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Incidentally Detected Lipoma of the Right Diaphragmatic Crus in a Young Adult With Pompe Disease: A Case Report

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ABSTRACT

Diaphragmatic lipomas are rare benign fat-containing lesions that are usually detected incidentally. Their recognition is important because they can mimic a fat-containing diaphragmatic hernia or, less commonly, an atypical lipomatous tumor/well-differentiated liposarcoma. We report a 25-year-old man with late-onset Pompe disease treated with alglucosidase alfa who underwent chest computed tomography (CT) for a 2-week productive cough. No pulmonary or airway abnormality was identified. CT demonstrated an incidental 15 x 10 x 7 mm, well-circumscribed, homogeneous lesion centered in the right diaphragmatic crus. The lesion showed macroscopic fat attenuation (mean, -112 Hounsfield units [HU]), without thick septa, a non-adipose nodule, enhancement, a diaphragmatic defect, or continuity with intra-abdominal fat. These findings supported an imaging diagnosis of right diaphragmatic crus lipoma. Given the lesion's small size, benign imaging appearance, and absence of related symptoms, conservative management was selected. This case illustrates the value of multiplanar CT in establishing the diaphragmatic origin of a fat-containing lesion and differentiating lipoma from Bochdalek hernia and suspicious lipomatous neoplasms. The coexistence of Pompe disease was considered incidental because no causal association has been reported.

KEYWORDS

Diaphragm; lipoma; Pompe disease; computed tomography; incidental finding

MAIN ARTICLE

INTRODUCTION

Primary tumors of the diaphragm are rare, and lipomas represent only a small subset. They are typically asymptomatic and discovered incidentally on thoracic or abdominal imaging [1-4]. The first published case is attributed to Clark in 1886; subsequent literature has consisted mainly of isolated reports and small series [1,2].

Computed tomography (CT) allows identification of macroscopic fat, confirmation of the lesion's diaphragmatic origin, and assessment of diaphragmatic continuity [1,3]. Thick or irregular septa, nodular non-adipose components, or enhancement should raise concern for an atypical lipomatous tumor/well-differentiated liposarcoma [5,6].

Pompe disease (glycogen storage disease type II) is an autosomal recessive lysosomal storage disorder caused by acid alpha-glucosidase deficiency. In late-onset Pompe disease, respiratory muscle involvement, particularly diaphragmatic weakness, may occur early and can be disproportionate to limb-girdle weakness [7-9]. No association between Pompe disease and diaphragmatic lipoma has been reported.

We report a small lipoma of the right diaphragmatic crus incidentally detected in a young adult with Pompe disease, emphasizing the CT features that support a benign diagnosis and distinguish this lesion from its principal imaging mimics.

CASE REPORT

A 25-year-old man with late-onset Pompe disease had been followed since childhood and was receiving enzyme replacement therapy with alglucosidase alfa. His phenotype included moderate proximal skeletal muscle weakness. He presented with a productive cough of 2 weeks' duration, without fever or other clinical features of lower respiratory tract infection. Oxygen saturation was 98% on room air, and pulmonary auscultation was normal. Because his neuromuscular disease increased the risk of respiratory complications, chest CT was performed to exclude an occult pulmonary cause rather than as routine surveillance.

Chest CT was acquired before and after intravenous administration of iodinated contrast material. The lungs and central airways were unremarkable, with no consolidation, ground-glass opacity, bronchiectasis, or pleural effusion. An oval, sharply marginated, homogeneous lesion measuring 15 x 10 x 7 mm was incidentally identified in the posterior right hemidiaphragm, centered on the right diaphragmatic crus (Figures 1-3). Its attenuation was identical to that of subcutaneous fat, with a mean value of -112 Hounsfield units (HU; range, -

167 to -74). No thick septa, non-adipose nodules, calcification, or soft-tissue component were present, and no enhancement was observed. Coronal and sagittal multiplanar reconstructions demonstrated preserved diaphragmatic continuity, without a posterolateral defect or communication with intra-abdominal fat. The lesion mildly protruded into the right thoracic cavity without significant mass effect on the adjacent lung.

The findings supported a presumed lipoma of the right diaphragmatic crus. Because the lesion was small, asymptomatic, and composed entirely of macroscopic fat without suspicious features, neither biopsy nor surgical resection was performed, and conservative management was selected. The cough resolved spontaneously within a few weeks and was considered unrelated to the diaphragmatic lesion.

