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

Anne Sophie Lambert (1), Danielle Rodrigue (1), Agnès Linglart (1), Jean François Papon (2) and Pierre Bougnères (1)
1. Department of Pediatric Endocrinology, Bicêtre Hospital, Paris, France
2. Department of Otolaryngology-Head and Neck Surgery, Bicêtre Hospital, Paris, France

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

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 Patient 1.

Ultrasoundography 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.