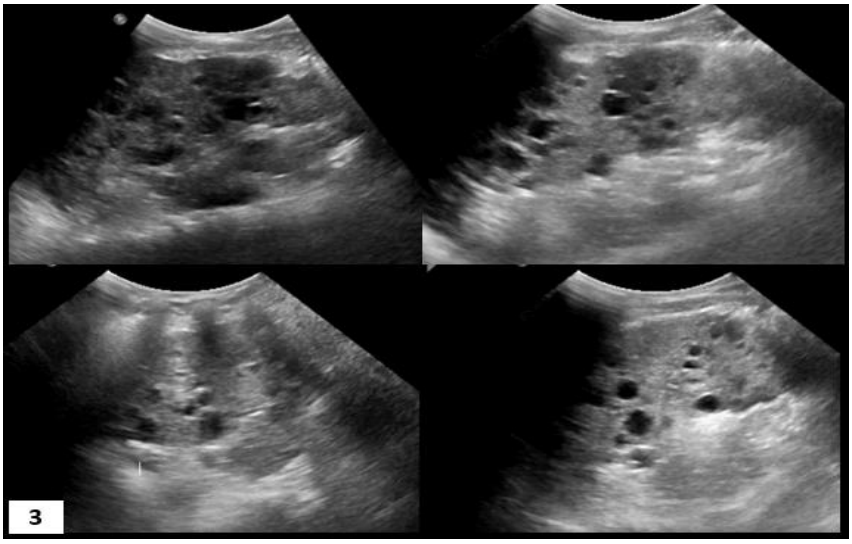

Fig 3: Ultrasound grey scale images of the spleen showing multiple variable sized anechoic cystic lesions throughout spleen.

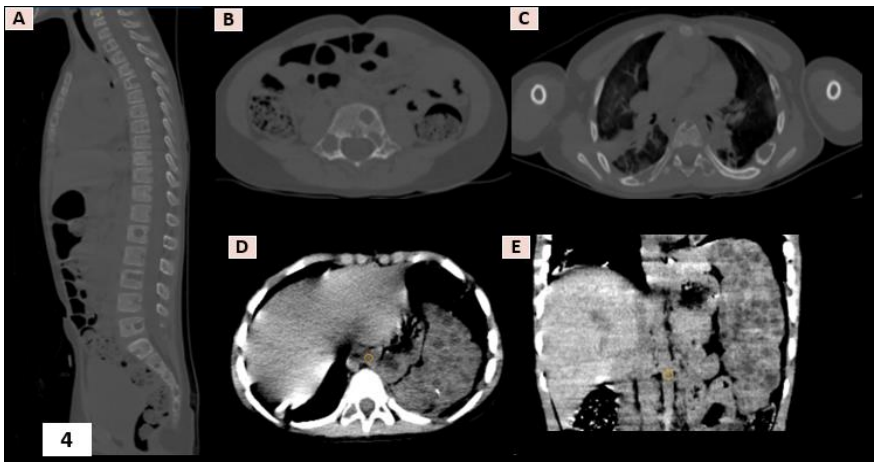

Fig 4: CT scan bone window of chest-abdomen sagittal (A) and axial (B, C) shows multiple lytic lesions in spinal vertebral bodies and ribs. Few patchy areas of consolidation due to aspiration pneumonia in bilateral visualised lungs (C). CT abdomen axial (D) and coronal (E) images showing enlarged spleen with multiple hypodense cysts within spleen.

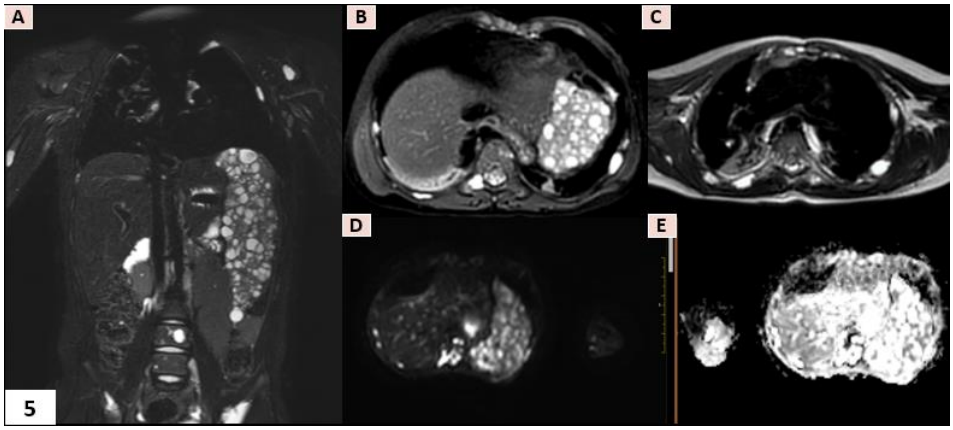

Fig 5: T2 weighted coronal (A) and axial (B, C) images of abdomen-chest (A) showing multiple hyperintense cysts involving spleen, vertebral bodies, ribs, visualised bilateral humeri and ulna. Axial DWI (D) and corresponding ADC images (E) at the level of spleen, the lesions show no restricted diffusion.