DISCUSSION

A 2020 literature review summarized 34 previously reported cases of diaphragmatic lipoma and added a 35th case; most lesions were identified incidentally or at autopsy, and the posterolateral diaphragm was the most frequent location [1]. Clinical manifestations depend largely on size and mass effect. Small lesions are generally asymptomatic, whereas large or giant lipomas may cause chest discomfort, dyspnea, or compression of adjacent thoracic structures [1,2,4]. In the present case, the 15-mm lesion was unlikely to explain the patient's transient cough.

On CT, a simple lipoma appears as a well-defined, homogeneous lesion with attenuation equivalent to macroscopic fat. Multiplanar reformations help confirm that the lesion is centered within the diaphragm and demonstrate preserved muscle continuity [1,3]. Benign features include an entirely adipose composition, thin or absent septa, and no nodular soft-tissue component or enhancement. By contrast, thick or irregular septa, non-adipose nodules, enhancement, reduced fat content, large size, or invasion increase suspicion for an atypical lipomatous tumor/well-differentiated liposarcoma, although none of these features is individually diagnostic [5,6]. In our patient, the lesion was homogeneous and entirely fatty, centered in the crus, and associated with no diaphragmatic defect. Because no tissue sampling was performed, it is most accurately described as a presumed or imaging-diagnosed lipoma.

The principal differential diagnosis is a fat-containing Bochdalek hernia. A Bochdalek hernia is characterized by a posterolateral diaphragmatic defect and continuity of herniated fat or abdominal viscera with the abdominal cavity, whereas a diaphragmatic lipoma is enclosed within or attached to intact diaphragmatic tissue [1,3]. In our patient, multiplanar CT showed

neither a defect nor transdiaphragmatic continuity. A pleural or extrapleural lipoma may also abut the diaphragm, but its center lies outside the diaphragmatic muscle and it typically displaces rather than arises from the diaphragm. Finally, atypical lipomatous tumor/well-differentiated liposarcoma should be considered when a fat-containing mass is large, heterogeneous, enhancing, or contains thick septa or nodular non-adipose components [5,6]; none of these findings was present in this 15-mm lesion.

Respiratory muscle dysfunction is a major feature of late-onset Pompe disease. Diaphragmatic weakness may occur early and can be disproportionate to limb-girdle weakness [7-9]. Respiratory surveillance therefore relies primarily on clinical assessment, upright and supine spirometry, respiratory muscle strength testing, cough assessment, and evaluation for sleep-related hypoventilation [8,9]. CT is not routinely required solely to monitor diaphragmatic weakness; in this patient, it was obtained for an acute respiratory symptom. To our knowledge, no causal relationship between chronic diaphragmatic weakness in Pompe disease and diaphragmatic lipoma has been established. The coexistence of the two conditions was therefore considered incidental.

Because diaphragmatic lipomas are rare, no standardized size threshold or universally accepted imaging surveillance interval has been established. Management should be individualized: observation is reasonable for a small, asymptomatic lesion with unequivocally benign imaging features, whereas excision or further evaluation is appropriate for a symptomatic, enlarging, compressive, or indeterminate lesion [1,2,4]. In a young patient, the potential value of serial CT should be balanced against cumulative radiation exposure; MRI offers a non-ionizing alternative when additional characterization is clinically required [5,6]. In this case, the small size and typical CT appearance supported conservative management.

CONCLUSION

A diaphragmatic crus lipoma can be confidently suggested on CT when a homogeneous macroscopic fat-containing lesion is centered within an intact diaphragmatic crus and lacks thick septa, non-adipose nodules, or enhancement. Multiplanar reconstructions are particularly useful for excluding a diaphragmatic defect and continuity with intra-abdominal fat. Small, asymptomatic lesions with unequivocally benign imaging features may be managed conservatively, whereas interval growth, related symptoms, or diagnostic uncertainty should prompt further evaluation. In this patient, the coexistence of late-onset Pompe disease was considered incidental.

