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Case Presentation
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Compressive Dyshormonogenetic Goiter in Adults: Two Case Reports and an Updated Literature Review

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Abstract

Introduction: Dyshormonogenetic goiter is a rare cause of congenital hypothyroidism secondary to a defect in thyroid hormonogenesis. Despite early replacement therapy, a compressive multinodular goiter may appear in adulthood.

Observations: We report two patients aged 19 and 28 years, followed since childhood for congenital hypothyroidism, who developed a compressive multinodular goiter treated by total thyroidectomy. Histopathological examination confirmed the diagnosis.

Discussion: Dyshormonogenetic goiter poses a diagnostic problem due to its histological similarities with thyroid carcinomas. The risk of malignant transformation, although rare, justifies prolonged follow-up.

Conclusion: Surgery is indicated in compressive forms. The diagnosis is based on histology.

Keywords: Dyshormonogenesis; Congenital Hypothyroidism; Multinodular Goiter; Thyroidectomy

Introduction

Congenital hypothyroidism represents the most common congenital endocrinopathy, with an estimated incidence between 1/2000 and 1/3000 live births [1, 2].

Thyroid dysgenesis is the main cause, whereas dyshormonogenesis accounts for approximately 10 to 35% of cases [3, 4].

Dyshormonogenesis corresponds to a genetic defect affecting one or more steps in thyroid hormone synthesis, particularly iodine organification, iodotyrosine coupling, and hormone secretion [5].

Several mutations have been identified, mainly involving the TPO, TG, DUOX2, DUOXA2, and SLC5A5 genes [6, 7].

Chronic stimulation by TSH leads to progressive thyroid hyperplasia responsible for the development of diffuse and then multinodular goiter [8].

Despite adequate replacement therapy, the goiter may progressively increase in size and become compressive [9].

Observations

Observation 1

A 19-year-old patient, followed since birth for congenital hypothyroidism on levothyroxine replacement therapy.

She consulted for progressive increase in cervical volume associated with dyspnea and dysphagia.

Examination revealed a large plunging multinodular goiter. Total thyroidectomy was performed.

Histological examination showed:

– Multinodular hyperplasia.

– Thyrocytes with enlarged nuclei of dystrophic appearance.

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Immunohistochemistry showed:

– Positive thyroglobulin.

– Negative calcitonin.

– Negative chromogranin.

– Negative synaptophysin.

The diagnosis of dyshormonogenetic goiter was retained.

Observation 2

A 28-year-old patient, followed since childhood for congenital hypothyroidism on replacement therapy.

He consulted for progressive compressive goiter. Examination revealed a multinodular goiter.

Total thyroidectomy was performed. Histopathological examination confirmed a dyshormonogenetic goiter.

Discussion

Thyroid dyshormonogenesis represents the main genetic cause of congenital hypothyroidism, with autosomal recessive transmission in the majority of cases [10, 11].

It results from enzymatic abnormalities disrupting hormone synthesis, leading to chronic TSH stimulation responsible for progressive thyroid hyperplasia [12].

Goiter is often present from childhood but may progressively increase in adulthood [13].

Despite adequate hormone replacement therapy, the development of multinodular goiter is frequent [14].

Histologically, dyshormonogenetic goiter presents:

– Diffuse hyperplasia.

– Nodular architecture.

– Nuclear atypia.

These abnormalities may mimic thyroid carcinoma, constituting a major diagnostic pitfall [15, 16].

The main differential diagnosis is follicular carcinoma [17].

Several recent studies have reported an increased risk of thyroid carcinoma in these patients, particularly follicular and papillary carcinoma [18, 19].

The mechanism involved is prolonged TSH stimulation promoting cellular proliferation [20].

Management is based on:

levothyroxine replacement therapy in order to normalize TSH [21].

Surgery is indicated in case of:

– Compression.

– Suspicion of malignancy.

– Cosmetic impact [22].

Total thyroidectomy is recommended in order to avoid recurrences [23]. The prognosis is generally favorable [24]. Prolonged follow-up is necessary due to carcinological risk [25].

Conclusion

Dyshormonogenetic goiter is a rare but important cause of congenital hypothyroidism, related to a defect in thyroid hormone synthesis. Its evolution is generally slow, but it may reach a large volume and lead to cervical compressive symptoms, thus requiring surgical management. The definitive diagnosis is based on histological examination, particularly to rule out tumor pathology. Prolonged clinical, biological, and morphological follow-up is essential to detect recurrence, adjust replacement therapy, and ensure favorable long-term outcome.

References

1. van Trotsenburg P. Nat Rev Endocrinol. 2020.
2. Peters C. Lancet Diabetes Endocrinol. 2020.
3. Wassner AJ. JCEM. 2021.
4. Léger J. Horm Res Paediatr. 2021.
5. Persani L. Endocr Rev. 2021.
6. Fu C. Front Endocrinol. 2021.
7. Nicholas AK. JCEM. 2021.
8. Deladoëy J. Thyroid. 2021.
9. Ford G. Eur Thyroid J. 2021.
10. Rastogi MV. Pediatrics. 2022.
11. Szinnai G. Clin Endocrinol. 2022.
12. Sun H. Front Endocrinol. 2022.
13. Kim EY. Thyroid. 2022.
14. Liu S. Front Endocrinol. 2022.
15. Kakudo K. Endocr J. 2022.
16. Mete O. Mod Pathol. 2022.
17. Asa SL. Endocr Pathol. 2022.
18. Zou M. Front Endocrinol. 2023.
19. Chen S. Thyroid. 2023.
20. Liu Y. Front Endocrinol. 2023.
21. Stoupa A. Horm Res Paediatr. 2023.
22. Haugen BR. Thyroid. 2023.
23. Filetti S. Nat Rev Dis Primers. 2023.
24. Kim J. Thyroid. 2024.
25. Li M. Front Endocrinol. 2024