



## Pendred Syndrome

### Alternative Names

PDS  
Deafness with Goiter  
Goiter-Deafness Syndrome  
Thyroid Hormonogenesis, Genetic Defect In, 2B  
Hypothyroidism, Congenital, due to  
Dyshormonogenesis, 2B  
Thyroid Hormone Organification Defect IIB

### WHO International Classification of Diseases

Endocrine, nutritional and metabolic diseases

### OMIM Number

274600

### Mode of Inheritance

Autosomal recessive

### Gene Map Locus

7q31

### Description

Pendred syndrome (PDS) is a common form of syndromic deafness. At least 5% of the total cases with congenital deafness are PDS. Typical PDS has the association of congenital bilateral neurosensory deafness, thyroid goiter, cochleovestibular malformation and potential vestibular dysfunction. The severity of the symptoms and the age of onset vary from case to case. Deafness often appears at birth, but it may develop in late infancy or early childhood. Sometimes, deafness is asymmetrical or fluctuant and often it is progressive. Abnormal bones of the inner ear can be observed in PDS. Thyroid status varies from euthyroid (goiter) to hypothyroidism. Goiter is secondary to abnormal iodine transport across the thyrocyte and it develops most commonly during adolescence. Incidence differs according to geographic location with a range between 1/100,000 births and 10/100,000 births. Audio-prosthetic management of deafness can be helpful and if the goiter becomes compressive

because of excessive size, a thyroidectomy must be performed.

### Molecular Genetics

Pendred Syndrome (PDS) is inherited as an autosomal recessive trait. The only gene known to be associated with PDS is the solute carrier family 26, member 4 gene (SLC26A4), or PDS gene. The normal PDS gene makes a protein (pendrin) that is found at significant levels only in the thyroid. Pendrin transports anions, particularly chloride (Cl<sup>-</sup>) and iodide (I<sup>-</sup>), into and out of the cells. This action is important for thyroid function and inner ear development. Alteration in PDS gene will impair the function of pendrin leading to thyroid malfunction and improper development of the ear. Mutations in PDS gene are found in more than 90% of typical cases and they differ according to ethnic group.

### Epidemiology in the Arab World

#### Egypt

Al Attia et al. (1986) reported an 18-year-old Egyptian male with Pendred Syndrome (PDS). His parents were first cousins and they were residing in the UAE. Around the age of one year, his mother suspected hear defect and a year later, she noticed that he was also dumb. At the age of 15, he developed goiter and being shorter in stature than the relevant group. Hormonal assays, radioactive iodine uptake, and radiological studies of bone age showed results of primary hypothyroidism with bone age of 11-13 years. Bradycardia and slow relaxing ankle jerks was observed in 17 year of age along with goiter. At 18 years, he had moderately sized multinodular goiter and a right hydrocele. His TSH level was elevated. Potassium Perchlorate Discharge test was performed showing a positive perchlorate discharge and indicated an organification defect (T3&T4 forming defect). The patient improved due to daily thyroxine



therapy. A radioactive iodine uptake (RAIU) was within normal range because of thyroxine therapy. A month later, the patient displayed a regression of his goiter and his pulse rate became within the normal range.

#### **United Arab Emirates**

[See also: Egypt > Al Attia et al., 1986]

#### **References**

Al Attia HM, El-Hag IA, Habab NH, Kazim S. Pendred's Syndrome. *Emirates Med J.* 1986; 4:140-2.

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