Dyshormonogenetic Goiter: A Case Report

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ABSTRACT

Several types of familial goiter have been defined on the basis of clinical and biochemical data. Those goiters caused by inherited enzyme defects in hormone synthesis are called dyshormonogenetic goiters. Because this form of goiter is commonly associated with pleomorphism, hypercellularity and mitotic activity of the follicular epithelium, one should be particularly strict with the criteria for the correct diagnosis, especially in FNA. We report an 11-year-old boy presented with hypothyroidism and thyroid enlargement. FNA result was malignancy. Total thyroidectomy was performed and the pathology report was dyshormonogenetic goiter.

Key words: Goiter, Thyroid hormones, Nodular

Introduction

Those goiters caused by inherited enzyme defects in hormone synthesis are known as dyshormonogenetic goiter. The enzyme defects in DG are inherited as autosomal recessive (1). The genetic mutations responsible for these various defects are being currently unraveled (2). The architectural polymorphism and cellular atypia in these types of goiters mimic thyroid neoplasm specifically nothing short of clear cut capsular or blood invasion should be accepted (3,4). Thus, because it is important to correct the diagnosis of this disease that mimics malignant lesions in FNA we presented this case to avoid the over-diagnosis of malignancy and prevent unnecessary surgery.

Case report

The patient was an eleven-year-old boy who was admitted with thyroid enlargement and a past history of hypothyroidism from birth. In out-patient clinic, FNA was performed and the result was malignancy. He had hypothyroidism in his family. Before surgery done all the thyroid hormones tests were normal and he was euthyroid due to hormone therapy. Finally, total thyroidectomy was done. On gross examination, the thyroid was enlarged, bosselated and on cut section the surface was nodular. The thyroid weight was 70 g. Histopathological examination revealed multinodular formations that surrounded by fibrous bands with marked follicular hyperplasia and hypercellularity with decreased colloid material. The follicular cells had a severe cytologic atypia including bizarre and markedly enlarged, hyperchromatic nuclei. These nuclear changes were mostly seen in inter-nodular areas (Figures 1-3). Finally, dyshormonogenetic goiter was confirmed by consultation with other four pathologists in our center.
Discussion

Thyroid gland is controlled by TSH which in turn is influenced by TRH. TSH permits growth, cellular differentiation and thyroid hormone production and secretion by the thyroid gland. A deficiency in thyroid hormone synthesis or intake leads to increased TSH production. If this process is sustained, a goiter is established (5). Congenital primary hypothyroidism occurs in 1 of 4000 births, whereas the majority of the cases are due to developmental defects of thyroid gland. In this respect, 20% of these cases carry a defect in thyroid hormonogenesis (6). There are several types of goiter resulting from enzyme defects in hormone synthesis (7,8). These include lack of responsiveness to TSH, defects in coupling, abnormalities of thyroglobulin synthesis and secretion and others (9). Grossly, the gland is enlarged and multinodular. Microscopically, the most common alteration consists of hypercellular nodules exhibiting a variety of architectural appearances with a predominance of solid and micro follicular patterns (10,11). Fibrosis is a common finding (12). Other common features include marked nuclear atypia and minimal amount of colloid and mitotic figures are often seen. The differential diagnosis could be follicular carcinoma, atypical bizarre adenoma, papillary carcinoma, Grave’s disease but strict histological criteria will help to separate these lesions, presumably as a result of the continuous TSH stimulation (13). The possible role of growth factor and oncogenes in this form of goiterogenesis has also been mentioned (14). Cases of thyroid carcinoma have also been reported in patients with DG, but the number of well-documented cases are very low (15). Mostly, they have been of follicular type and others have been incidental papillary microcarcinoma (11). Correlation with the history, clinical findings, levels of thyroid hormones, and other investigations is imperative for its diagnosis.

The degree of hyperplasia in this form of goiter is severe enough to prompt a diagnosis of malignancy, particularly in FNA. For this reason, it is important that in young patients who have a past history of hypothyroidism in himself and family and come with thyroid enlargement and nodule, the FNA should be reported with caution to avoid the unnecessary surgery.
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References