FIGURES:

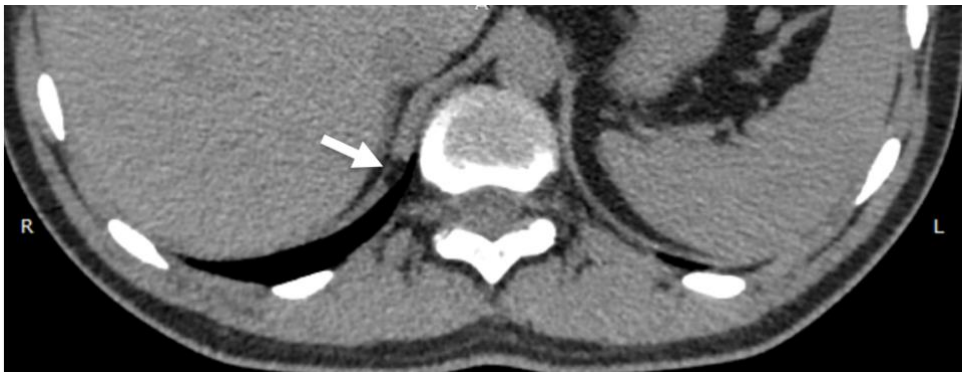

Figure 1. Axial CT image showing a small, well-circumscribed, homogeneous fat-attenuation lesion (white arrow) centered in the right diaphragmatic crus, without a visible soft-tissue component.

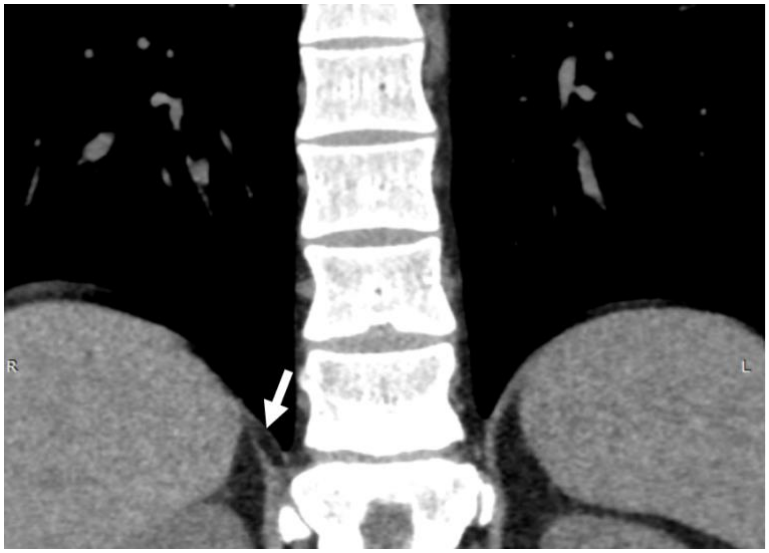

Figure 2. Coronal multiplanar CT reconstruction confirming the right posteromedial diaphragmatic location of the lesion (white arrow) and preserved diaphragmatic continuity.

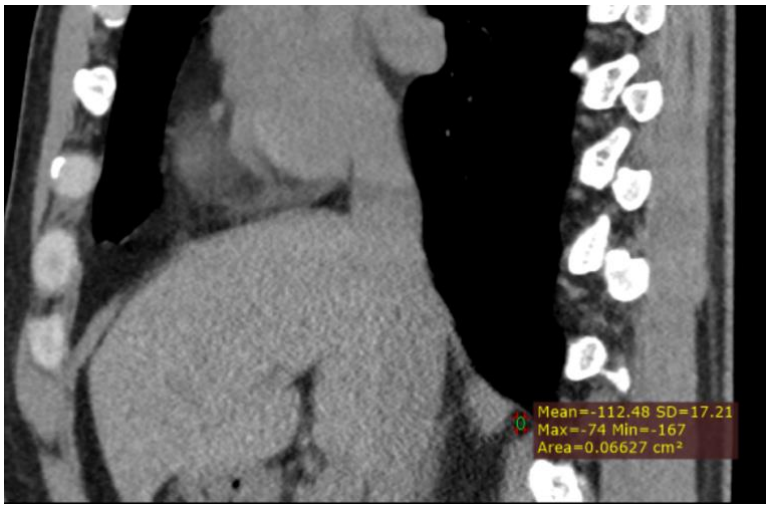

Figure 3. Sagittal CT image with a region-of-interest measurement demonstrating a mean attenuation of -112 Hounsfield units (HU; range, -167 to -74), confirming macroscopic fat.

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Patient Consent: Written informed consent for publication of this case and the accompanying imaging was obtained from the patient.

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