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(CASE REPORT)

# PENDRED syndrome with ectopic thyroid: About a delayed diagnosis

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## Abstract

We describe a case of a delayed diagnosis of Pendred syndrome. The patient had a history of total thyroidectomy 13 years ago .And presented when aged 30 with a cervical abscess of ectopic thyroid. The patient had a hearing loss from the early childhood and she had a 27 year old sister who present the same history of deafness and goiter.

Pendred syndrome is an autosomal recessive disorder characterized by sensorineural hearing loss and thyroid dyshormonogenesis. It is caused by mutations in the PDS/SLC26A4 gene encoding for pendrin. Hypothyroidism in Pendred syndrome can be--although rarely--present from birth and therefore diagnosed by neonatal screening. We highlight to the general physician the classical features of this syndrome that would aid early diagnosis.

**Keywords:** Goiter; Sensorineural deafness; Thyroid hormone; Pendred syndrome

## 1. Introduction

Pendred syndrome, an autosomal recessive disorder, is the most common syndromal form of hearing loss and involves abnormalities of the cochlea, varying degrees of sensorineural hearing loss and diffuse thyroid enlargement/goiter.

## 2. Case report

A 30-year-old patient who presented for a cervical abscess with breast abscess that had been developing in parallel for a few days before hospitalization.

Our patient had a total thyroidectomy 13 years ago at the age of 17 years for multinodular goiter.

She had a medical history of severe hearing loss since early childhood. Her main mode of communication is signing (Arabic sign language) and lip-reading.

The patient's parents were non-consanguineous. With a similar case in the 27-year-old sister including deafness and mutism since early childhood. The sister had never consulted for her symptomatology.

On examination our patient had a inflammatory cervical mass measuring 6 cm in diameter not fistulized to the skin; without compression signs.

She had a Inflammatory breast mass that drains purulent material related to breast abscess. And without abnormal pigmentation or proximal myopathy with normal brisk deep tendon reflexes elicited in all four extremities

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The biologic assessment showed elevated markers of infection, thyroid-stimulating hormone (TSH) elevated to 46 (normal range 0.3–5); in the past she had a elevated TSH

Ultrasound scan showed: multiple nodular formations under the left submaxillary gland on a probable left thyroid ectopy of which one of the nodules is classified EUTIRADS 4

Cervical computer scan: Left cervical lesions located in vascular structures with neighboring infiltration and compression of neighboring structures

The cervical and breast abscess was treated by antibiotic after incision and drainage. Investigations for deafness are planned including audiometric assessment, genetic testing is not yet available.

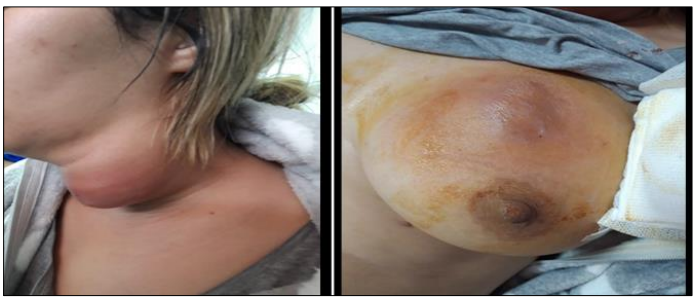

**Figure 1A** Inflammatory cervical mass related to abscess of left thyroid ectopy **1B** Left breast abscess

## 3. Discussion

Pendred syndrome was first described by Vaughan Pendred in 1896, after noting the coincident combination of hearing loss, mutism and goitre in an Irish family. It is an autosomal recessive inherited disorder, which may be accompanied by a goitre associated with sensorineural hearing loss. (1)

The gene responsible for Pendred syndrome was discovered and mapped to chromosome 7q31. The gene, *SLC26A4*, codes for the protein pendrin (PDS), a chloride-iodide cotransporter found in the thyroid, inner ear and kidney.

In the thyroid, pendrin transports iodide out of thyrocytes to allow for iodination of thyroglobulin. Mutated pendrin results in less iodide transported out and causes reduced organification of thyroglobulin, and goitre may form, though this is dependent on iodine intake.(2,3) Figure 2

More than 80 hearing loss genes, with more than 1200 mutations, have been identified, making it one of the most genetically heterogeneous traits. Advances in next-generation sequencing gene panel testing are now increasingly used to enable prompt, cost-effective and reliable genetic diagnoses to be made. Recent guidelines advise that following a detailed medical history (including birth history), family history, physical examination and audiometric assessment, genetic testing should be considered in cases of acquired hearing loss.(4,5,6)

Clinical presentation of Pendred syndrome can vary. Sensorineural hearing loss is often prelingual and profound, however it can fluctuate, developing later on in life, sometimes following on from a head injury.

