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Case Report

Surgical Treatment of Pendred Syndrome with Thyroid Nodule: A Case Report

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ABSTRACT

Introduction and importance: Pendred syndrome (PS) is a genetic disease inherited in an autosomal recessive pattern. In PS patients, the ability of thyroid gland to accumulate iodide in the follicle lumen is disrupted, causing insufficient thyroid hormone synthesis and goiter to occur as compensation. However, this symptom does not often appear, as goiter is only detected in 30–75% of PS patients. **Presentation of case**: A 33-year-old Buginese man complained of a painless mass in the neck. In the past 3 months, the mass has shown rapid growth, followed by symptoms of agitation, irritability, and hyperhidrosis. The patient was previously diagnosed with nodular goiter and was taking Levothyroxine. Furthermore, there was a history of the same disease in the patient's grandmother and younger sibling. Physical examination showed grade III goiter (WHO criteria) with hard consistency and no palpable nodules. Laboratory tests showed routine blood tests, kidney and liver function in normal limits, 1.14 ng/dL FT4, free triiodothyronine level of 6.07 pg/dL, and 2,401 mIU/L TSH level. Chest X-ray results showed tracheal narrowing at the level of the superior thoracic aperture. Similarly, thyroid USG showed that the size of the right lobe was enlarged and anechoic. CT (Computerized Tomography) scan of the cervix showed diffuse enlargement in each lobe and the patient

was diagnosed with PS. Right isthmolobectomy and thyroid biopsy were carried out and pathological anatomical examination results showed thyroid follicular nodular disease with cystic degeneration. After surgery, thyroxine hormone replacement therapy was continued.
Discussion: Surgical intervention strategies may be employed to inhibit the advancement of disease and mitigate thyroid enlargement, consequently decreasing the likelihood of obstruction and malignancy.
Conclusions: In cases of PS with large thyroid nodules pressing on the trachea, isthmolobectomy was carried out to obtain good results and no recurrence.

1. INTRODUCTION

Pendred syndrome (PS) is an inherited disease with an autosomal recessive (AR) pattern associated with sensorineural deafness, inner ear malformations, and goiter with or without hypothyroidism.[1,2] In PS patients, goiter occurs due to the disruption to the ability of thyroid gland to accumulate iodide in the follicle lumen, causing goiter to occur as compensation and insufficient thyroid hormone synthesis.[3] However, these symptoms does not often appear and goiter was only detected in 30–75% of PS patients. This condition becomes apparent in late childhood or early puberty or even only detected in adulthood.[4,5]

2. CASE PRESENTATION

A 33-year-old Buginese man visited Hasanuddin University General Hospital because of a painless mass in the neck, which was incidentally identified 10 years earlier. The mass had recently shown rapid growth and he reported symptoms of hyperhidrosis, irritability, agitation, and discomfort when swallowing. Before being admitted to the institution, he had been diagnosed with nodular goiter by a doctor at another hospital and had been taking Levothyroxine periodically for several years. There was a report of hearing loss and difficulty learning language in childhood, but clinical examination showed normal physical and mental development was normal. There was a history of the same disease in the patient grandmother and younger sibling.

