Graves' disease with thyroid hemiagenesis: A rare abnormality with rarer presentation

Sir,

Thyroid hemiagenesis is a rare developmental abnormality of thyroid gland. Most of the cases of thyroid hemiagenesis are euthyroid, but rarely can be associated with hyperthyroidism, hypothyroidism or malignancy. The incidence of Graves' disease in thyroid hemiagenesis is rare and we report such a case.

A 50-year-old female patient presented with a history of swelling in the left side of the front of neck of 6 months duration. There was no family history of thyroid disease or any other developmental abnormalities. On examination, goitre was present, World Health Organization grade 2, firm, which was on the left side [Figure 1]. The right lobe of thyroid was not palpable. The thyroid function tests carried out were suggestive of thyrotoxicosis, with T3 227 ng/dl (80-200), T4 of 14.9 μ g/dl (4.5-12.5) and thyroid-stimulating hormone 0.02 µiu/ml (0.35-5.50). Anti-thyroid peroxidase antibody was positive (>1300 IU ml). An ultrasound thyroid was done, which showed an enlarged left lobe of thyroid, with mildly coarsened echo texture and increased vascularity, with absent right lobe and isthmus [Figure 2]. A technetium uptake study was carried out, the perfusion study showed increased vascularity to the left lobe of the thyroid gland and delayed static image showed enlarged left lobe of thyroid with increased tracer uptake with uniform distribution. The total uptake was 5.1%. The right lobe was not visualized [Figure 3]. Based on the above clinical details and investigations, a diagnosis of Graves' disease with hemiageneis of the right lobe of the thyroid was made.

Congenital abnormalities of the thyroid gland is rare and may be related to abnormal descent of thyroid gland, presenting as single or multiple ectopic thyroid tissue or may include structural abnormalities such as hypoplasia or hemiagenesis. [1] Thyroid hemiagenesis is an uncommon condition with only about 250 cases reported in literature until date. [2] Hemiagenesis is more common on left side with a left: Right ratio of 4:1. Most cases are sporadic but a few cases of familial hemiagenesis have been reported. [3,4]

Most of the patients with thyroid hemiagenesis are euthyroid, but hyperthyroidism, hypothyroidism and malignancy have been reported.^[5] The incidence of hyperthyroidism of the remaining lobe, as with our case is, is not well-known and literature review yielded only six case reports of the above condition.

When a patient presents with unilateral thyroid swelling, the possibilities, which are usually considered are thyroid adenoma, post-hemithyroidectomy status, or a large nodule. If the uptake



Figure 1: Unilateral thyroid swelling (left side)

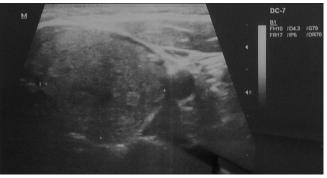


Figure 2: Ultrasound thyroid showing enlarged left lobe and absent right lobe



Figure 3: Technetium uptake study showing increased uptake in left lobe and non-visualized right lobe

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study shows absent uptake on one side, the pertinent differential diagnosis include a hyper functioning nodule of the same side which is suppressing the rest of the gland, or a cold nodule on the same side. Thyroid hemiagenisis is a rare, but important differential diagnosis in this scenario and unless the thyroid uptake study is interpreted along with an ultrasound of thyroid, which picks up hemiagenesis, this rare diagnosis will be missed. Radio iodine ablation is the safe and convenient modality of treatment for Graves' disease in the presence of structural abnormalities of thyroid gland.^[1]

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