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Case Report

Costal Amyloidosis: First Case in Literature

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Abstract

Amyloidosis is a heterogeneous group of diseases characterized by the extracellular deposition of amyloid proteins in various tissues and organs, which leads to significant morbidity and mortality. The most commonly affected organs include the kidneys, liver, spleen, heart, and nervous system, where the deposition of amyloid proteins disrupts normal function and causes a wide range of clinical manifestations. Involvement of osseous structures, particularly the ribs, is exceedingly rare and has not been extensively documented in the medical literature, making it a subject of significant clinical interest. This study aims to present a rare and fortuitously discovered case of costal amyloidosis in a patient who initially presented with a chest wall mass. The patient's presentation, diagnostic workup, and subsequent management are detailed to provide a comprehensive overview of this unusual manifestation of amyloidosis. Clinical evaluation revealed a firm, non-tender mass on the chest wall, prompting further investigation. Biological analyses included a series of laboratory tests to rule out common differential diagnoses and identify potential systemic involvement. Radiological evaluation comprised of advanced imaging techniques, including chest X-ray, computed tomography (CT), and magnetic resonance imaging (MRI), which highlighted the nature and extent of the osseous involvement. A biopsy of the mass was performed, and histopathological examination confirmed the diagnosis of amyloidosis through the identification of amyloid deposits using specific staining techniques. The case also examines the evolutionary aspects of the disease, discussing the progression and changes observed over time through regular follow-ups. Therapeutic strategies were implemented based on the latest guidelines and tailored to the patient's specific condition, including both pharmacological and surgical interventions. The short-term and long-term outcomes of these interventions are analyzed, highlighting the challenges and successes encountered during the treatment process. By presenting this case, we aim to shed light on the rare occurrence of costal amyloidosis, its diagnostic challenges, and therapeutic considerations. This case underscores the importance of considering amyloidosis in the differential diagnosis of chest wall masses and contributes to the broader understanding of the disease's clinical spectrum. Additionally, it emphasizes the necessity of a multidisciplinary approach in managing such rare and complex cases, ensuring comprehensive care and improved patient outcomes.

Keywords

Chest Wall, Amyloidosis, Rare Disease

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1. Introduction

Amyloidosis is a heterogeneous group of disorders characterized by the extracellular deposition of insoluble amyloid fibrils in various tissues, leading to the disruption of normal organ function. These amyloid proteins are abnormally folded and accumulate extracellularly, causing progressive damage and organ dysfunction. The disease can present in several forms, including systemic and localized amyloidosis, depending on the extent and location of amyloid deposits.

Commonly affected organs include the kidneys, where amyloid deposition can lead to nephrotic syndrome and renal failure; the liver, which can exhibit hepatomegaly and impaired liver function; and the heart, where amyloid infiltration may cause restrictive cardiomyopathy, heart failure, and arrhythmias. The involvement of the nervous system, particularly peripheral and autonomic neuropathy, is also frequently observed. While these manifestations are well-documented, amyloid deposition in osseous structures, specifically the ribs, is exceptionally rare and not extensively described in medical literature. This rarity poses unique challenges in both diagnosis and management, often leading to delayed recognition and treatment.

This case study explores an unusual presentation of amyloidosis fortuitously discovered during the evaluation of a patient presenting with a chest wall mass. The incidental finding of costal amyloidosis in this context underscores the necessity for a high index of suspicion and a thorough diagnostic approach when encountering atypical presentations of common diseases. The diagnostic process involved a comprehensive workup, including advanced imaging techniques and histopathological examination, to accurately identify the amyloid deposits and determine their systemic involvement.

Furthermore, the therapeutic approach to managing amyloidosis in such an atypical anatomical context requires careful consideration of the potential complications and the patient's overall health status. This case highlights the importance of a multidisciplinary team in addressing the complexities of the disease, integrating input from specialists in radiology, pathology, oncology, and thoracic surgery to devise an optimal treatment plan.

2. Methods

Patient Information: The patient is a 40-year-old male with no significant medical history who presented to the thoracic surgery department with a painless, non-inflammatory mass on the anterior chest wall, specifically involving the 6th and 7th ribs. Upon initial examination, the swelling was noted to be rigid and non-mobile, without any accompanying systemic symptoms such as weight loss, fever, or fatigue. Additionally, there were no signs of hepatomegaly, renal impairment, or lymphadenopathy, indicating an absence of other systemic involvements commonly associated with amyloidosis.

