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Case Report

Persistent Hypothyroidism Syndrome in a Teenager Followed up for Athyreosis

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Abstract

Congenital Hypothyroidism is a rare but serious malformative pathology, subject to congenital screening in several countries around the world. its impact once omitted is serious and irreversible. The form with agenesis of the thyroid gland called athyreosis can be associated with several malformations, cardiac, renal or skeletal. Patients living with athyreosis most often have symptoms of chronic hypothyroidism, which can be attributed either to a FT3/F4 ratio that is too low or a defect in the peripheral deiodation of T4 to T3. We report the case of a young girl with athyreosis who maintains a refractory hypothyroidism syndrome.

Keywords: Congenital hypothyroidism- Athyreosis– Malformations -Hypothyroidism syndrome.

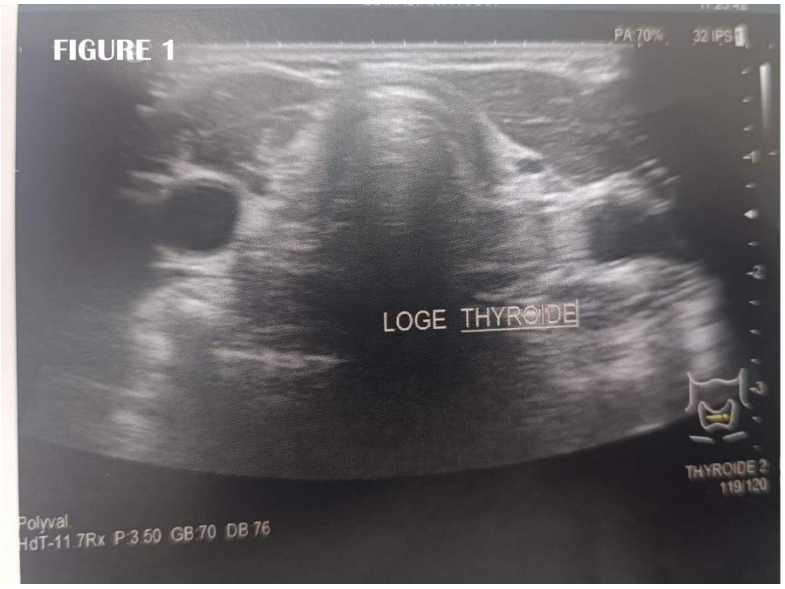
INTRODUCTION

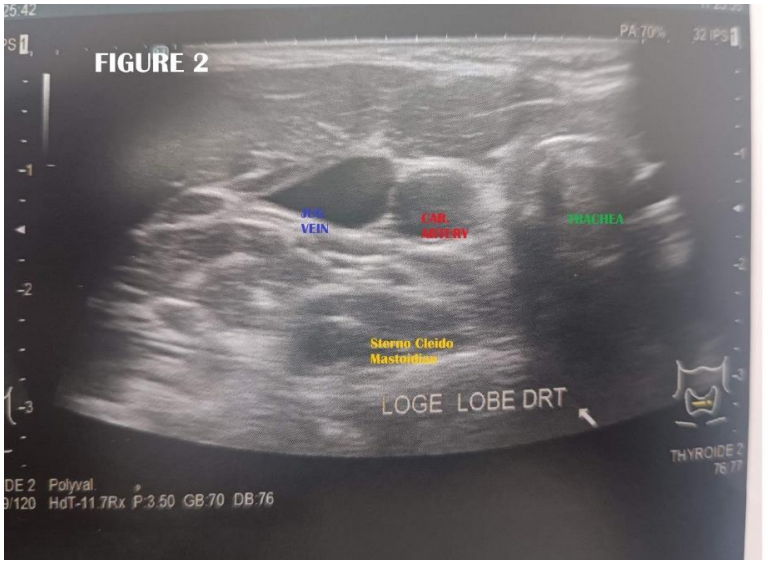
Congenital hypothyroidism (CH) is the most frequent neonatal endocrine disorder [1], and, historically, thyroid dysgenesis was thought to account for approximately 80% of cases [2]. Congenital hypothyroidism (CH) occurs in one out of every 2500 to 1 of 4000 neonates, but the incidence varies by geographic location and by ethnicity [3,4]. The cardiac and musculoskeletal anomalies were the most frequent malformations associated with congenital hypothyroidism in the Egyptian population [5]. Levothyroxine monotherapy is the treatment of choice for hypothyroid patients because peripheral T4 to T3 conversion is believed to account for the overall tissue requirement for thyroid hormones. However, there are indirect evidences that this may not be the case in all patients. [6].