A congenital goiter may be apparent and prompt the diagnosis, but some cases may have no thyroid enlargement at all. This varying clinical presentation can make diagnosis difficult, perhaps underestimating the prevalence of the syndrome

Goiter combined with a history of hearing loss should evoke suspicion of

Pendred syndrome. MRI of the auditory canals can aid in supporting these suspicions, and molecular testing can confirm diagnosis.(7,8,9,10)

Sensorineural hearing loss, though often prelingual and profound, can fluctuate and develop later on in life, sometimes following on from a head injury. It can also be associated with vestibular dysfunction.

There is no specific treatment of cure of Pendred Syndrome .Thyroid hormone deficiencies can be supplemented.

Speech and language therapy and cochlear implants can be offered. Genetic counselling can be offered to those planning a family.(11,12,13,14)

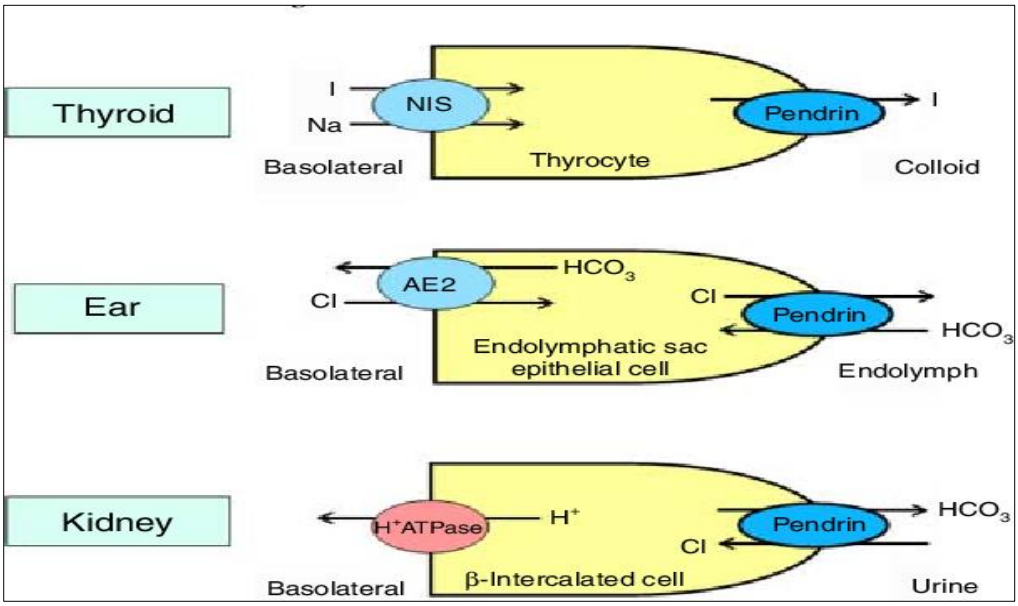


**Figure 2** Cellular functions of pendrin. In the thyroid, pendrin is involved in apical iodide transport; in the inner ear it transports bicarbonate into endolymph in exchange for chloride; and in renal collecting duct b-intercalated cells, it participates in urinary bicarbonate excretion with tubular chloride reabsorption. NIS, sodium iodide symporter; I, iodide; Cl, chloride; HCO3, bicarbonate. (4)

## 4. Conclusion

In conclusion, patients with Pendred syndrome exhibit distinct clinical features and the mechanisms associated with the molecular genetics of this disease have been clarified. it is important to determine an early diagnosis of Pendred syndrome; however, reducing the rate of missed diagnoses and misdiagnoses requires further investigation, thus, it is essential for clinicians to improve their understanding of Pendred syndrome. Misdiagnosis may be reduced, in part, by screening for suspected cases, obtaining a full report of the family history and performing relevant imaging examinations. The clinical understanding of Pendred syndrome, as well as the molecular and genetic research based on clinical diagnosis, will have a vital role in the prevention, early detection, diagnosis and treatment of this disease.

## Compliance with ethical standards

### *Acknowledgments*

Department of otorhinolaryngology.

### *Disclosure of conflict of interest*

No conflict of interest.

### *Statement of informed consent*

Informed consent was obtained from all individual participants included in the study.

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