CONGENITAL DEAFNESS AND GOITER*

(Pendred's Syndrome)

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Pendred's syndrome is characterized by congenital deafness and goiter and is transmitted as an autosomal recessive disease. Some patients may also show symptoms of hypothyroidism. The association of congenital deafness and goiter was first described in 1824 by Wood[1] and later by Pendred[2] in 1896. In 1927 Brain reported cases of five families in which 12 members had congenital deafness and had shown symptoms of goiter during childhood.[3] After making extensive studies on his patients he came to the conclusion that deafness and goiter were two different expressions of the same recessive gene. In 1958 Morgans and Trotter[4] studied the thyroid functions of their two patients and found that the uptake of radioactive iodine was usually rapid with a significant and swift discharge of thyroidal radioactivity following potassium thiocyanate or potassium perchlorate administration. The case presented was also diagnosed on the basis of combined congenital deafness and goiter.

Case Report

A. C. (66—26715), a ten year old boy with complaints of deafness and of a swelling on his neck, was first seen at Hacettepe Medical Center on June 6, 1966. His history revealed that deafness had been noticed at about two years of age and that he had developed the swelling on his neck which had grown progressively larger.

Physical examination. It was found that he was a deaf mute and a 3+ goiter was noted. His blood pressure was 95/70 mm Hg, pulse rate was 92/min, weight, 27 kg and height 125 cm. The swelling primarily involved the right lobe and isthmus of the thyroid gland, with the left lobe only slightly enlarged. Superimposed on this swelling were four palpable nodules, which were easily movable, rubberlike in consistency and not tender (Figure 1). The rest of the physical examination was noncontributory.

Laboratory examination revealed a PBI of 3.0 microgram per 100 ml, BEI 1.04 microgram per 100 ml, cholesterol, 184 mg per 100 ml. The radioactive Iodine uptake (RAI) was 40 per cent and 35 per cent at four and at 24 hours respectively. The scanogram showed diffuse hyperplasia of the gland. The patient showed 25 per cent discharge at one hour following potassium thiocyanate

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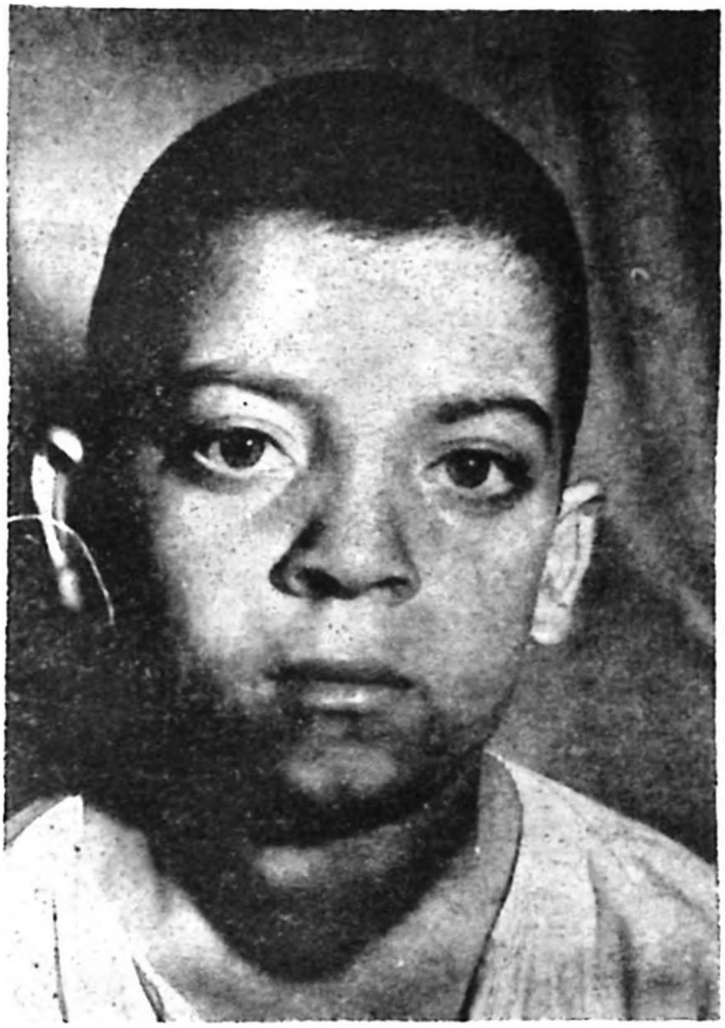