Clinical Findings: A thorough local examination of the chest wall mass was performed. The mass was well-defined and located over the 6th and 7th ribs. Radiological imaging, including X-rays and computed tomography (CT) scans, confirmed the involvement of the costal bones. Importantly, there was no evidence of adjacent soft tissue infiltration or lung parenchymal involvement, suggesting that the mass was localized to the bony structures. A biopsy of the subcostal mass was conducted, and histopathological analysis revealed extracellular deposits of an amorphous, weakly eosinophilic substance, which is characteristic of amyloid. Immunohistochemical studies were performed to identify the specific type of amyloid, confirming the presence of type AA amyloidosis localized to the costal region.

Diagnostic Assessment: To further assess the extent of the disease and rule out systemic involvement, comprehensive laboratory tests and a cardiac ultrasound were conducted. These investigations showed no evidence of systemic amyloidosis or cardiac abnormalities. The patient then underwent a detailed preoperative assessment to ensure he was a suitable candidate for surgical resection. This evaluation was crucial to anticipate and mitigate potential complications such as pleural invasion or vascular compromise during surgery.

Therapeutic Intervention: The patient underwent surgical excision of the costal mass under general anesthesia. The surgery aimed to achieve clear margins, and a 2 cm resection of the mass and adjacent pleura was performed. A pleural drain was placed to prevent postoperative complications such as fluid accumulation. The surgical procedure was successful, and the patient tolerated it well.

Follow-Up and Outcomes: Postoperatively, the patient's recovery was uneventful. The pleural drain was removed on the second postoperative day, and the patient was discharged on the fifth day. Follow-up at one year showed no signs of recurrence or progression of amyloidosis. The patient maintained preserved organ function and did not develop any systemic complications related to amyloidosis. Long-term monitoring is planned to continue assessing for potential recurrence or deposition of amyloid in other organs.

3. Results

The case presented a 40-year-old male patient with a rare and localized form of amyloidosis affecting the ribs. The clinical and radiological evaluations were pivotal in defining the extent and nature of the disease. The histopathological and immunohistochemical analyses confirmed the diagnosis of type AA amyloidosis, specifically localized to the costal bones, without systemic involvement.

The surgical intervention, involving resection of the mass with clear margins and adjunct pleurectomy, was successful. The patient's postoperative course was smooth, with rapid recovery and discharge within five days. The absence of recurrence or systemic progression at the one-year follow-up

highlighted the efficacy of the surgical approach for this rare presentation.

The successful management of this case underscores the importance of considering amyloidosis in the differential diagnosis of atypical chest wall masses and demonstrates the critical role of a multidisciplinary approach in the diagnosis, treatment, and long-term monitoring of patients with rare forms of amyloidosis. The patient's favorable outcomes emphasize the potential for effective localized treatment even in the context of a disease typically associated with systemic involvement.

4. Discussion

Amyloidosis represents a heterogeneous group of diseases characterized by the abnormal deposition of amyloid fibrils in various organs and tissues, disrupting normal physiological function [1, 2]. The pathogenesis of amyloidosis is complex and multifactorial, primarily involving the misfolding of soluble proteins into insoluble fibrils that aggregate extracellularly [1, 3]. These amyloid deposits can accumulate in different anatomical locations, including vascular walls, interstitial tissues, and parenchymal organs, leading to progressive organ dysfunction and potential systemic complications [1, 3, 4].

The diagnosis of amyloidosis relies heavily on biopsy, which remains pivotal in identifying the specific subtype of amyloid and guiding subsequent therapeutic strategies. Histopathological examination of affected tissues, often obtained through biopsy, reveals characteristic amyloid deposits that stain with Congo red and exhibit apple-green birefringence under polarized light microscopy [4-6]. Immunohistochemical staining further refines the diagnosis by identifying the specific type of amyloid protein involved, such as amyloid A (AA), immunoglobulin light chains (AL), or others [1, 7, 8]. This precise identification is crucial for determining the most appropriate treatment approach.

Costal involvement in amyloidosis is exceptionally rare and poses unique challenges in diagnosis and management. The ribs, particularly the costal bones, may be affected by amyloid deposition, leading to localized masses or structural changes that can mimic other pathological conditions such as neoplasms or inflammatory diseases [9, 10]. Diagnostic imaging, including computed tomography (CT) and magnetic resonance imaging (MRI), plays a crucial role in assessing the extent of bone involvement and evaluating potential complications such as pleural invasion or compression of nearby structures [11-13]. These imaging modalities provide detailed information that is essential for accurate diagnosis and effective treatment planning.

