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Familial Occurrence of Congenital Hypothyroidism Due to Lingual Thyroid Gland

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Two sisters who presented with midline neck masses proved to be biochemically hypothyroid. Thyroid scintigraphy supplemented with perchlorate discharge testing showed lingual thyroid glands and ruled out the expected autosomal recessive organification defect. The related literature is reviewed.

Key words: Congenital, Familial, Lingual Hypothyroidism.

PERMANENT congenital hypothyroidism results from various types of thyroid gland defects. These include thyroid aplasia or hypoplasia and dysgenic lingual or sublingual glands, which occur sporadically, and the autosomally inherited form, which results from an inborn error of thyroid hormone synthesis (dyshormonogenesis) primarily because of a lack of the peroxidase enzyme (1–3). The latter is characterized by high Tc-99m pertechnetate and I-123 uptake with rapid and high perchlorate discharge (4).

In this report, we describe two sisters who had lingual thyroid glands with low Tc-99m uptake and normal perchlorate discharge.

Case Reports

Case 1

A 9-year-old Saudi girl presented with a midline neck swelling that measured 0.5 × 0.5 cm. The swelling was firm and moved with tongue movements (Fig. 1). There was also a protruding mass at the back of the tongue (Fig. 2). She had a poor appetite and a small build. Her past medical and developmental history were unremarkable, and her performance at school was good. Her growth parameters were normal for her age, and the results of her physical examination were normal. Laboratory investigation revealed a free triiodothyronine level of 9.2 pmol/l (normal range, 4.6–9.2 pmol/l), free thyroxine level of 13.3 pmol/l (normal range, 10.3–25.8 pmol/l), and thyroid-stimulating hormone level of 7.8 mIU/l (normal range, 0.25–5 mIU/l).

Thyrotrophin-releasing hormone stimulation test was performed before therapy was initiated using standard procedures (5), with thyroid-stimulating hormone levels measured at 0, 30, 60, 90, 120, and 180 minutes. The results, shown in Table 1, are consistent with primary hypothyroidism.

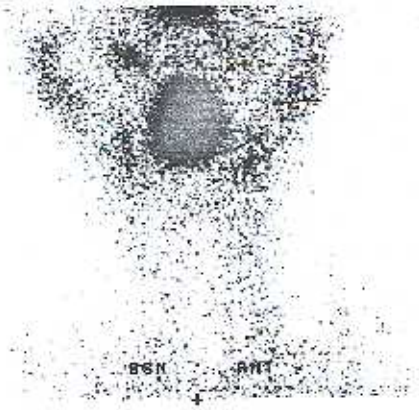

Fig. 1. A Tc-99m pertechnetate scan shows a lingual thyroid gland.

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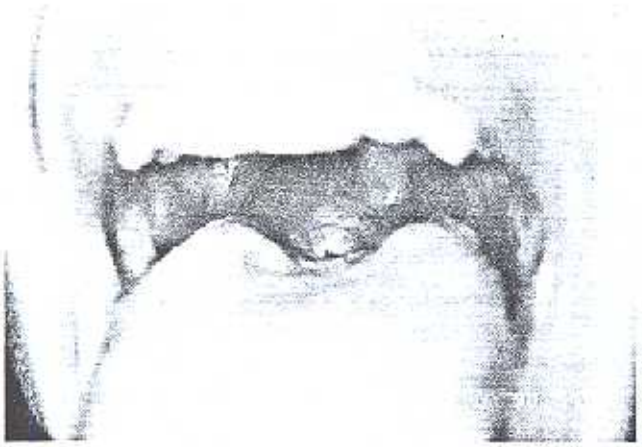

Fig. 2. An open-mouth view shows a protruding lingual thyroid gland at the back of the tongue.

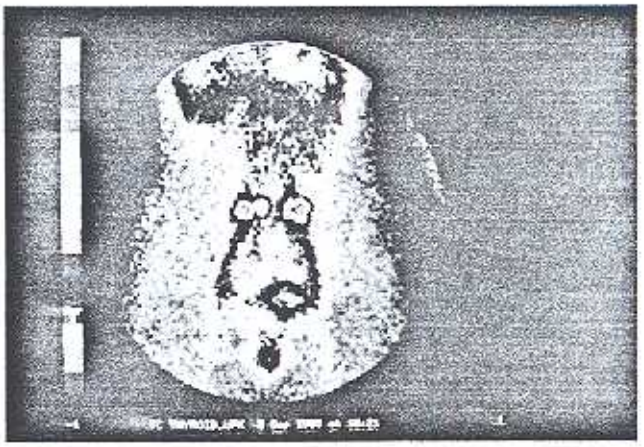

Fig. 3. An anterior abdomen view of the Tc-99m pertechnetate scan shows the autotransplanted thyroid gland.

Case 2

A 20-year-old Saudi woman, the sister of our first patient, was evaluated when she was 9 years old because of a swelling at the posterior third of the tongue of which she had been aware for 6 years. She was treated with autotransplantation, after which she remained in good health (Fig. 3). The results of her physical examination were unremarkable. Laboratory investigations revealed a free triiodothyronine level of 7.25 pmol/l, a free thyroxine level of 11.68 pmol/l, and thyroid-stimulating hormone level of 9.92 mIU/l. The results of a thyrotrophin-releasing hormone stimulation test were consistent with primary hypothyroidism (Table 1). There was no similar family history, and their parents were consanguineous. L-thyroxine was begun in both sisters, who continue to be clinically and biochemically euthyroid.

Discussion

Thyroid dysgenesis is the most common cause of congenital hypothyroidism (1,3). It usually occurs sporadically. Dyshormonogenesis usually follows an autosomal recessive pattern of inheritance. Organification defects, coupling defects, thyroglobulin defects, and others are forms of this inherited disorder of thyroid metabolism (2). These are more common in the Saudi community, where the consanguinity rate is relatively high (6). Majeed-Saidan et al. (7), Al-Jurayyan et al. (8), and Al-Nuaim et al. (9) showed that the prevalence in such a community is higher compared with other series (1,3,6).

O'Connor et al. (10) observed increased uptake in some of their patients with lingual thyroid glands and suggested the possibility that dyshormonogenesis may coexist with an inability of the gland to descend (10). However, the perchlorate discharge test was not performed in their series. In 1985, Hilditch and Jackson (11) were the first to report a positive perchlorate discharge test result in infants with ectopic thyroid glands. Recently, Al-Jurayyan and El-Desouki (12) reported the transient nature of the iodine organification defect in infants with ectopic thyroid glands and suggested that a delay in the development of synthetic mechanisms occurs in dysgenic glands.

The familial occurrence of congenital hypothyroidism resulting from thyroid ectopy has been reported by Orti et al. (13) and Kaplan et al. (14), who suggested a hereditary factor in controlling the development and descent of the thyroid gland. The results in our sibling patients, although they might represent coincidental findings, support this theory and indicate further the need to study the genetic role in the development and descent of the thyroid gland.

TABLE 1. *Thyroid Stimulating Hormone Values After Thyrotrophin-Releasing Hormone Stimulation*

		0 min	30 min	60 min	90 min	120 min	180 min
Case 1	TSH (mIU/l)	7.8	55	39	27	18	13
Case 2	TSH (MIU/l)	9.6	104	88	39	28	21

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