Figure 1.

ingestion. X-ray of the hand and wrist revealed a bone age of six years (Figure 2). The audiogram showed a high tone nerve deafness. The hearing loss was reported to be 85 decibels on the right and 75 decibels on the left.

A diagnosis of Pendred's syndrome was made on the basis of this perceptive type of deafness, goiter, high uptake of radioactive iodine and a significant discharge following potassium thiocyonate.* Although the patient did not show any significant clinical symptoms of hypothyroidism, the low BEI level and retarded bone age indicated the inadequate functioning of the thyroid gland.

Surgery was considered necessary because of the large size of the goiter and the presence of nodules. At the time of the operation, the right lobe and the isthmus of the thyroid gland were excised totally, but the left lobe was only partially excised.

Histopathologic examination revealed well delineated nodules in the thyroid tissue. The nodules showed areas of a papillomatous structure, microfollicles and degenerative changes. No invasion of the vessels was noted (Figure 3a,b,c).

After a diagnosis of multinodular goiter, the patient was put on a thyroid extract treatment and was discharged from the hospital on the seventh day after operation.

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* According to Thould and Scowen, 15 per cent or higher discharge is significant.[5]

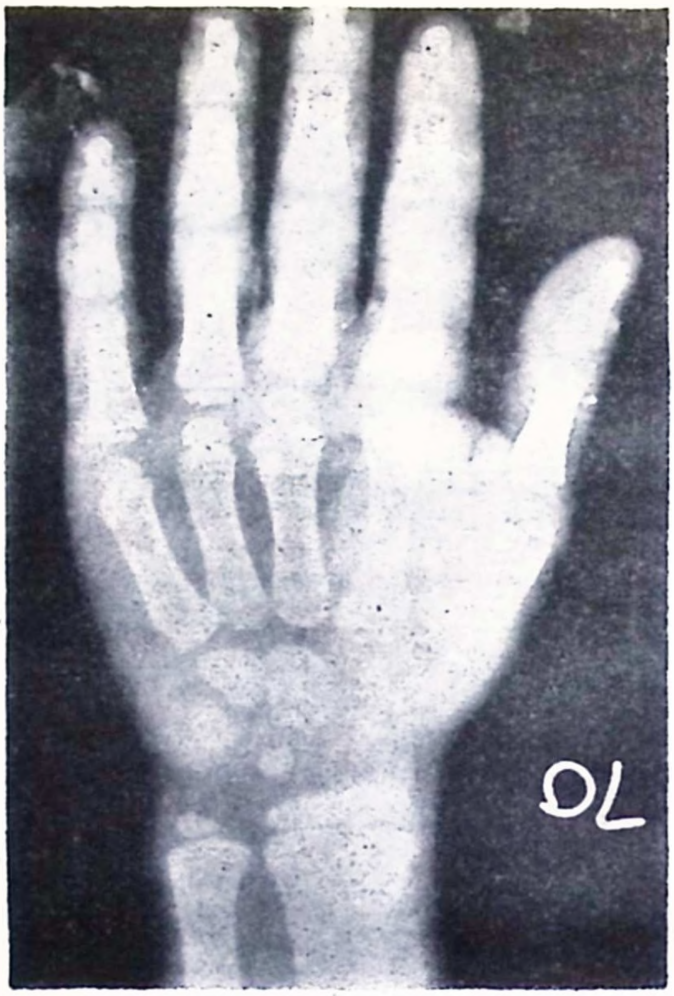

Figure 2. X-ray reveals a bone age of six years.