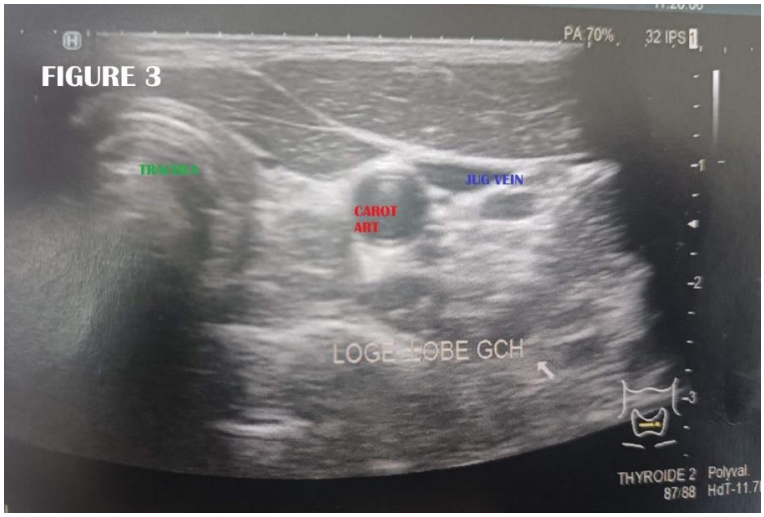
CLINICAL CASE

She is a 16-year-old teenager, followed since the age of 4 months for congenital hypothyroidism. Disease declared in the presence of poor weight development, constipation, severe delay in psychomotor acquisitions, sub-icterus and seizures (according to family). She was put under levothyroxine at the age of 04 months and underwent additional examinations with the aim of detecting other malformations, which returned negative. The family reports the performance of many cervical ultrasound examinations in childhood having not shown thyroid tissue but not reporting the performance of a scintigraphy. The functional signs reported by the young girl and her family are numerous and worrying: permanent asthenia, difficulty in acquiring and school retardation, a hypo tinnitus, and delayed puberty. The physical examination found a patient at the beginning of her puberty; Her Tanner score is B2H3, without obvious dysmorphic syndrome. She is thin P = 39 Kg, BMI = 16.44 KG/m, T = 1.54m (-2 DS relative to the population and -1.5 relative to her target heigh). Biological exploration finds the following: TSH = 7.27, FT4 = 13 pmol/L (N: 12-22); under 75 ug/day of Levothyroxine (1.92 ug/kg/day) but with poor adherence. Collapsed thyroglobulin = 0.1 ng/mL in favor of the absence of thyroid tissue. 17-Beta estradiol = 33 pmol/L FSH = 5.8 LH = 3.9, Prolactin = 17 ng/mL normal, Phosphocalcic assessment finds a profile of secondary hyperparathyroidism PTH = 97 pg/mL with Ca++ normal at 95 mg/L and Vitamin D low = 12 ng/mL. moreover NFS, Ferritin, normal liver and kidney functions. Morphological exploration finds 2 empty left and right thyroid chambers, the thymus was easily objectified on the lower median sections, appears homogeneous without ectopic visible thyroid tissue at this level (see figures from 1 to 4). The thyroid scintigraphy finds a white map confirming a form of athyreosis CH (see figure 5). In view of the delayed puberty associated with short stature, a karyotype was requested that returned to normal 46 XX (image 6). The trans-thoracic ETT ultrasound as well as the renal

ultrasound were requested and returned to normal without cardiac or renal malformative syndrome. The exploration of deafness has been requested and is ongoing.







