

Congenital Hypothyroidism By Athyreosis: About A New Case From The Dysmorphology, Congenital And Developmental Anomalies Unit

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ABSTRACT

The objective of this work is to contribute to the promotion of neonatal screening for congenital hypothyroidism in our daily practice. In the Kingdom of Morocco, neonatal screening is in the process of generalization, but some newborns escape this screening activity. The authors report the case of a female infant aged 14 months, resulting from a pregnancy followed and carried to term in a maternity hospital. She presented with a delay of psychomotor acquisitions with chronic constipation, macroglossia and respiratory difficulties. The clinical examination

found a staturoponderal delay, facial puffiness, a hoarseness of the voice, an umbilical hernia and a psychomotor delay concerning the holding of the head and the sitting position. The biological assessment showed a deep peripheral hypothyroidism. The cervical ultrasound showed the absence of thyroid gland visualization. Thyroid scintigraphy with Tc99m showed the total absence of fixation of the tracer at the level of the thyroid lodge, absence of ectopic focus at the level of the axis of the thyroglossal tract. The bone age was in favor of the bone age of a newborn. Through this case, the authors underline the importance of carrying out neonatal screening for congenital hypothyroidism by all physician taking care all newborns while awaiting the generalization throughout the Morocco. All this, to avoid the serious and avoidable complications of this disease.

Keywords : congenital hypothyroidism, screening ,intellectual disability

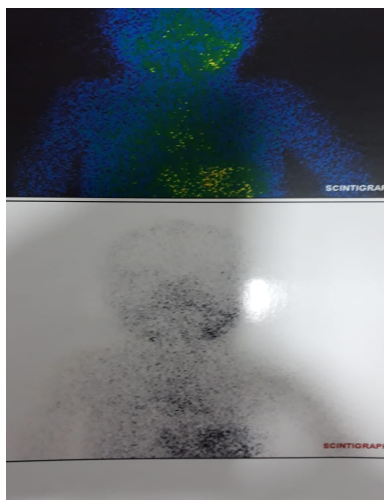
INTRODUCTION

Congenital hypothyroidism, is the most common preventable causes of intellectual disability with an incidence of 1:2000 to 1:4000, [1]. The first screening experiments carried out since the 1970s in Canada [2], the United States of America[3] and Europe [4], had shown the effectiveness of neonatal screening by measuring TSH (thyroid-stimulating hormone - thyreostimuline) and/or T4 (thyroxine). In fact, the screening did not eliminate the disease, but it did change the expression of the disease.[5]. In Morocco, we do not have data on the frequency of congenital hypothyroidism, and little of studies have taken an interest in this subject. The first neonatal hypothyroidism screening campaign congenital (DNHC) which took place in Rabat between 1995 and 1997 was not completed due to various problems: it only affected 3,300 newborns instead of the 15,000 initially planned. [6].

CAS REPPORT

Female infant, 14 months old, 4th of 4 siblings, without any particular antecedents, presenting at 8 months of age a delay in psychomotor acquisition, generalised oedemas and chronic constipation, evolving in a context of apyrexia, the clinical examination found a staturoweight delay: 8,5 (-3DS) Height : 63 cm (-3DS) Pc : 43 cm ,puffiness of the face, hoarseness of the voice, umbilical hernia and a psychomotor delay concerning the sitting position and the holding of the head. The biological assessment showed a deep peripheral hypothyroidism: TSH us : 100 uIU / ml, FT3: 1pg /ml, FT4: 0,4ng/dl, cervical ultrasound showed the absence of thyroid gland visualisation. Thyroid scintigraphy with Tc99m (before treatment) showed the total absence of tracer fixation in the thyroid lodge, absence of ectopic focus in the axis of the thyroid tract. The osseous age was in favour at the osseous age of a newborn baby, the treatment with Levothyroxine cp: 75ug/m2/d was started with biological monitoring of T4 levels. The evolution was marked on the clinical level by the acquisition of the sitting position, the decrease of facial puffiness, the regression of edemas, macroglossia, aquisitions of sitting and walking, a near normal weight recovery at the age of 4 years, a weight of 17kg, a height of 99cm.

On the biological level by the increase of T4 levels and the normalisation of TSH.