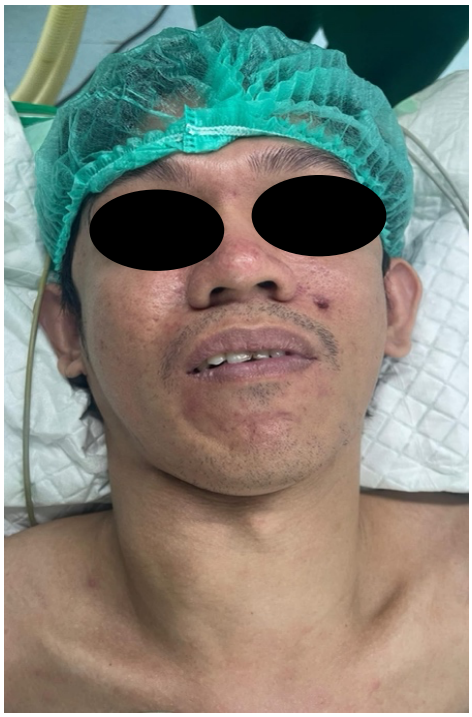

Figure 1. The right thyroid nodule in the patient

Physical examination showed a visible goiter of grade III according to the WHO criteria. Thyroid was hard and enlarged, with the border was clear and no palpable nodules, as shown in Figure 1. Laboratory tests showed routine blood tests, normal renal and liver function, free T4 of 1.14 ng/dL (normal range, 0.93–1.7 ng/dL), free triiodothyronine level of 6.07 pg/dL (normal range, 130–450 pg/dL) and thyroid-stimulating hormone level of 2,401 mIU/L (0.5–5.0 mIU/L). Tracheal narrowing at the level of the superior thoracic aperture was seen through Chest X-ray, and thyroid ultrasound showed an enlarged right lobe, anechoic, solid intraocular components, normal isthmus, and right thyroid. In addition, there was no enlargement of the paracervical lymph nodes. Manifestations of cervical CT scan include thickening of the isthmus and diffuse enlargement of each lobe of thyroid gland, as shown in Figure 2. Numerous oval-shaped, low-density, and slightly elevated nodules were identified in thyroid.

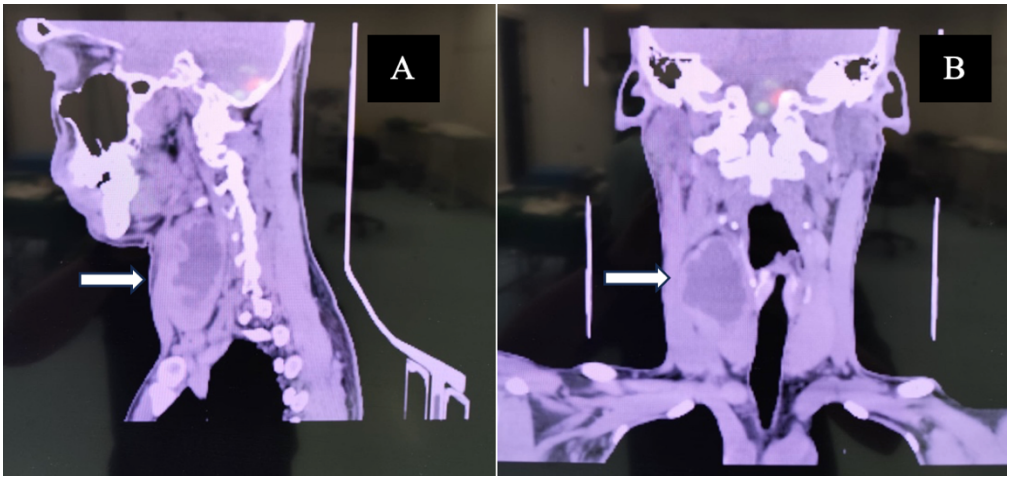

Figure 2. The right thyroid gland is seen to be enlarged to 70x42 mm (A), nodular, accompanied by necrosis pressing the trachea to the left (B).

Surgical procedure was carried out under general anesthesia and right isthmolobectomy was performed, as shown in Figure 3. The results of the anatomical pathology examination showed thyroid follicular nodular disease with cystic degeneration. Postoperatively, the patient was given permanent hormone replacement therapy with thyroxine.

