

CONGENITAL HYPOTHYROIDISM IN AL-KADHIYMA TEACHING HOSPITAL, BAGHDAD-IRAQ

Nasheit A. Nasheit¹ *CABP*, Husam Al-alwani¹ *CBAP*, Iman S. Mahmoud²

Abstract

Background: Congenital hypothyroidism is one of the most encountered endocrine diseases in childhood and the importance of this diagnosis is because early and adequate treatment prevents developmental retardation and other sequelae of the condition.

Objective: Looking for the congenital hyperthyroidism in Iraqi children.

Methods: A retrospective study involved 40 children who were attended Pediatric Endocrine and diabetic Clinic for children in Al-Kadhiymia Teaching Hospital, College of Medicine, Al-Nahrain University Baghdad, Iraq, over period from Jan 1993-Jan 2003 .

Results: Of the 40 patients, 24 were female and 16 male. Female to male ratio 1.5:1. A history of parental consanguinity was positive in 36(80%) of the patients and 25(62.5%) had family history of hypothyroidism, and 24(60%) of the patients were

from urban area 16(40%) from rural area west to Baghdad. Ten children (25%) detected in first month of life, 15(37.5%) in first 3 months and 25(62.5%) within the first 6 months. Seven patients had aplasia of thyroid gland, five had ectopia, and 8 had thyroid in normal position . Developmental assessment performed on all patients, also clinical manifestation in 20 patients diagnosed before 6 months of life.

Conclusion: Although we do not know much about the overall prevalence of congenital hypothyroidism in Iraq, the first observation to make is that it is not a rare disease in this country. A genetic explanation suggested by the involvement of multiple siblings and the high rate of consanguinity in this population.

Key Words: Congenital hypothyroidism, Retrospective

IRAQI J MED SCI, 2005; Vol. 4 (1): 89-92

Introduction

Congenital hypothyroidism (CH) results from deficient production of thyroid hormone or defect in thyroid hormone receptor activity. Congenital causes of hypothyroidism may be sporadic or familial, goitrous or non-goitrous. In many cases the deficiency of the thyroid hormone is severe and symptoms develop in early weeks of life, in others lesser degree of deficiency occur or

manifestations may be delayed for months^[1].

Early treatment of CH particularly within first 3 months of life usually results in normal psychomotor development since the mass neonatal thyroid screening programmes have been introduced in many industrialized countries aiming at prevention of mental retardation by early diagnosis of CH and prompt thyroid replacement therapy^[2,3]. However, in most of the developing countries the diagnosis of CH still based on clinical grounds. This may result in a delay in the diagnosis and the initiation of thyroid replacement therapy with consequent mental retardation.

¹ Dept. Pediatrics, College of Medicine Al-Nahrain University ² Central Arab Child Hospital

Address correspondence to Dr. Nasheit A. Nasheit e-mail: naazna2003@yahoo.com
Received 5th December 2004: Accepted 14th February 2005.

Patients & Methods

This retrospective study included children who were attended Pediatric Endocrine and Diabetes Clinic in Al-Kadhiymia Teaching Hospital, Baghdad-Iraq, over period of 10 years from Jan. 1993-Jan. 2003. They were clinically diagnosed as having CH. Data recorded includes sex, age ,residency, parental consanguinity, family history of thyroid disease, pregnancy history, place of birth, postnatal fallow up, clinical manifestation, age at diagnosis and treatment, and assessment of mental development by Stanford-Binet scale.

Diagnosis of CH was confirmed by thyroid function tests (T4, T3, TSH and TC99M thyroid scan), the values considered according to the age (some investigation performed out side Iraq Perchlorate discharge test). Skeletal maturity was assessed by X-ray of the knee for infants and other joints for older children. Thyroid function tests made by radioimmunoassay methods using commercially available kits.

Results

Forty Patients with CH, Twenty-four (60%) from urban area and 16(40%) from

rural area west to Baghdad. The female to male (24/16) ratio was 1.5:1.