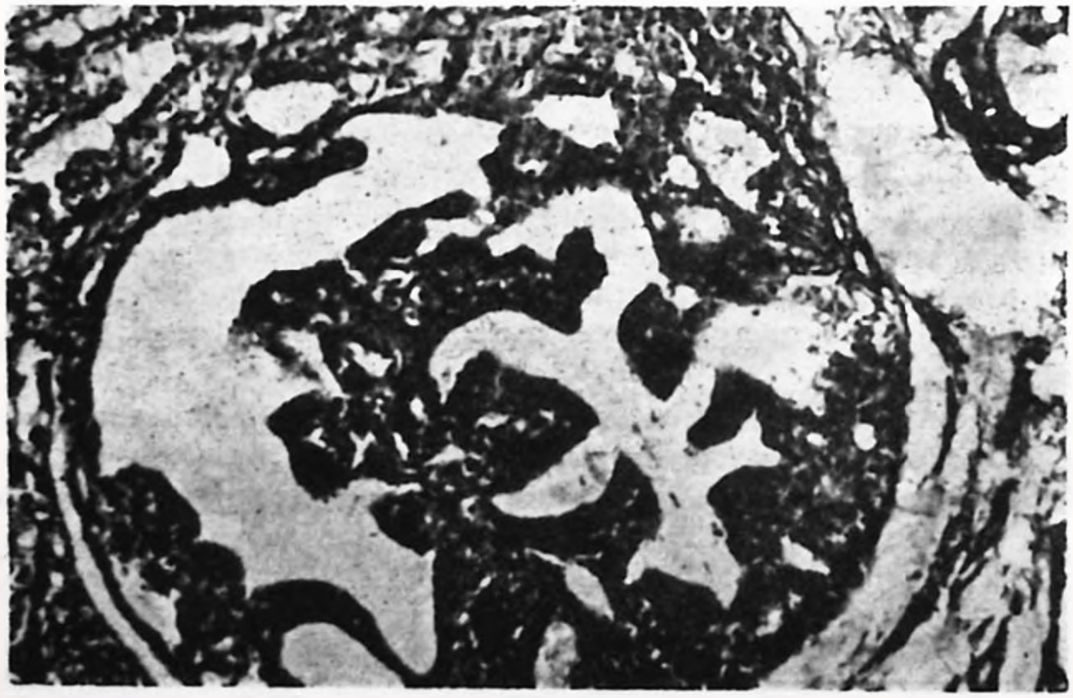

Figure 3a.

