

Case Report

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Cretinism: Case report

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Abstract

Congenital hypothyroidism is the most common congenital endocrinopathy; it is also the leading cause of preventable mental retardation and has severe socioeconomical consequences. We report the case of a 10 year-old girl admitted for a typical clinical presentation of hypothyroidism with cretinism and dwarfism (height at 78 cm). Treatment was started immediately with levothyroxine.

Neonatal screening for this disease should be generalized in our country, not only because it is a cause of avoidable mental retardation, but also because this would be one of the most cost-effective secondary prevention programs.

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Introduction

Congenital hypothyroidism is a rare, but serious disease. It is one of the causes of mental retardation in children and has heavy socioeconomical consequences. We must address the needs of infants in nonaffluent parts of the world where congenital hypothyroidism of all etiologies, including iodine deficiency, is still a major health problem. Cretinism is an untreated congenital hypothyroidism, regardless of the presence or not of a goiter. Iodine deficiency is still a major cause of endemic cretinism. In most Western countries, neonatal screening programs have successfully been in place for over 30 years.

Objectives

We report a historical case of cretinism, with a literature review, to recall the genetic, pathogenic, clinical, and therapeutic characteristics of this disease.

Case report

We report the case of a 10 year-old girl, who has been brought for psychological and developmental retardation with failure to thrive. Upon examination, she had generalized hypotonia, umbilical hernia, and constipation (Figure 1). The weight was 11 kg, the waist perimeter was 77 cm, and the head circumference was 47 cm (dwarfism).

The biological analysis confirmed the diagnosis, the Thyroid Stimulating Hormone (TSH) dosage was 218 pUI / ml, with Triiodothyronine (T3) and Thyroxine (T4) collapse.

Treatment was started immediately with levothyroxine sodium. The evolution was marked by the partial recovery of the tone and melting of the myxedema.



Figure 1

Discussion

Hypothyroidism, or thyroid insufficiency, is a metabolic disease that is characterized by the absence or insufficient production of Thyroid Hormones (T3 and T4).

In children, these hormones play a major role in brain growth and differentiation, the neonatal onset in severe congenital forms, associated with late diagnosis, is responsible for severe mental retardation: Cretinism [1].

Infantile hypothyroidism was described by Thomas Curling in 1850, who reported two cases of absence of the thyroid gland in children with “sporadic cretinism” [2].

This justifies the major interest of systematic screening for congenital hypothyroidism in all newborns to initiate an early dose-adapted treatment to allow the child to develop all his intellectual potential and avoid mental delay [3].

The symptoms of congenital hypothyroidism are not specific or often absent in the neonatal period and early childhood. Therefore, it is difficult to identify this disease based only on clinical symptoms, thus, it is often under diagnosed. Indeed, based solely on clinical data, the syndrome is recognized in only 10% of infants in the first month, 35% in the first three months, 70% in the first year, and 100% at the age of 3-4 years [4]. In advanced stages, the most frequent complaint is psychomotor retardation.

Signs and symptoms of congenital hypothyroidism vary depending on the cause, severity, and duration of thyroid hormone deficiency during the prenatal period [5].

The delay in bone maturation is almost constant and intense.

Thyroid ultrasound is the best examination to assess the size and volume of the gland by assessing the transverse and longitudinal measurements of each lobe and isthmus. This examination has been established for two decades as the morphological examination of reference [6].

Levothyroxine is the treatment of choice. In drops, it has a better bioavailability and allows lower initial doses [7].

Several prognostic factors determine mental development:

- The severity of hypothyroidism with very low levels of thyroxine and a significant delay in bone maturation at birth.
- Quality of medical care: The dosage of levothyroxine and the early start of hormone therapy.
- The quality of hormonal control during the first year of treatment.
- And the socio-economic level of the family [8].

The neonatal diagnosis and treatment of congenital hypothyroidism prevents severe intellectual delay. In Morocco, neonatal screening was implemented in April 2012 but only in Rabat Salé, Zemmour Zair region. The challenge is to generalize this screening program to all regions of the country given the heavy consequences of its late diagnosis.

Conclusion

Cretinism is a serious affection. If not diagnosed early and treated correctly, it inevitably causes heavy repercussions. A generalized neonatal screen in Morocco is crucial to prevent neuro-developmental and socioeconomical consequences by providing an early diagnosis and management.

Conflict of interest: The author declares that no conflict of interests.

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