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CHIRURGIE CERVICO-FACIALE ET CANCÉROLOGIE

An uncommon and distinctive type of goiter to recognize: about 5 cases

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Introduction

Dyshormonogenetic goiter is a rare entity that affects 1 in 30,000 to 50,000 live births, and it is the second most common cause (10% to 15%) of permanent congenital hypothyroidism. It is morphologically characterized by architectural and cellular pleomorphism that may mimic thyroid malignancy and cause difficulties in differential diagnosis.

Aim of the study

To discuss the cases of the dyshormonogenetic goiter and to show its particularities

Methods

We present five cases of dyshormonogenetic goiter gathered at our department.

Résultats

- The cohort comprised three patients, 2 females, and 3 males
- **Average age** of 18.6 years (range: 7 to 32 years).
- All five were offspring of **consanguineous marriages** and were under observation for **congenital hypothyroidism**.
- **Chief complaint**: (all patients) was a basal cervical swelling.
- **Clinical examination:** multinodular goiter in 4 patients and a right lobar nodule in one patient. Delayed growth and psychomotor development were noted in two patients.
- **Cervical ultrasound and thyroid scintigraphy** were performed in all cases.
- **Ultrasound:** multinodular goiter in all patients. (Fig,1)
- **Scintigraphy:** thyroid hyperfixation in all cases, with a hypofixation zone on the right side in one patient.
- **Thyroid function tests:** hypothyroidism in all five patients.
- All patients underwent surgical treatment: total thyroidectomy in four cases and right lobectomy in one case.
- **Histopathological examination** confirmed dyshormonogenetic goiter in all five cases.
- **Favorable outcomes** were observed in all cases, with no recurrence .

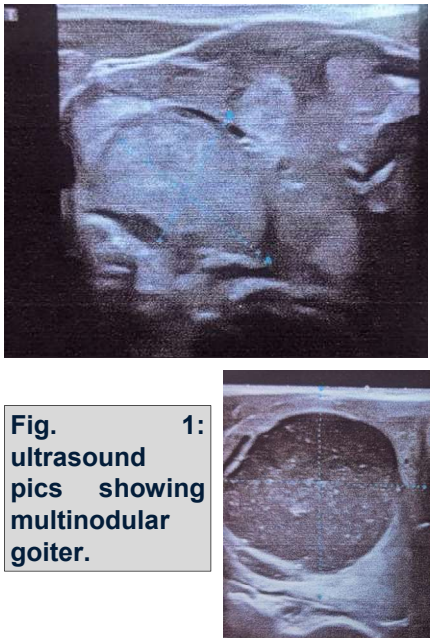

Fig. 1: ultrasound pics showing multinodular goiter.

Conclusion

Dyshormonogenetic goiter represents a rare benign condition characterized by architectural and cytological characteristics that, if not well understood, may contribute to the overdiagnosis of malignancy. Recognition of this entity becomes crucial in cases lacking strict histological criteria for malignancy, particularly in individuals with a history of hypothyroidism since infancy. An exact molecular diagnosis allows genetic counseling and the identification of asymptomatic mutation carriers..