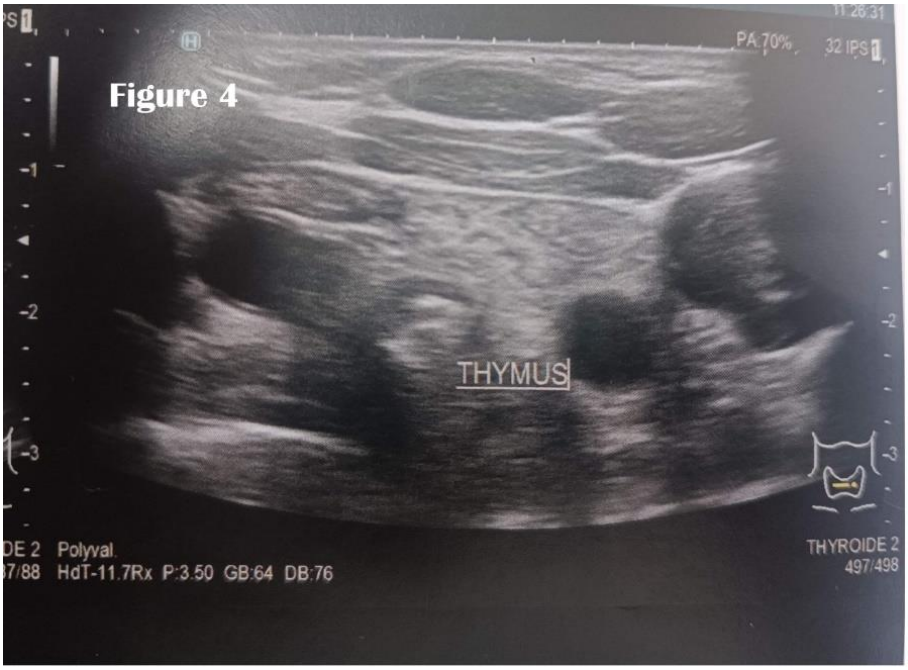


Figure 4
PA:70%
32 IPS
THYMUS
THYROIDE 2
497/498
Polyval
HdT-11.7Rx P:3.50 GB:64 DB:76