The management of costal amyloidosis hinges on the localization and severity of the disease. While there is no curative treatment for amyloidosis, current therapeutic strategies aim to mitigate symptoms, slow disease progression, and improve quality of life [13, 15, 16]. Systemic therapies targeting the underlying amyloidogenic process, such as chemotherapy in AL amyloidosis or anti-inflammatory agents in AA amyloidosis, may be indicated in cases of systemic involvement [1, 7]. However, in localized forms of amyloidosis, particularly those involving the ribs, surgical resection remains a viable option to achieve complete removal of amyloid deposits, alleviate local symptoms, and prevent further tissue damage [13, 18].

Surgical intervention in costal amyloidosis requires meticulous preoperative assessment to evaluate the extent of bone and soft tissue involvement. This ensures adequate surgical planning and minimizes the risk of complications during and after surgery [11, 12, 16, 17]. The goals of surgery include achieving clear resection margins and preventing local recurrence, while also considering the patient's overall health and potential comorbidities. Postoperative monitoring is essential to assess for recurrence or progression of amyloid deposition in other sites, as amyloidosis can exhibit variable clinical courses and patterns of organ involvement over time [11, 14]. Long-term follow-up is critical to ensure early detection and management of any new or recurring manifestations of the disease.

In conclusion, costal amyloidosis represents a rare manifestation of a complex disease process characterized by abnormal protein deposition. The diagnosis relies on histopathological examination and imaging studies, which together provide a comprehensive understanding of the extent and nature of the disease. Management strategies may include surgical resection in localized cases to optimize outcomes and preserve organ function. Continued research and clinical vigilance are crucial to advancing our understanding of amyloidosis and improving therapeutic outcomes for affected patients [17-19]. By sharing insights from rare cases such as this, we can enhance the collective knowledge and foster the development of more effective diagnostic and treatment modalities for amyloidosis.

5. Conclusion

Amyloidosis remains a complex and multifaceted disease with diverse clinical manifestations and variable prognostic outcomes. The disease's heterogeneity, resulting from different types of amyloid proteins and their deposition patterns, complicates both diagnosis and management. Costal involvement, as seen in this case, represents an exceedingly rare manifestation of amyloidosis. This rarity underscores the critical importance of comprehensive diagnostic evaluation, including detailed imaging and histopathological analysis, to accurately identify and localize amyloid deposits.

Tailored therapeutic interventions are essential to address the unique challenges posed by such atypical presentations. Surgical resection, as demonstrated in this case study, can offer favorable outcomes for patients with localized forms of amyloidosis. The success of the surgical approach in this case highlights the potential for achieving complete removal of

amyloid deposits, alleviating local symptoms, and preventing further tissue damage. This emphasizes the need for a multidisciplinary approach, involving thoracic surgeons, radiologists, pathologists, and other specialists, to ensure optimal patient care.

Continued research and clinical vigilance are paramount in managing this challenging condition. Understanding the underlying mechanisms of amyloid deposition, exploring new diagnostic techniques, and developing targeted therapies are essential steps toward improving patient outcomes. By documenting and sharing rare cases like this one, we contribute to the collective knowledge and foster advancements in the field of amyloidosis.

Ongoing long-term monitoring of patients with amyloidosis is crucial to detect any recurrence or progression of the disease. This vigilance helps in timely intervention and management, ultimately enhancing the quality of life for affected individuals. In summary, while amyloidosis remains a challenging condition to manage, the insights gained from rare presentations such as costal amyloidosis can inform and improve clinical practice, leading to better diagnostic and therapeutic strategies for all patients with this complex disease.

Abbreviations

CT Computed Tomography

Author Contributions

Mohammed Hachmi: Data Collection
Imane Belfkih: Supervision
Fatimezzahra Ammor: Supervision
Mouhssine Makloul: Supervision, Validation
Maidi Elmehdi: Supervision, Validation

Declarations

Ethics approval and consent to participate, Verbally consent obtained.

Data Availability Statement

All data are available and can be consult by contacting the corresponding author.

Conflicts of Interest

The authors declare no conflicts of interest.

References

[1] Desai SS, Rizzo MG, Rush AJ, Rosenberg AE, Al Maaieh M. Amyloidoma: A review and case report. Skeletal Radiol. 2021; 50(2): 437–444. https://doi.org/10.1007/s00256-020-03560-3

[2] Milani P, Basset M, Russo F, Foli A, Palladini G, Merlini G. The lung in amyloidosis. Eur Respir Rev. 2017; 26(145) https://doi.org/10.1183/16000617.0046-2017 Published 2017 Sep 6.

