

Giant Amyloid Goiter: Case Report

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Abstract

Amyloidosis refers to the abnormal deposition of amyloid proteins in organs and tissues. The disease can affect almost any organ in the body and when the thyroid gland is involved it results in a very rare condition known as amyloid goiter. An amyloid goiter is often characterized by progressive, rapidly growing, and painless bilateral thyroid enlargement without any major negative impact on its normal function as a gland. Symptoms include dysphagia, dyspnea and/or hoarseness. Here, we describe the case of a 32-year-old male patient who presented with a giant goiter and all the symptoms of upper airway obstruction, mainly voice change and breathing difficulty. He has been undergoing hemodialysis treatment as he suffered from chronic renal failure. Computer tomography confirmed the clinical exam and a total thyroidectomy was performed without complications.

Keywords: Amyloidosis; Amyloid Goiter; Thyroid gland; Renal failure; Thyroidectomy.

Introduction

Amyloidosis is characterized by insoluble, antiparallel β -pleated sheets of fibrils of proteins deposition in the extracellular space of different organs and tissues (1,2). It is called primary amyloidosis (PA) when it originates from plasma cells diseases such as multiple myeloma or other immunocyte dyscrasias while secondary amyloidosis (SA) is caused by a variety of degenerative, metabolic and inflammatory diseases (2). While the disease is usually systemic, localized cases were reported. In systemic forms, soluble fibril protein originating in the liver or bone marrow is delivered through blood plasma to different organs where it is accumulated as amyloid by a mysterious mechanism yet to be uncovered (12).

One extremely rare manifestation of amyloidosis is amyloid goiter. Although Eiselberg coined the term "amyloid goiter" in 1904, it's Beckman who first described the unusual disease entity in 1858 and recorded the first example of the condition (3,4). Amyloid goiter refers to the complete or near complete replacement of the thyroid parenchyma by amyloid, the process is rapid (from a few weeks to a couple of years) and gradual culminating in a firm, fibrous mass, and an enlarged neck (5,11).

The gradual increase in the thyroid gland might result in

tracheal deviation, upper airway obstruction, dyspnea, dysphagia, and changes in the voice owing to the compression exerted on the area. The condition's etiology remains unknown, but it is often associated with chronic inflammatory disorders (such as familiar Mediterranean fever, rheumatoid arthritis, ankylopoietic espodilitis, chronic osteomyelitis, tuberculosis, bronchiectasis) and, to a much lesser extent, with neoplastic processes and renal failure (6).

In the absence of an effective medical therapy, surgery or total thyroidectomy remains the treatment of choice for amyloid goiter (1,7). It is often required to proceed with fine-needle aspiration biopsy to rule out any malignancies (7). Amyloid goiter preoperative diagnosis often represents a challenging dilemma since thyroid function tests often turn out normal despite the infiltration (1). Differential diagnosis includes a malignant neoplasm such as anaplastic carcinoma or a non-Hodgkin's malignant lymphoma (11).

Case Report

In November 2021, a 32-year-old male patient presented to our hospital with a giant palpable goiter, complaining of dysphonia but no dysphagia. The goiter steadily developed and increased in size over the previous 3 years. The mass was heterogenous and has a bumpy external surface with the presence of fatty tissues identified as fatty metaplasia.

The patient's medical history revealed that he suffered chronic kidney failure and that he was undergoing hemodialysis treatment. The patient was diagnosed with high blood pressure 5 years ago. But no surgical interventions.

Preoperative computed tomography showed a bilateral enlargement of thyroid (the right lobe had dimensions of 41 x 52 x 113.4 mm whereas the left lobe was 53 x 54 x 128 mm) that extends upwards into the oropharynx descending to the thoracic cavity. The mass also exerted a severe compression of the adjacent neck structures that lead to dysphonia. Multiple bilateral cervical lymph nodes and air bubbles in the jugular vein were detected

but no bone lesions or damage was observed. It displaced the lateral vascular system which remained functional though.

