Prevalence and Characteristics of thyroid nodules in a pediatric population with congenital hypothyroidism

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INTRODUCTION
Thyroid dyshormonogenesis accounts for 10-15% of permanent congenital hypothyroidism. Patients can present with goiter. Thyroid nodules may also be associated.

AIM
To assess the characteristics of thyroid nodules among infants diagnosed with thyroid dyshormonogenesis and to discuss the interest of a systematic screening.

METHOD
A retrospective study of children with congenital hypothyroidism due to dyshormonogenesis was carried out at the pediatric endocrine center in Kremlin Bicêtre hospital, Paris, between 1999 to 2020. A screening ultrasound for thyroid nodules was then performed from July 2019 to July 2020 if no recent analysis was found (>3 years).

RESULTS

78 patients
Median age 14 ans [8-21]
47 boys / 31 girls

Disorder of iodine organification
50/78 : 64% 
Genetic results available 13/50 :
2 Pendred syndrome
10 TPO gene mutation
1 DUOX gene mutation

Disorder of iodine uptake
3/78 : 4% 
Genétique : 1 NIS

No diagnostic in 18/78 patients.

Disorder of thyroglobulin synthesis
7/78 10% 
Génétique : 3 TG

42/78 patients were treated by L-thyroxine (54%)
TSH : 2.6 mUI/l [0.1-6.2]

10 Nodules (median size of 17 mm [5-37]) and 1 multinodular goiter were detected in 8 and 1 patient respectively during follow-up (median average of 2.5 [1-3])

Nodule classification :
1/10 TI-RADS 2
5/10 TI-RADS 3
4/10 TI-RADS 4.

Fine needle aspiration biopsy were realised in 3 patients, and 5 had thyroidectomy.

Diagnosis was thyroid cyst in 6/10 patients, Follicular lesion in 1/10, papillar carcinoma in 2/10 (1/2 patient with TPO gene mutation) et 1 multinodular thyroid.

CONCLUSIONS
The prevalence of thyroid cancer in our pediatric population is 2.5%, the prevalence of nodules is 7.6% including 20% of malignant nodules. Thyroid cancer occur in younger patients in the context of dyshormonogenesis. Given the importance of this pathology in patients with dyshormonogenesis, we recommend a regular ultrasound follow-up at least every 3 years, and a systematic genetic analysis at diagnosis to identify possible pathogenic variants.