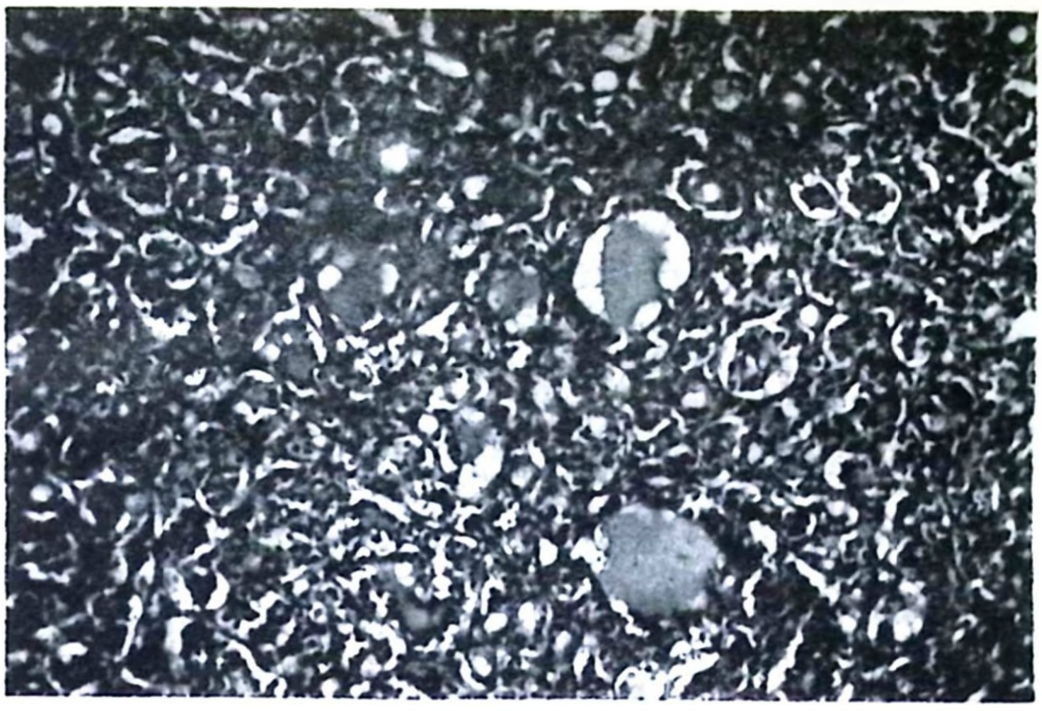

Figure 3b.

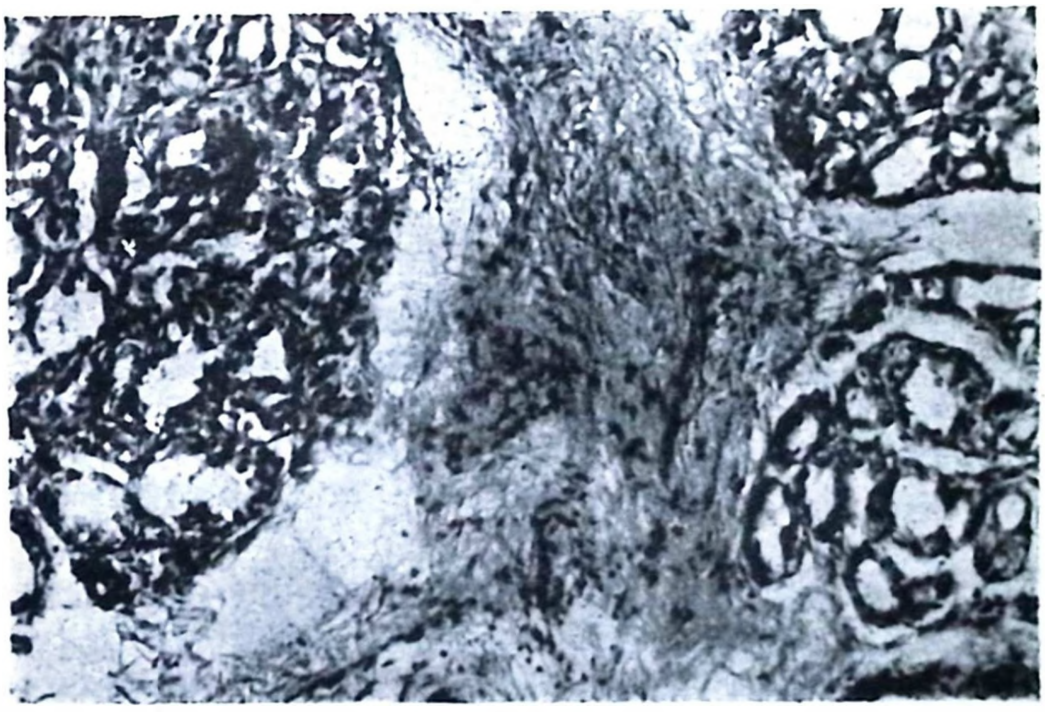

Figure 3c.

Figure 3a, b, c. Nodules show areas of papillomatous structure, microfollicles and degenerative changes.

