

Introduction:

Thyroid dysmorphogenesis (TD) accounts for 15–20 % of congenital hypothyroidism (CH). Levothyroxine is the treatment of choice with the immediate goal to raise the serum T4 as rapidly as possible. Surgical treatment is indicated in case of compressive or suspected goiter.

Methods:

Seventeen patients with thyroid dysmorphogenesis belonging to four Tunisian families were included in a descriptive prospective study. They were followed up for a period of 12 years.

Results:

*11 males and 6 females.

***Average age at diagnosis:** 6.97 years (range: 1 month-30 years).

***Diagnosis of thyroid dysmorphogenesis :**

-developing clinical signs of hypothyroidism for 11 patients (64%),

-family screening for 4 patients (32%)

-exploration of goiter for 2 patients(14%).

*6 patients had goiter at diagnosis.

***Treatment with levothyroxine** has been decided for all our patients after confirmation of hypothyroidism.

-The **average dose of L-thyroxine**, evaluated in 13 patients, was 134.37 ug / day with extremes of 50 and 200 ug.

-**Adherence** was evaluated good for 6 patients (35%) and poor for 9 patients (52%). 2 patients were not evaluated. -

Normal thyroid function was obtained for 86% of patients.

-Under treatment, 7 patients have had developed multinodular goiter. An increase of goiter volume was observed for 4 patients among 6 patients who initially presented goiter.

***Total thyroidectomy** was decided secondarily for 2 patients after 27 and 34 years of diagnosing thyroid dysmorphogenesis. This thyroidectomy has been indicated for a compressive goiter for one patient and for a suspected nodules for the second. The histopathologic findings were in favor of benignity.

Discussion:

*The active thyroid hormone tri-iodothyronine (T3) is essential for a normal development of children.

*Especially within the first years of life, thyroid hormone is pivotal in enabling maturation of complex brain function and somatic growth.

*The most compelling example for a life without thyroid hormone are those historical cases of children who came to birth without a thyroid gland as shown in autopsy-studies- and who suffered from untreated hypothyroidism, at that time initially called “sporadic congenital hypothyroidism” (CH).

*In the last decades huge achievements resulted in a normal development of these children based on newborn screening programs that enable an early onset of a high dose LT4-treatment. Unfortunately, this is not the case in Tunisia.

*Further progress will be necessary to further tailor an individualized thyroid hormone substitution approach and to identify those more complex patients with congenital hypothyroidism and associated defects, who will not benefit from an even optimized LT4 therapy.

*Besides the primary production of thyroid hormone a variety of further mechanisms are necessary to mediate the function of T3 on normal development that are located downstream of thyroid hormone production.

Conclusion:

The absence of neonatal screening of congenital hypothyroidism in Tunisia is responsible of diagnostic and therapeutic important delay.

Bibliographie:

- 1-Gruters A, Krude H. Detection and treatment of congenital hypothyroidism. Nat Rev Endocrinol 2012;8:104e13.
- 2-Man EB, Mermann AC, Cooke RE. The development of children with congenital hypothyroidism. A note on early, temporary replacement therapy for 2 goitrous infants. J Pediatr 1963;63:926e41.
- 3-Klein AH, Agustin AV, Foley Jr TP. Successful laboratory screening for congenital hypothyroidism. Lancet 1974;2:77e9.
- 4-Glorieux J, Desjardins M, Letarte J, et al. Useful parameters to predict the eventual mental outcome of hypothyroid children. Pediatr Res 1988;24:6e8.
- 5-Rovet JF. Children with congenital hypothyroidism and their siblings: do they really differ? Pediatrics 2005;115:e52e7.