[3] Crain MA, Lakhani DA, Balar AB, Hogg JP, Adelanwa A, Hailemichael E. Tracheobronchial amyloidosis: A case report and review of literature. Radiol Case Rep. 2021; 16(9): 2399–2403.

[4] Kapoor R, Bhattacharyya T, Bahl A, Agarwal R, Bal A, Gulati A. Primary amyloidoma of lung treated with radiation: A rare case report. Lung India. 2014; 31(4): 404–406. https://doi.org/10.4103/0970-2113.142151

[5] Barešić M, Sreter KB, Brčić L, Hećimović A, Janevski Z, Anić B. Solitary pulmonary amyloidoma mimicking lung cancer on 18F-FDG PET-CT scan in systemic lupus erythematosus patient. Lupus. 2015; 24(14): 1546–1551. https://doi.org/10.1177/0961203315591025

[6] Brandelik SC, Heussel CP, Kauczor HU, Rocken C, Huber L, Basset M., et al. CT features in amyloidosis of the respiratory system - Comprehensive analysis in a tertiary referral center cohort. Eur J Radiol. 2020; 129.

[7] Yamada M, Takayanagi N, Yamakawa H, Ishiguiro T, Baba T, Shimizu Y, et al. Amyloidosis of the respiratory system: 16 patients with amyloidosis initially diagnosed ante mortem by pulmonologists. ERJ Open Res. 2020; 6(3): 00313–02019. https://doi.org/10.1183/23120541.00313-2019

[8] Kumar S, Kumar S, Ahmad A, Kumar A. Primary amyloidoma of chest wall–a rare condition. BMJ Case Rep. 2012; 2012 https://doi.org/10.1136/bcr.01.2012.5552 Published 2012 Mar 27

[9] Cordier JF, Loire R, Brune J. Amyloidosis of the lower respiratory tract. Clinical and pathologic features in a series of 21 patients. Chest. 1986; 90(6): 827–831. https://doi.org/10.1378/chest.90.6.827

[10] Iannettoni MD, Wiechmann RJ, Kirsh MM. Primary amyloidoma of the chest wall. Ann Thorac Surg. 1995; 59(6): 1583–1586. https://doi.org/10.1016/0003-4975(94)00907-O

[11] Matsumoto K, Ueno M, Matsuo Y, Kudo S, Horita K, Sakao Y. Primary solitary amyloidoma of the lung: Findings on CT and MRI. Eur Radiol. 1997; 7(4): 586–588. https://doi.org/10.1007/s003300050211

[12] Kim HY, Im JG, Song KS, Lee KS, Kim SJ, Kim JS, et al. Localized amyloidosis of the respiratory system: CT features. J Comput Assist Tomogr. 1999; 23(4): 627–631. https://doi.org/10.1097/00004728-199907000-00026

[13] Fierens J, Mees U, Vanbockrijck M, Hendrikx M. Amyloidoma of the chest wall: A rare entity. Interact Cardiovasc Thorac Surg. 2008; 7(6): 1194–1195. https://doi.org/10.1510/icvts.2008.178921

[14] Sipe JD, Cohen AS. Review: History of the amyloid fibril. J Struct Biol. 2000; 130(2-3): 88–98. https://doi.org/10.1006/jsbi.2000.4221

[15] Gillmore JD, Hawkins PN. Amyloidosis and the respiratory tract. Thorax. 1999; 54(5): 444–451. https://doi.org/10.1136/thx.54.5.444

[16] Lachmann HJ, Hawkins PN. Amyloidosis and the lung. Chron Respir Dis. 2006; 3(4): 203–214. https://doi.org/10.1177/1479972306070066

[17] Tabassomi M, Almasri E, Abdulhaq H. Unusual presentation of pulmonary plasmacytoma and amyloidoma in a patient with multiple myeloma. Am J Respir Crit. 2012; 185: A6649. https://doi.org/10.1164/ajrccm-conference.2012.185.1_MeetingAbstracts.A6649

[18] Safi S, Op den Winkel J, Schnabel PA, Hegenbart U, Neben K, Schneider T, et al. Extended resection of a plasmacytoma of bone and an amyloidoma of the chest wall. Ann Thorac Surg. 2013; 96(6): 2223–2225. https://doi.org/10.1016/j.athoracsur.2013.04.109

[19] Sravanthi N, Naveen G, Dinesh J. Primary pulmonary plasmacytoma: Unusual presentation with coexisting amyloidosis and bone metaplasia. Chest. 2014; 146(4): 661A. https://doi.org/10.1378/chest.1989411