Discussion

GCL predominantly affects pediatric populations, although cases have been reported in adults as well [1, 2]. The exact incidence and prevalence of GCL remains unknown, likely due to under-recognition and under-reporting of this rare condition. It was first described by Rodenber in 1828. There is no gender predilection. It can occur sporadically, there have been occasional reports of familial clustering, suggesting a potential genetic predisposition. Nearly, 90% of cases are usually diagnosed within first 2 years of age, and it is rarely presented in adults. Intra-abdominal lymphangiomatosis occurs in less than 1% of all cases [1].

Pathologically these cystic areas are dilated chyle-filled spaces along the lymphatic system. It is seen to involve wide range of systems including the peritoneum, pleural cavity, spleen and musculoskeletal system [3]. Histologically it can be challenging to diagnose GCL as its morphology overlaps with other similar diseases such as generalised fibromatosis and diffuse haemangiomatosis. Therefore, cross-sectional imaging, such as computed tomography and MRI, play an important role in differentiating these pathologies [3, 4].

GCL poses diagnostic challenges due to its variable clinical presentation and radiologic features. Aim of this report is to discuss the radiological imaging features of GCL. In our case, the characteristic radiologic findings, including multiple cystic lesions in the spleen, peritoneum and bones with past history of operation for mediastinal cystic lesion were instrumental in establishing the diagnosis [2]. The differential diagnosis of cystic lesions in these organs may include other entities such as lymphangiomas, lymphangiectasia, lymphaticovenous malformation, cystic metastases, and congenital cystic lesions [3, 4]; however, the presence of diffuse involvement of multiple organs favours the diagnosis of generalized cystic lymphangiomatosis. Isolated presentation is shown to have better prognosis than multi-organ involvement. Rare presentation of subcutaneous nodules has been occasionally been described in the literature [1, 4].

Patients usually present with severe bone pain due to bony lesions. Other features due to extreme osteolysis cause pathological fractures and joints deformity in patients [1].

Ultrasound is the initial useful modality in imaging for evaluation of characteristics of the lesion like location, morphology, vascularity. Usually it is diagnosed as a multiloculated cystic lesion with septa [5]. Echogenicity of the cyst varies based on its fluid content which can be either blood, pus or chyle. Cross sectional imaging provide more clear information about the extension, size, and relationship to surrounding structures [1]. CT scan helps in detecting fatty components and calcifications within the cysts or along the septae. One differentiating feature of abdominal and superficial lymphangioma is that, lesions located in abdomen show linear, thin and small septal calcifications [1, 2, 5].

Histopathological examination remains the gold standard for definitive diagnosis, although imaging modalities such as CT and MRI play a crucial role in delineating the extent of disease and guiding treatment planning. Few case reports have shown that adult patients with systemic lymphangiomatosis have PIK3CA mutations [1].

Kaposiform lymphangiomatosis and Gorham-Stout disease (GSD) are other differential diagnosis for complex lymphatic anomalies. These conditions are differentiated mainly on imaging findings [1, 2].

In Kaposiform lymphangiomatosis, lesions usually show haemorrhagic content with consumptive coagulopathy. Intralesional hemorrhage is very rare in case of GCL [1, 2]. However, in our patient rare presentation of haemorrhagic debris in few of the cysts was seen.

Key feature of GSD is contiguous involvement with progressive osteolysis and cortical loss. In lymphangiomatosis, osteolytic lesions are non-contiguous, non-progressive and round lesions. Multiple focal bone involvement is noted in lymphangiomatosis. Vertebral involvement is rare in GSD as compared to lymphangiomatosis. GCL lesions rarely involve lumbosacral spine with extension into spinal canal and neural foramen [1]. In case of direct neural compression or cauda equina compression, it leads to severe lower back pain in the patient [3].

Treatment and Outcome: Management of GCL is primarily supportive, focusing on symptomatic relief and prevention of complications. Therapeutic options include dietary modifications, lymphatic drainage procedures, and sclerotherapy. Regular follow-up for monitoring of disease progression and response to treatment is indicated, and for early detection of complications such as infection or hemorrhage. Bisphosphonate medication has been investigated for preventing bone loss in skeletal lymphangiomatosis.

Conclusion

Generalized cystic lymphangiomatosis is a rare disorder characterized by the proliferation of lymphatic vessels in multiple organs with diverse clinical manifestations and radiological findings. Early recognition, high index of suspicion is required for timely diagnosis and appropriate management are essential for optimizing patient outcomes. Radiologic imaging plays a crucial role in the diagnosis and evaluation of this condition, with characteristic findings including multiple cystic lesions in the lungs, liver, and spleen. However, multidisciplinary collaboration involving radiologists, pathologists, and clinicians is essential for optimizing patient outcomes and advancing our understanding of this challenging condition. Further research is needed to elucidate the underlying pathophysiology and explore novel therapeutic interventions for this challenging condition.

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