

## Case Report

# A rare presentation of amyloid goiter with renal amyloidosis in a young female

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### ABSTRACT

Amyloidosis is a rare systemic disorder caused by abnormal folding of normal soluble proteins leading to fibril formation in one or more body organs, systems or soft tissues. Amyloid goiter is characterized by deposits of amyloid protein in the thyroid tissue. Amyloid infiltration of thyroid gland with development of secondary goiter is rare. Here we report a case of 36-year-old female presented with progressive painless swelling over neck. Thyroid profile was normal. Ultrasound neck showed enlarged bilateral thyroid gland and isthmus. Fine needle aspiration cytology suggestive of subacute thyroiditis (granulomatous thyroiditis). Total thyroidectomy was done and biopsy sample revealed amyloid goiter.

**Keywords:** Amyloid goiter, Amyloidosis, Subacute thyroiditis

### INTRODUCTION

Amyloidosis is a group of diseases characterized by extracellular deposition of beta-sheet fibrils. These are broadly divided into localised and systemic amyloidosis. Localised amyloidosis is at the site of amyloid formation and the systemic amyloidosis is due to the circulation of amyloid deposits to various tissues and organs. The accumulation of amyloid deposits causes the progressive malfunction and eventual failure of the affected organ. The common sites of amyloid deposition include kidney, heart, liver, spleen, brain, nerves and skin. Amyloid goiter is defined as the presence of amyloid within the thyroid gland in such quantities that produces a clinically apparent enlargement of the thyroid gland. Amyloid goiter is very rare and constitutes about 0.04% of all the systemic amyloidosis.

Types of amyloidosis include AL amyloidosis (AL amyloidosis is due to deposition of protein derived from immunoglobulin light chain fragments), AA amyloidosis

(the fibrils are composed of fragments of the acute phase reactant serum amyloid A protein), wild-type transthyretin systemic amyloidosis (deposition of otherwise normal (wild-type) TTR, dialysis-related amyloidosis, hereditary amyloidosis. AL type is called as primary amyloidosis, AA type is called as secondary amyloidosis is caused by amyloid aggregates derived from predisposing conditions such as chronic inflammatory diseases, infections and haematological disorders.

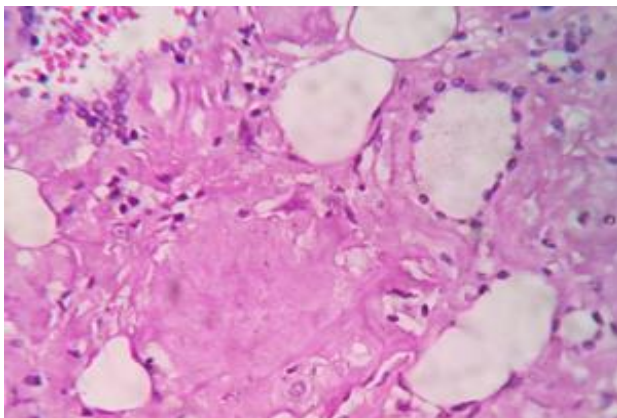
### CASE REPORT

A 36-year-old female who presented with swelling over the neck which is slowly progressive and painless since 6 months, history of difficulty in swallowing and hoarseness of voice since 5 months, history of breathing difficulty and amenorrhoea since 4 months. On physical examination, thyroid gland was diffusely enlarged, non-tender with no apparent nodularity and firm in consistency. Systemic examination was normal. Patient was an old case of biopsy proven renal amyloidosis and was on treatment with tablet

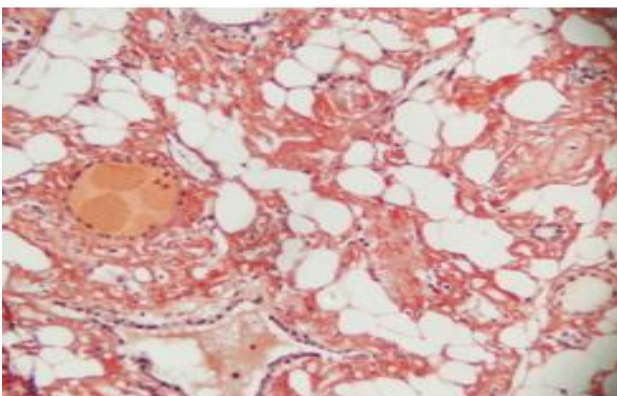
colchicine 0.5 mg once a day. There is no similar history in family members.