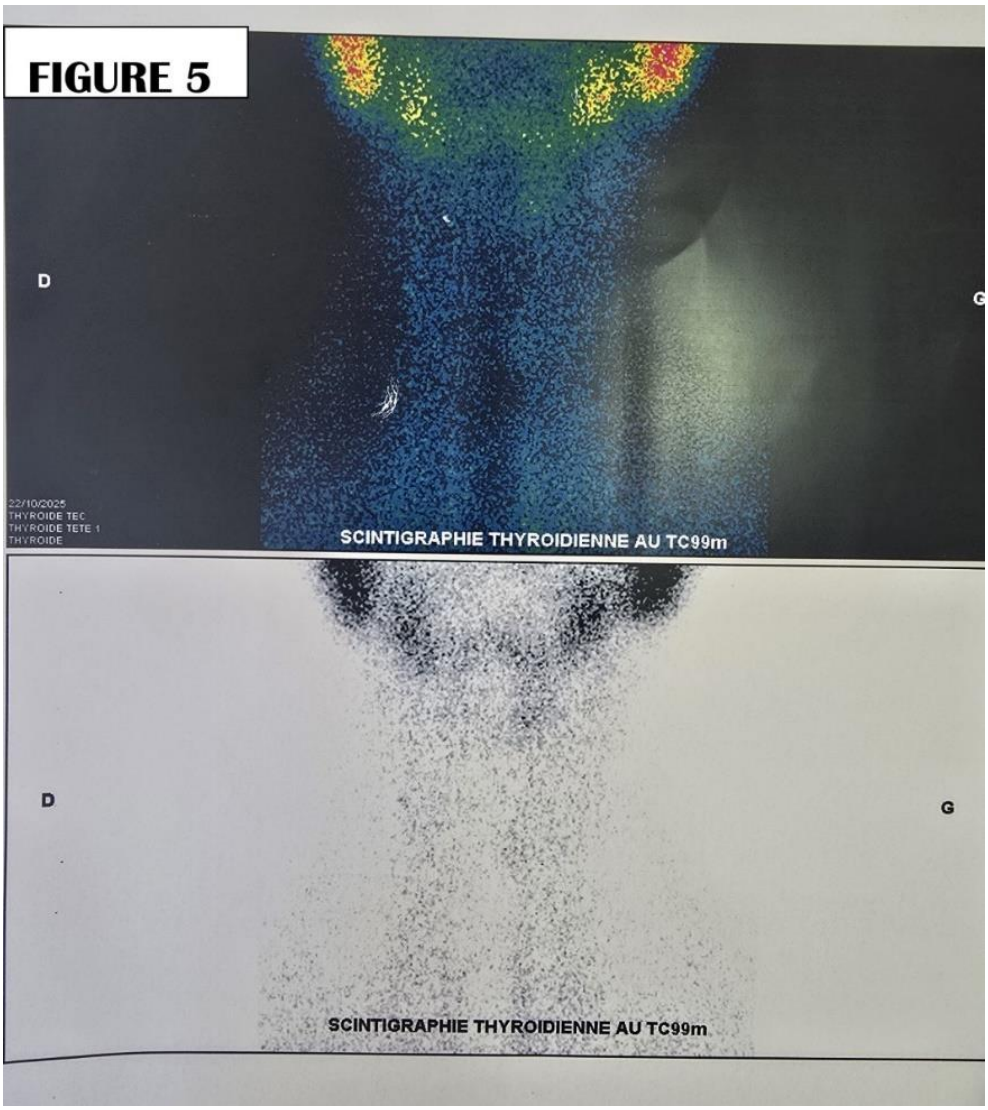


FIGURE 5
D
G
SCINTIGRAPHIE THYROIDIENNE AU TC99m
D
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SCINTIGRAPHIE THYROIDIENNE AU TC99m