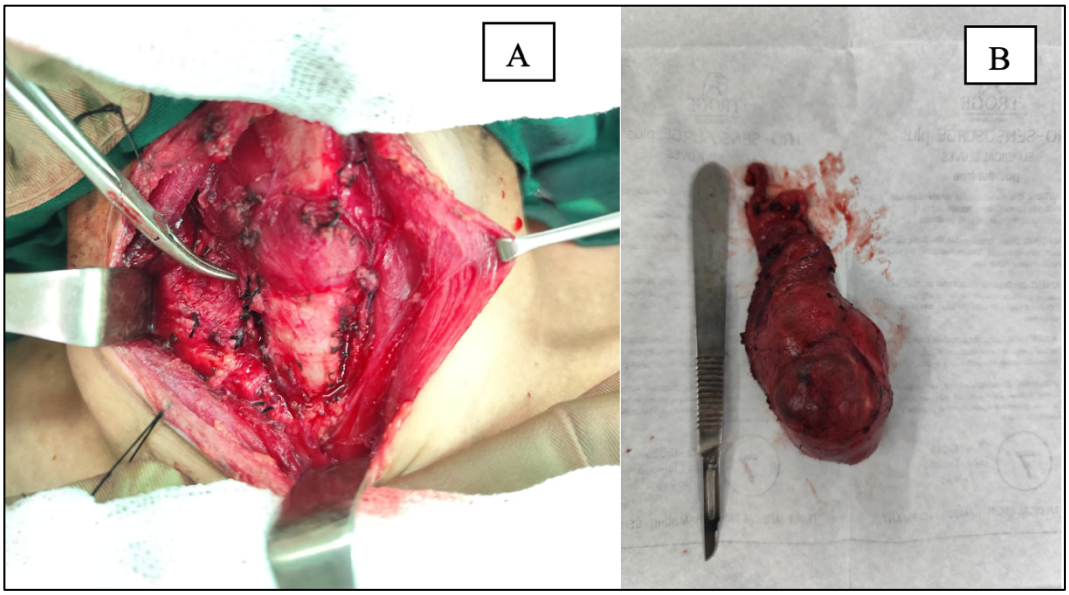

Figure 3. Istmolobectomy procedure (A) and macroscopic image of thyroid tissue (B).

3. DISCUSSION

PS is a rare disease, with an estimated prevalence ranging between 7.5 and 10 per 100,000 individuals. This disease accounts for approximately 4–10% of congenital deafness cases.[6,7] Therefore, PS is probably the most common cause of syndromic deafness. Based on the AR mode of inheritance, the risk of contracting from a heterozygous parent is 25% due to the AR mode of inheritance. [2]

The diagnosis of PS has evolved over the years through clinical presentation and family history, as well as thyroid function tests, perchlorate tests, and petrous bone radiography. Hearing loss is a consistent condition in PS and often manifests during the congenital or prelingual stage and is sensorineural.8 The onset of goiter varies significantly in severity and time which can be influenced by iodide intake. Recently, the utilization of molecular assays to detect mutations in the SLC26A4 gene has emerged as the preferred approach for the presumptive identification of PS. Molecular diagnosis requires the identification of bi-allelic mutations of PS gene in homozygous or compound heterozygous form.8 In addition, perchlorate testing can be carried out to screen for iodide organification diseases. This test includes measuring the intrathyroidal radioactive iodine content and administering radioactive iodine. A measurement that is less than 10% shows an organification diseases. However, a negative test result does not rule out the diagnosis of PS as many factors can influence the results. The previous use of high-dose iodine intake is a factor that can interfere with the test results.[7]

Although PS lacks a cure, the fine needle aspiration results showing a 1% malignancy risk enable informed management decisions and close monitoring. Large retrosternal goiter can cause obstructive symptoms in the future resulting in the consideration of total thyroidectomy.[9–12] However, total thyroidectomy can still cause recurrence due to persistent pathogenic factors. Thyroidectomy is not recommended in patients without symptoms of airway obstruction and malignancy. Early intervention measures can be taken to reduce thyroid growth and prevent disease progression, thereby reducing the possibility of obstruction and cancer. In addition, hearing loss in PS patients is often bilateral with the level of impairment ranging from "severe" to "very severe". Cochlear implantation is often performed in PS patients with quite satisfactory results.[13]

Loss of pendrin function results in a defect in organification and can lead to dyshormonogenetic goiter. This condition is an important clinical challenge due to malignant transformation, possibly caused by prolonged thyrotropin stimulation. In PS, the incidence of thyroid cancer is estimated to be approximately 1%. The most common histology of thyroid cancer arising from dyshormonogenetic goiter in patients with PS is follicular carcinoma, although papillary carcinoma also occurs.[14]

4. CONCLUSION

In conclusion, this study found a promising result in the case of PS in an adult man with an enlarged thyroid gland and hearing loss who underwent an isthmolobectomy on a large thyroid nodule.

CONSENT FOR PUBLICATION

Written informed consent was obtained from patient for publication in this case report. A copy of the written consent is available for review by the Editor-in-Chief of this journal upon request.

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Conflict of Interest Statement:

The author declares that the case report was conducted in the absence of any commercial or financial relationships that could be construed as a potential conflict of interest.