Laboratory investigations revealed mild anemia with haemoglobin of 10 gm/dl, leucocyte count and platelets were normal. Peripheral smear showed microcytic hypochromic anemia. Urea (40 mg/dl) and creatinine (1.6 mg/dl) were mildly elevated. Liver function tests were normal. Lipid profile was normal. 2D echo was normal. Thyroid profile was normal. Anti-TPO and thyroglobulin were negative. ANA profile was negative. Ultrasound neck showed bilateral thyroid gland and isthmus enlarged with right lobe of size of 2.2×3.6×4.3 centimetres with colloid nodule of size 7×5 mm and left lobe of 3.2×3.0×4.3 centimetres.

Fine needle aspiration cytology revealed follicular epithelial cells in clusters, micro follicles and scattered lymphocytes in both the lobes along with the few multinucleated giant cells and occasional clusters of epithelioid histiocytes with background of colloid suggestive of subacute thyroiditis (Granulomatous thyroiditis). Patient was taken up surgery and total thyroidectomy was done.

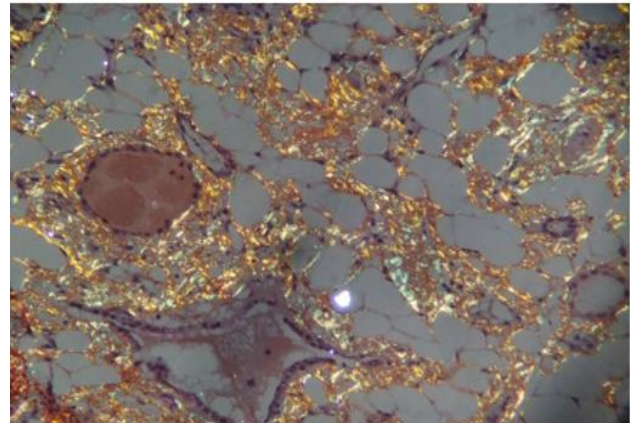


**Figure 1: Homogenous eosinophilic extracellular amyloid surrounding the thyroid follicles.**



**Figure 2: Congo red stain under light microscope showed amyloid that is stained red suggestive of amyloidosis.**

Histopathology report of the biopsy specimen showed homogenous eosinophilic extracellular amyloid surrounding the thyroid follicles and with the special stain of Congo red stain showed red colour staining of the diffuse fibrillary material under light microscopy and apple green birefringence of the diffuse fibrillary material under polarised microscopy suggestive of amyloid goiter (Figures 1-3). Patient was treated with tablet colchicine 0.5 mg twice a day, tablet thyroxine 100 mcg once a day and tablet ferrous ascorbate 100 mg once a day. Patient was on follow up and remained asymptomatic.



**Figure 3: Congo red stain under fluorescent microscope showed apple green birefringence suggestive of amyloidosis.**

## DISCUSSION

The term "amyloid," first introduced by Schleiden in 1838 to describe plant starch. Amyloid goiter is defined as the presence of amyloid within the thyroid gland in such quantities as to produce a clinically apparent enlargement of the gland. Amyloid accumulates extracellularly in the thyroid parenchyma and disrupts the normal follicular patterns. This was first described in 1858 by Beckman. It may be associated with either primary or secondary amyloidosis. The majority of cases are associated with secondary amyloidosis, which is also called reactive amyloidosis.

Jacobovic et al reported a case of 40-year-old male admitted with goiter and upper airway obstructive symptoms with normal thyroid function and concluded that amyloid goiter had no major impact on thyroid function even in cases where a greater amount of amyloid replaces the normal thyroid parenchyma.

Eisalari et al reported a case of 53-year-old male presented with painless neck swelling over neck since 2 years without obstructive symptoms, surgery was done and biopsy revealed amyloidosis of thyroid gland and he concluded that amyloid goiter is a rare entity and high index of suspicion is required in patients with an enlarging thyroid gland to not to miss the diagnosis.

Juhlin et al reported a case of 59-year-old male who presented with a slowly progressive enlargement of both thyroid lobes, eventually causing significant tracheal displacement, dysphagia and dyspnea. Later on evaluation patient has amyloidosis. He concluded that patients with progressive thyroid enlargement should have suspicion of amyloidosis.

In our case patient had a past history of biopsy proven renal Amyloidosis now presented with progressive thyroid enlargement. Total thyroidectomy was done and the biopsy sample revealed amyloid goiter. This is very rare case report in which amyloid goiter present along with the renal amyloidosis in the same patient

## CONCLUSION

Here we report a case of 36-year-old female presented with painless progressive thyroid swelling and on further evaluation patient amyloid goiter. we report this case due to rarity of the condition. Here we conclude that patients presenting with progressive thyroid enlargement with normal thyroid function tests should be suspected highly for amyloidosis.

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