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Case Report

Amyloidosis, A Rare Cause of Recurrent Pleural Effusions. A Case Presentation

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Abstract

We present a case of a 72-year-old patient presenting with recurrent pleural effusion, extensive workup revealed amyloidosis to be the cause of this presentation.

This case highlights the challenges in diagnosing pleural effusions associated with amyloidosis, particularly in patients with complex medical histories. It underscores the importance of considering amyloidosis in cases of recurrent pleural effusions, especially when combined with systemic organ dysfunction.

Background

Amyloidosis is a rare disease characterized by the abnormal buildup of amyloid proteins in various tissues and organs throughout the body. This protein accumulation can lead to organ dysfunction and failure if left untreated. In the United States, the most common form is AL (light chain) amyloidosis, which accounts for the majority of cases. The prevalence of AL amyloidosis in the United States has been increasing over time. A study analyzing health insurance claims data found that the adjusted prevalence rose significantly from 20.1 cases per million in 2007 to 50.1 cases per million in 2015, representing an annual increase of 12% [1]. Based on these figures, it was estimated that at least 12,000 adults in the US were living with AL amyloidosis in 2015, with numbers likely continuing to rise.

Regarding incidence rates, studies have reported varying figures:

- Oneanalysis found adjusted incidence rates ranging from 10.8 to 15.2 cases per million person-years between 2007 and 2015 [1]
- Another study estimated the incidence to be approximately 9 cases per million person-years, suggesting around 2,200 new cases diagnosed annually in the US [1].
- Morerecent estimates indicate an incidence of 1 case per 100,000 person-years in Western countries, translating to approximately 1,275 to 3,200 new cases per year in the United States [2].

Amyloidosis is a highly variable disease, with its symptoms and severity depending on which organ system is primarily affected, be it cardiac, renal, gastrointestinal, or neurological. This variability often makes the diagnosis challenging and elusive.

A high level of clinical suspicion is crucial for diagnosis. Key diagnostic steps include:

- Comprehensive clinical examination and detailed patient history
- Serumandurine protein electrophoresis with immunofixation
- Serumfreelight chain analysis
- Tissuebiopsy with Congo red staining (the gold standard for diagnosis)
- Organ-specific imaging and biomarker evaluation

Case Presentation

Our patient, a 72-year-old male with a previous medical history of ischemic heart disease, congestive heart failure and atrial fibrillation, was referred to the clinic for the evaluation of a pleural effusion. During his initial work-up, right groin/inguinal lymphadenopathy was identified, leading to a surgical referral. While undergoing evaluation, he reported shortness of breath and was subsequently referred to Interventional Radiology (IR) for pleural drainage, as a CT scan confirmed the presence of a right-sided pleural effusion.

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The patient underwent thoracentesis on 9/19/2023, and 1.5 liters of pleural fluid was removed. Cytological analysis was negative, with the fluid being predominantly lymphocytic. Cultures revealed light growth of Cutibacterium acnes, which was considered likely a skin contaminant. Surgical excision of the inguinal lymph nodes revealed no malignancy.

The patient re-presented with recurrent pleural effusions occurring approximately every two weeks. A CT scan of the chest, abdomen, and pelvis was negative for any malignancy. We discussed treatment options with the patient, including pleural biopsy and pleurodesis. An empirical trial of Augmentin was initiated but did not resolve the effusion.

In December 2023, the patient consented to undergo pleural biopsy and pleurodesis, with biopsy results confirming amyloidosis. The patient was subsequently referred to oncology for further management. At his follow-up visit in August 2024, no recurrence of the pleural effusion was noted post-procedure. He reports occasional right-sided chest pain but is otherwise asymptomatic.

Differential Diagnosis

At the patient's initial presentation, our differential diagnosis was broad, encompassing both traditional transudative and exudative etiologies. Upon identifying lymphocyte-rich fluid, we refined our differential to focus on potential infectious and malignant causes.

Treatment

The patient's unilateral pleural effusion raised concerns for malignancy or pneumonia, despite a lack of typical pneumonia signs. After a week of Augmentin and multiple thoracenteses, lymphocyte-predominant fluid was repeatedly found with negative cytology and cultures. Following VATS with pleural and lung biopsy, amyloidosis was confirmed. The patient is now under oncology care. A CT abdomen suggested liver cirrhosis, but this was questioned due to negative ultrasound and normal LFTs. Follow-up chest CT showed significant improvement in pleural effusion, with mild pleural thickening and hilar/mediastinal lymphadenopathy. A newly identified 8 mm right lower lobe nodule was detected, while left lung nodules remained stable.

We also did workup for rheumatological causes, however the workup was negative.

Outcome and follow-up

Our chief diagnosis for this patient was pleural effusion caused by amyloidosis. We have discussed with the patient for possible EBUS for the 8mm lung nodule.

Discussion

Amyloidosis-related pleural effusion is driven by multiple pathophysiological mechanisms. Direct amyloid deposition within the pleura increases vascular permeability, resulting in fluid accumulation. Cardiac amyloidosis frequently leads to restrictive cardiomyopathy, precipitating heart failure and, consequently, transudative pleural effusions. In some cases, amyloid infiltration directly into the pleural space produces an exudative effusion. Rarely, hepatic involvement, particularly with hypoalbuminemia or nephrotic syndrome, can compound the effusion by promoting fluid retention.

Clinically, patients with amyloid-related pleural effusions typically present with nonspecific symptoms like dyspnea, chest discomfort, and cough, mirroring other causes of pleural effusion. However, pleural effusions in amyloidosis tend to be recurrent and refractory to standard interventions such as thoracentesis and diuretics, largely due to the persistent nature of amyloid infiltration in the pleura or ongoing systemic involvement.

Diagnostic evaluation hinges on a high index of clinical suspicion, appropriate imaging, and histopathological analysis. Thoracentesis commonly reveals an exudative effusion with a lymphocytic predominance, but cytological analysis is often negative for malignancy. Definitive diagnosis is obtained via pleural biopsy, demonstrating amyloid deposits confirmed by Congo red staining, with electron microscopy or mass spectrometry providing further confirmation. Imaging modalities, particularly chest CT, may reveal pleural thickening, recurrent effusions, and potential involvement of other organ systems, including the heart and liver, further aiding in diagnosis and guiding management.

Learning points

Pleural effusion is a rare but significant manifestation of amyloidosis. It often presents as recurrent, refractory effusions that can complicate the clinical management of these patients. Diagnosis relies on histopathological identification of amyloid deposits, and treatment options range from symptomatic relief through thoracentesis to more definitive procedures such as pleurodesis. The prognosis is generally poor in cases associated with cardiac amyloidosis, highlighting the importance of early diagnosis and targeted therapy.

Author Statements

Ethics Statements

We obtained informed consent from the patient.

References

1. (https://ojrd.biomedcentral.com/articles/10.1186/s13023-022-02414-6)

2. (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5965052/)

3. Falk RH, Comenzo RL, Skinner M. The systemic amyloidoses. New England Journal of Medicine. 1997; 337: 898-909.