Discussion

Morgans and Trotter studied extensively the nature of the thyroid disorder in Pendred's syndrome and suggested the presence of a partial defect in the binding of iodine to protein (iodination). Because of this defect, the rate of iodination would become less than the entering rate of iodine and subsequently would cause the iodine

to accumulate in the gland. Biosynthetic pathway of thyroxin is shown in Figure 4. In 1952 Stanbury and Wyngaarden showed that this accumulated iodine in the thyroid gland could be discharged following the administration of perchlorate or thiocyanate.[7] Because of the defect in the synthesis of the thyroid hormone TSH secretion is increased eventually causing goiter. Although goiters are often present at the age of seven or eight years, it may not be noticed until puberty. Some patients may also show symptoms of hypothyroidism.

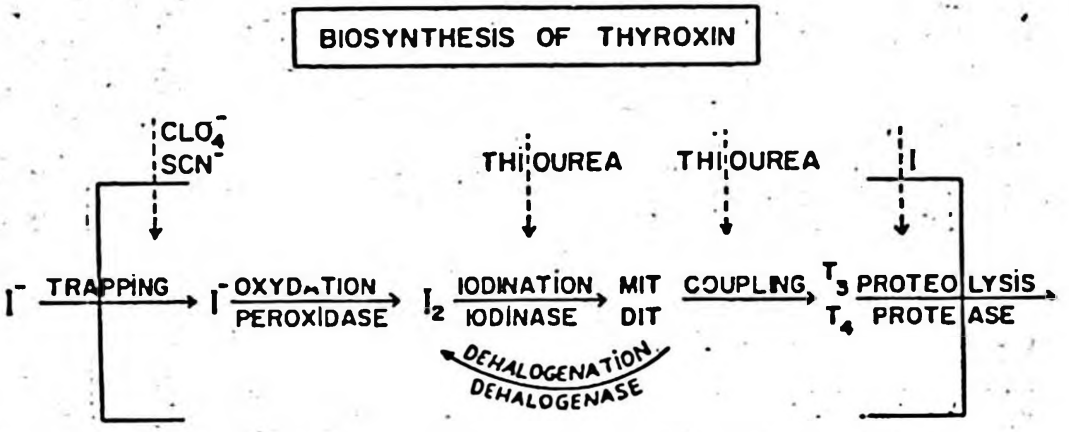

Figure 4.

The nature of the hearing loss is more complex. The thyroid hormone is necessary for the normal development and maturation of the central nervous system especially during the fetal stage. Although the transmission of the thyroid hormone through the placenta has been shown by Grumbach and Werner,[8] because of the very slow rate of transfer, the maternal hormone may not be adequate for the normal development of the fetal brain. According to studies by Hodges and coworkers[9] the fetal thyroid begins to function at the twelfth week of fetal life. If the contribution of the fetus' thyroid hormone is inadequate because of an inherited defect, the normal development of the central nervous system will be incomplete. The affected fetal thyroid is unable to form enough hormone for the normal maturation of the acoustic nerve, hence irreversible damage may occur. Thould and Scowen believe that the damage to the acoustic nerve of children with congenital deafness and simple goiter, is associated with hypothyroidism **in utero.** The association of goiter and deaf-mutism in the endemic goiter region suggests the close relation between these two conditions. Wespi reported a distinct decrease in the percentage of goiter and deaf-mutism in the Swiss after introduction of iodine prophylaxis.[10] On the other hand, Fraser accounts for the common occurrence of goiter and deaf-mutism in

individuals in the endemic goiter regions by intermarriage in this area.[11]

The deafness in Pendred's syndrome is high tone nerve deafness and is congenital. This defect is usually detected at about two years of age.

Genetically, the mode of transmission in Pendred's syndrome is autosomal recessive and its gene frequency changes are between 1/1500 and 1/500.

Smith, in a study of fourteen cases, found that the pathologic findings were : 1. Macroscopically, an appearance of nodular goiter which recurred after subtotal thyroidectomy. 2. Microscopically, they showed epithelial hyperplasia, colloidal secretion and some degenerative changes such as fibrosis, calcification and hemorrhage.[12]

Summary

The case of a ten year old boy with goiter and deafness is presented. The diagnosis of Pendred's syndrome was made on the basis of a perceptive type of deafness and discharge studies. The patient underwent subtotal thyroidectomy with subsequent thyroid treatment.

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