“Semi-Hot” Thyroid Nodules Associated with GNAS Mutations in Four Adolescents

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BACKGROUND

Hot thyroid nodules are uncommon in children and adolescents. Hyperfunctioning adenoma do not always produce hyperthyroidism, but can precede the apparition of a truly toxic adenoma. Autonomous adenoma can be associated with somatic mutations of the TSH receptor or somatic activating mutations of Gsa (GNAS mutation).

PATIENTS

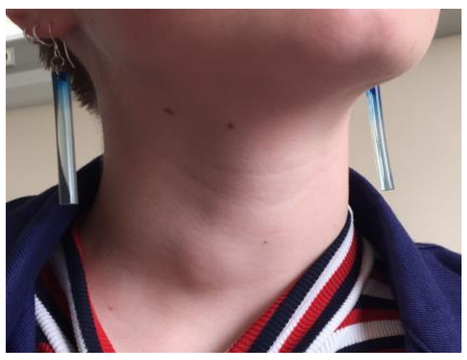

Figure 1 : Clinical Manifestation

Table 1 : Biological manifestation

	P1	P2	P3	P4	NV
T3L (pmol/l)	7	11,8	10,74	6,7	3,1-6,5
T4L (pmol/l)	18	18,3	18,5	12,6	10,2-22
TSH (mUI/L)	0,23	0,001	0,008	0,28	0,3-4,5

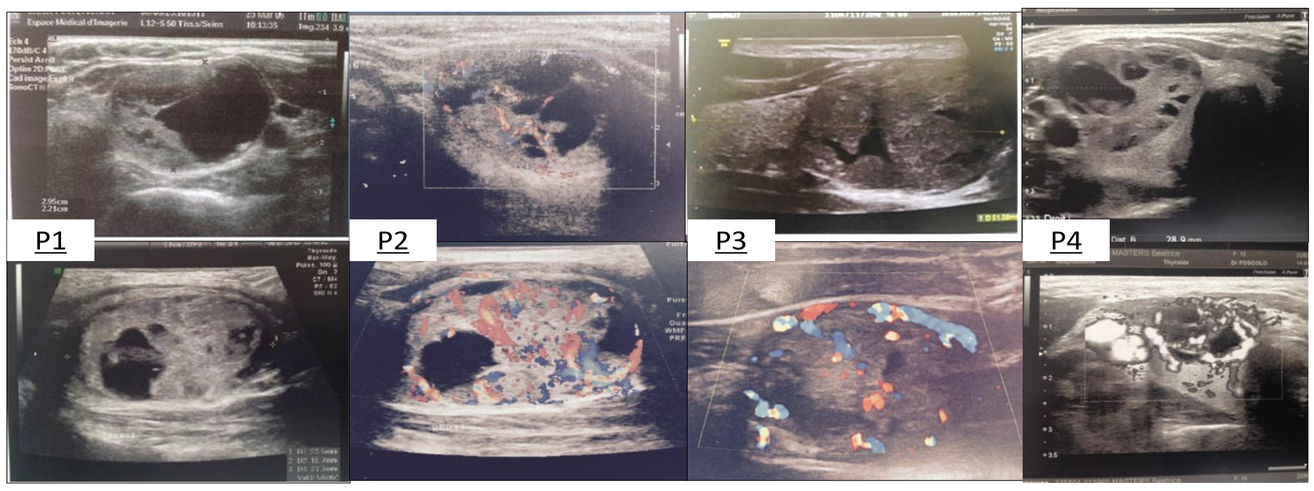

Figure 2 : Ultrasonography

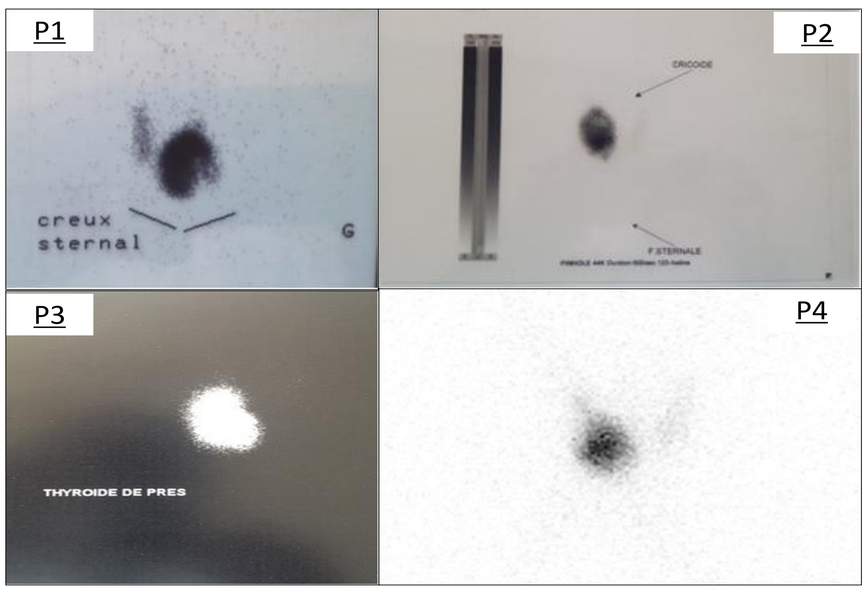

Figure 3 : Radionuclide scintigraphy

We report the clinical and molecular data of 4 patients referred in our unit for management of thyroid nodules (Figure 1),

Patient 1 (P1) presented mild clinical hyperthyroidism. Patients 2,3 and 4 (P2,P3 and P4) were asymptomatic and had clinical euthyroidism.

Examination revealed a unique isolated thyroid nodule in the 4 patients (diameter of 30.5, 30, 31 mm, respectively). Café-au-lait spots were present in Patient1.

Ultrasonography showed encapsulated nodules with increased vascularity in all 4 patients (Figure 2),

Radionuclide scintigraphy showed hyperfunctioning nodules with absent uptake in the surrounding tissues in 3/4 and decreased uptake in 1/4.

Partial thyroidectomy was performed in 4/4 patients. Molecular examination revealed GNAS mutations in the 4 patients. Postoperative period was uneventful.

CONCLUSION

Mild hyperthyroidism or thyroid palpation in asymptomatic patients can reveal hyperfunctioning nodules as the seemingly unique manifestation of GNAS activating mutations.

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