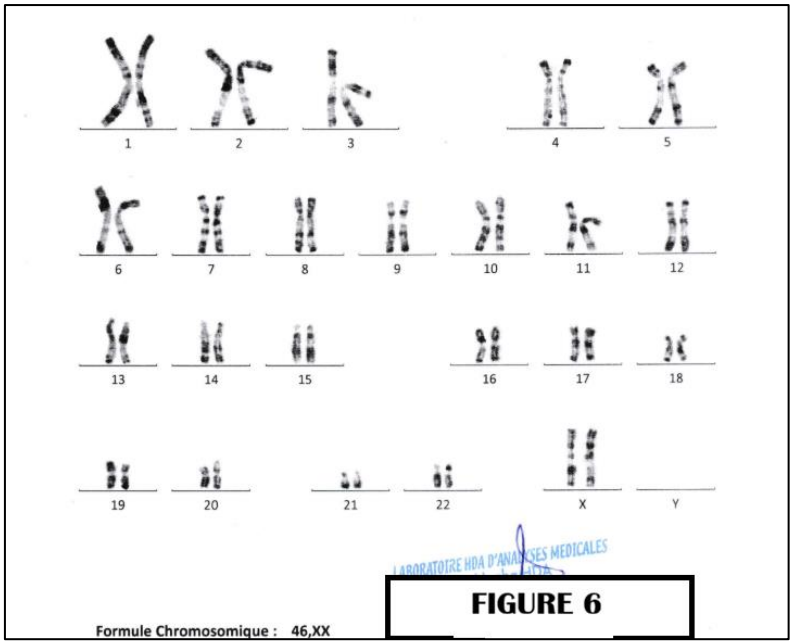

FIGURE 6

DISCUSSION

In the last two decades, the incidence of CH has doubled according to several publications [7]. A possible explanation for the increase in CH incidence is the change in the screening strategy, notably with the revision of TSH values used as a cut-off, which has significantly increased the sensitivity of the screening test [7]. The diagnosis of CH is therefore considered in all newborns with abnormal screening results. They should undergo serum FT4, FT3 and TSH measurements to confirm the findings: newborns with hypothyroidism typically have low FT4 and high TSH concentrations [8]. the diagnosis of congenital hypothyroidism involves an adequate etiological diagnosis (with in situ gland or athyreosis) and to determine the genetic syndrome responsible. and as well determine the possible malformations that can coincide and this pathology.

Malformations screening in athyreosis

In an Iranian cohort, a total of 150 infants with biochemically confirmed primary congenital hypothyroidism (72 females and 78 males) were recruited during the period between May 2006-2010. Overall, 30 (20%) infants had associated congenital anomalies. The most common type of anomaly was Down syndrome. Seven infants (3.1%) had congenital cardiac anomalies such as: ASD (n=3), VSD (n=2), PS (n =1), PDA (n=1). Three children (2.6%) had developmental dysplasia of the hip (n=3) [9]. In another retrospective study of 1,520 patients with CH, Gu *et al.,* observed that the incidences of extrathyroidal congenital anomalies (14.6%) and DS (5.7%) were significantly higher among the CH patients than among the general population [10]. Otherwise, Kreisner *et al.,* Carried out a prospective clinic-based study for evaluation of major congenital malformations in CH infants from Brazil. Among the 76 patients identified, 10 (13.2%) had major congenital malformations of whom 8 (10.5% of the total) had cardiac malformations [11]. in another experience driven by Stoll *et al.,* among 129 CH infants, 15.5% showed additional anomalies of whom 6.9% had congenital cardiac anomalies [12]. Let's return to the case of our patient, our so exhaustive report could not highlight an obvious malformative syndrome, but the clinical deafness she presents could be related to a congenital hearing disorder

Persistent hypothyroidism syndrome in athyreosis and its management

The decreased FT3/FT4 ratio is more marked in a subset of patients (approximately one third in our study) that do not reach a serum FT3/FT4 ratio within the reference range observed in euthyroid controls [13]. Analyzing the correlation between TSH and thyroid hormones serum levels in a large series of athyreotic patients under levothyroxine monotherapy, we observe that in these patients the pituitary response is significantly different from normal, being the pituitary feedback much less sensitive than in euthyroid controls [8]. This condition is more frequent in female and aged patients, indicating a significant gender and age influence on the individual capacity to produce T3 from exogenous levothyroxine. [14,15]

Athyreotic individuals show a relevant heterogeneity in their capacity to deiodinate exogenous levothyroxine to T3 as indicated by the variability of FT3 serum concentrations in patients with similar FT4 levels. While most patients under levothyroxine monotherapy will reach normal TSH serum levels, a normal FT3/FT4 ratio and also full improvement of signs and symptoms, a subset of patients is unable to convert the ingested levothyroxine into an adequate amount of T3. This may happen for a variety of reasons, including congenital or acquired deficiency of deiodinase function. [16,17]

These two hypotheses (FT3/FT4 ratio and exogenous Levothyroxin deiodation) may be the key to understanding the persistence of chronic asthenia syndrome observed in our patient. But the non-availability of T3 in Morocco did not allow us to try this therapeutic alternative of putting the young patient on a combination of T3 T4.

CONCLUSION

The management of HC is a long battle that has the essence of its importance in neonatal screening. Unfortunately, this procedure continues to be absent in our country, and as a result we continue to face handicapping forms in young children and adolescents

Abbreviation:
ASD: Atrial Septal Defect
CH: Congenital hypothyroidism
PDA: Patent ductus arteriosus
TSH: Thyroid stimulating hormone
VSD: Ventricular Septal Defect

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