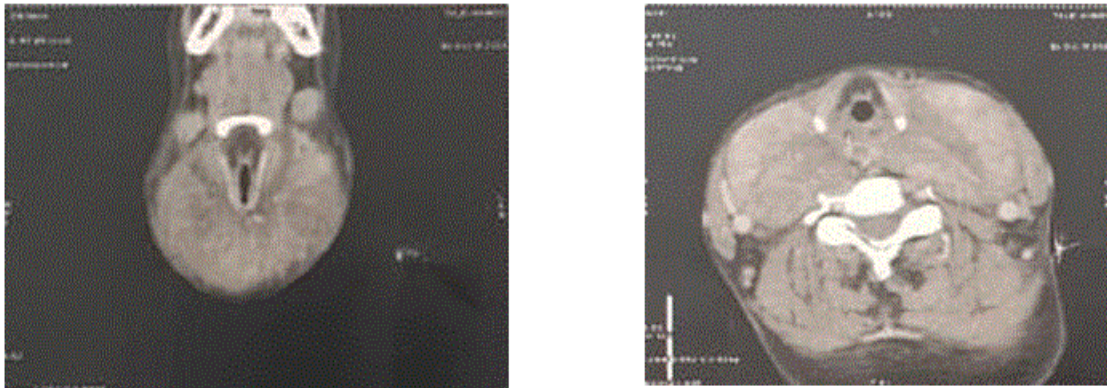


Figure 1-2: CT scan showing the huge goiter compressing the larynx



Figure 3: CT scan showing the goiter extending to the oropharynx

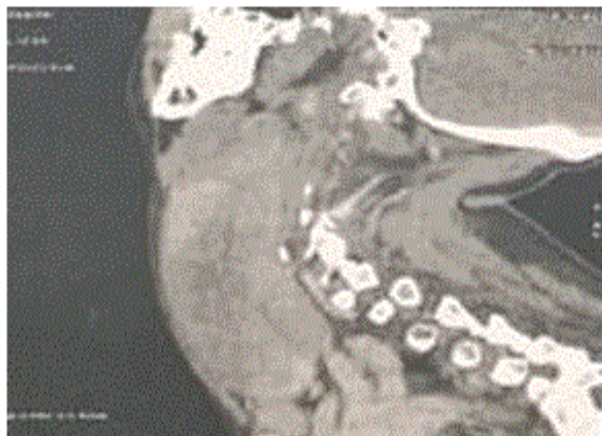


Figure 4: CT scan showing the goiter repels neighboring structures

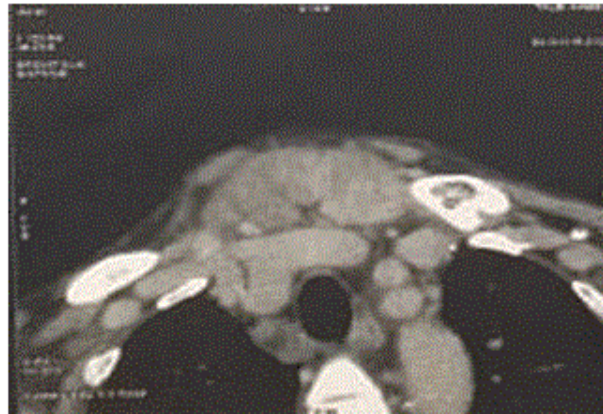


Figure 5: CT scan showing diffuse thyroid enlargement reaching to the thorax

Subsequently, the patient underwent total thyroidectomy without preoperative or postoperative complications and was discharged from hospital on day 4.

Histological findings confirmed the initial observations, and the patient was diagnosed with amyloid goiter associated to secondary amyloidosis.

Amyloidosis is the result of the accumulation of insoluble, fibrous amyloid proteins in the extracellular spaces of various organs and tissues. Amyloid fibrils deposition can affect almost any organ in the body and may have no clinical consequences as it may also have dire functional and structural changes in the involved organ (8,9). The disease, of unknown etiology, is extremely rare as it affects only eight people per million/year (9). One common technique to identify the presence of amyloid in tissues is to stain the structure with Congo red and by exposing it to cross polarized light and if the color turns apple-green birefringence it is indicative of an amyloid infiltration (9,13).