the scintigraphy of our patient



the image of our patient with hypotonia and puffiness of the face



Our patient at the age of 4 years, under substitution treatment

DISCUSSION

The primary function of the thyroid gland is to produce and secrete thyroid hormones, which exert important physiologic effects throughout the body.[1]. Congenital hypothyroidism (CH) is an inborn condition in which the thyroid gland does not produce sufficient thyroid hormone. The causes of congenital hypothyroidism can be divided into those that directly impair thyroidal synthesis of thyroid hormone and those that disrupt hypothalamic or pituitary control of the thyroid gland by decreasing the secretion or bioactivity of TSH . [1]. The most common cause of primary CH is failure of normal thyroid gland development or failure of a eutopic thyroid gland to produce TH normally . Thyroid dysgenesis accounts for ~85% permanent CH while dyshormonogenesis accounts for the remainder [7]. Despite the frequency of thyroid dysgenesis, and an increasing number of identified causative genes, the majority of cases are sporadic and a genetic etiology is found in only 2–5% [8]. Congenital central hypothyroidism is rare, with an incidence between 1:16,000 and 1:20,000, is usually caused by inborn structural or functional defects that may be associated with mutations in genes involved in hypothalamic and pituitary development [9]. Several extrinsic factors can cause hypothyroidism in neonates. Iodine deficiency is the common cause of neonatal hypothyroidism . [10]. Other extrinsic causes include transplacental passage of antithyroid medications used to treat hyperthyroidism (methimazole, carbimazole, or propylthiouracil), transfer of maternal IgG antibodies that block activation of the TSH receptor. [11].

Infants with severe congenital hypothyroidism may present with hypothermia, bradycardia, poor feeding, hypotonia, large fontanelles, myxedema, macroglossia, and umbilical hernia. [12]. thyroid hormone has important effects on many organ systems aside from its crucial role in neurodevelopment and growth. In the cardiovascular system, thyroid hormone increases cardiac rate contractility, output and decreases systemic vascular resistance . It also promotes renal excretion of salt and water, stimulates gastrointestinal motility, and increases the basal metabolic rate and body temperature [1]. it is important to recognize that these symptoms and signs are non specific and can be observed in many other neonatal illnesses.[1]. However, many neonates manifest few or no symptoms even with significant hypothyroidism, making clinical diagnosis difficult in this age group. [12]. which reinforces the central role of universal newborn screening in facilitating diagnosis and treatment. Specific screening practices in general begin in the first few days of life with collection of a heel-prick blood sample on a filter paper card that is sent to a central laboratory for analysis. Most screening programs around the world measure the blood concentration of TSH; in some programs, T4 is also measured routinely, or by reflex if the TSH is elevated [13]. The optimal time to collect a screening blood is between 48 and 72 hours of life [14]. If the newborn screening TSH is greater than 40 mIU/L, severe hypothyroidism presumed and treatment should be initiated as confirmatory serum laboratory tests are drawn. Once confirmatory measurements are available, treatment should be initiated in patient with a serum TSH of greater than 20 mIU/L, or with a low FT4 concentration regardless of TSH concentration [1]. Infants with a slight elevation of TSH and normal FT4 levels have mild hypothyroidism, and current data are indeterminate as to the neurodevelopmental risks posed by this degree of hypothyroidism [15]. In infants with primary hypothyroidism Thyroid imaging (either by ultrasound or scintigraphy) may establish a diagnosis of thyroid dysgenesis, which is likely to be permanent and therefore requires treatment even if mild. In contrast, the presence of a eutopic thyroid gland suggests the possibility of transient hypothyroidism.[16].Measurement of serum thyroglobulin levels provide useful information about the etiology of congenital hypothyroidism. When measured at a time of elevated TSH

concentrations, a low level of thyroglobulin indicates reduced thyroid mass, a rare mutation of the thyroglobulin gene, or impaired function of the TSH receptor itself, which may be caused by genetic defects in TSH receptor signaling or by maternal TSH receptor-blocking antibodies.[1]. the presence of a normal thyroid gland on anatomic imaging (ultrasound) combined with a low serum thyroglobulin concentration should prompt testing for TSH receptor antibodies.[11].

Levothyroxine (LT4) is the treatment for congenital hypothyroidism. The recommended starting dose is 10 to 15 mg/kg. Late initiation of treatment and lower initial doses of LT4 are associated with poorer neurodevelopmental outcomes [1]. The objective of treatment is to achieve euthyroidism rapidly and to maintain it as consistently as possible thereafter. Normalization of serum TSH and FT4 levels within 2 weeks after starting therapy seems to improve cognitive outcomes.[1]. Treatment of congenital hypothyroidism is controlled by serum TSH and FT4 measurements. These should be checked 1 to 2 weeks after starting treatment and every 2 to 4 weeks thereafter, with appropriate adjustment of LT4 dose, until they have normalized.[1]

our patient presents a congenital hypothyroidism by thyroid agenesis diagnosed late after the installation of psychomotor retardation and staturponderal retard, but under levothyroxine at adapted dose, the evolution was clinical improvement in the development of psychomotor and height-weight acquisitions.

CONCLUSION

Through this observation, the authors underline the importance of carrying out neonatal screening for congenital hypothyroidism by all physician taking care the newborns while awaiting the generalization throughout the Morocco. All this, to avoid the serious and avoidable complications of this disease.

Indeed, the congenital hypothyroidism is common and can cause severe neurodevelopmental morbidity. Early diagnosis and initiation of appropriate treatment are important to prevent these adverse effects and optimise long-term outcomes. Newborn screening is an important tool for the detection of congenital hypothyroidism and has led to a dramatic reduction in severe intellectual disability owing to this disease.

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