There are 30 different types of amyloidosis that englobe a variety of different conditions depending on the type of protein identified as the culprit, but we can distinguish three major forms: primary, secondary, and hereditary forms (13).

Primary amyloidosis involves an excessive presence of malformed "light chains" which is a form of amyloid proteins produced by dysfunctional plasma cells. These proteins gather in organs such as the heart, kidneys, nerves, and gastrointestinal system causing serious damage and leading up to the most common form of systemic amyloidosis (13).

Secondary amyloidosis or reactive amyloidosis also known as AA amyloidosis is linked to other chronic diseases (such as diabetes, tuberculosis, rheumatoid arthritis, and inflammatory bowel disease), certain types of cancer or simply aging. AA

amyloidosis derives its name from the "A protein", an amyloid protein that builds up in body organs such as the spleen, liver, kidneys, adrenal glands, and lymph nodes (1,13).

Hereditary amyloidosis is an even rarer condition that involves genes and is passed on to family members generation over generation. The protein responsible for hereditary amyloidosis may affect the heart and cause eye abnormalities. And in most cases the protein is known as transthyretin (TTR) hence the term ATTR amyloidosis which is another name for hereditary amyloidosis (13).

Both primary and secondary amyloidosis can cause an amyloid goiter, but the condition is most often associated with secondary amyloidosis. And our case confirms the view that chronic inflammation is the most common cause of secondary amyloidosis since our patient had kidney failure (1). Other causes include Hashimoto thyroiditis, Graves's disease, De Quervain and Riedle thyroiditis, thyroid cyst (13). The literature review revealed that most cases were associated with kidney disease (6).

Amyloid goiter must be differentiated from more common types of goiters, and the clinician has to consider the differential diagnosis of medullary carcinoma of the thyroid which can be ruled out by simply checking calcitonin levels that tend to be higher than normal in patients with medullary thyroid cancer. Other conditions with similar symptoms include multiple myeloma, rheumatoid arthritis, hyalinizing trabecular adenoma, solitary plasmacytoma, infections and familial Mediterranean fever. However, each of these lesions is easily recognizable to its unique histologic appearance. 8,13 It's also worth mentioning that thyroid lipomatosis, despite its rarity, can also lead to a massive goiter through the accumulation variable amounts of adipose tissue within the thyroid gland (4).

Preoperative diagnosis of a thyroid goiter is primarily achieved through fine-needle aspiration biopsy. The technique is also a reliable tool to rule out malignant thyroid tumor. A typical fine-needle aspiration cytology would demonstrate the presence of a substantial amount of adipose tissue and amyloid as an amorphous, eosinophilic, proteinaceous substance replacing and distorting normal tissue (1,7,13). Although the definitive diagnosis of an amyloid goiter is most often made post-operatively, FNA is always recommended as it is a safe and easy procedure (11).

The presence of proteins in the nodules produces images with high intensity on T1-weighted whereas fibrillar amyloid structures are associated with increased intensity on T2-weighted when the goiter is investigated with computerized tomography (CT) scans. MRI on the other hand shows an increased signal intensity on both T1 and T2 weighted images caused by fatty infiltration (4,6,13). Sonography reveals complex or hypoechoic masses (6,13).

Although people with familial Mediterranean fever are prescribed Colchicine to help prevent amyloid deposition and reports of patients who experienced improved thyroid function and decreased goiter size after being treated with dexamethasone, there aren't any effective treatment for amyloidosis. Total thyroidectomy remains the treatment of choice to an amyloid goiter with compressive symptoms or risks of malignancy development. Histological evaluation of the thyroid is often required for a definitive diagnosis (4,9,13).

Conclusion

A substantial increase in the size of the thyroid gland is a strong indicator of an amyloid goiter. Moreover, as it is the case with our patient, amyloid goiter should be suspected in all individuals suffering from chronic inflammatory diseases especially renal failure and undergoing hemodialysis treatment. Total thyroidectomy remains the treatment of choice and it can also help make a reliable and definitive postoperative diagnosis.

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