A history of parental consanguinity was positive in 36(80%) of the patients and 25(62.5%) had family history of thyroid disease. In seven families, multiple siblings affected and in none of the patients was maternal history of ingestion of any medication during pregnancy.

Eleven children (27.5%) were detected in first month of life, at age from 1-3 months 6 (15%) and at age 4-6 months 5(12.5%), at age 7-12 months 3(7.5%), and nine (22.5%) at age from 1-3 years and six (15%) were diagnosed after age of 3 years (Table 1).

TC-99M thyroid scan was performed for 20 patients, seven had no thyroid gland (aplasia), five had ectopia and 8 had thyroid gland in normal position (Table 1). The mean period of fallow up was 2.1 year (range 6 months- 5.5 year). Development assessment performed on all patients: Ten (25%) had moderately severe mental retardation (IQ more than 50), Sixteen children (40%) achieved normal psychomotor development and 14 patients (35%) had mild mental retardation (IQ 50-75; Table-1).

Table 1: Ages at diagnosis, Sex and the development outcome

Age of diagnosis	No.	%	Development outcome			Findings on (TC-99M)scan of the thyroid gland N = 20		
			Normal	Mild mental retardation (IQ 50-75)	Moderately sever mental retardation (IQ<50)	Normal position	Ectopic gland	Aplosia
< 1 month	11	27.5	8	3	-	1	1	2
1-3 months	6	15	4	2	-	1	1	2
4-6 months	5	12.5	3	1	1	-	1	2
7-12 months	3	7.5	2	1	-	2	-	1
1-3 years	9	22.5	4	3	2	2	-	1
>3 years	6	15	-	1	5	-	1	2
Total	40	100	21 (52.5%)	11 (27.5%)	8 (20%)	6 (30%)	4 (20%)	10 (50%)
			40 (100%)			20		

The clinical manifestation in 20 patients with congenital hypothyroidism diagnosed before 6 months (Table-2).

Table 2: Clinical manifestation in 22 patients with congenital hypothyroidism diagnosed before the age of 6 months

Manifestations	No.	%
Coarse faces	18	81.8
Delayed bone maturation	16	72.7
Constipation	15	68.2
Neonatal Jaundice	12	54.5
Umbilical hernia	11	50
Hoarse cry	8	36.3
Large anterior fontanel	8	36.3
Hypothermia	6	27.2
Macroglossia	6	27.2
Skin mottling	3	13.6

Discussion

Neonatal screening programmes for congenital hypothyroidism have revealed a variable incidence. In Iraq the incidence is not known, but in USA and other countries (1/2500)^[1,4,5].

Although we do not know much about the overall prevalence of CH in Iraq, the first observation to be made is that this is not a rare disease in this country.

Congenital hypothyroidism can be due to absent or hypoplastic gland 35%, an ectopic gland 43%, or inborn error of metabolism of thyroid hormone 22%⁶, while in our series the percentage of ectopic gland is 20%, aplasia is 50% and normal position 30%. Determination of the cause of CH has genetic, epidemiological and prognostic importance^[7,8]. The overall sex distributions showed the same pattern of female predominance reported elsewhere^[1,9,10], while there was even sex distribution in infants with thyroid aplasia or ectopia presented in the first 6 months of life, similar to the experience of Daoud et al^[9]. The most frequent initial clinical

manifestation in 22 patients presented in the first months of life was coarse facial features, constipation, prolonged neonatal jaundice, umbilical hernia and delayed skeletal maturation (Table-2) which is similar to those reported by Smith^[6,12], while in older age group the main reason for referral was psychomotor retardation, constipation and short stature. Though many of these features are rather non-specific, it is important to notice that most of the hypothyroid infants had several of these features at a time. Forty-two percent of our patients detected in the first 3 months (62.5%) in the first year. These results are similar to those reported by others^[11,12].

Children with thyroid gland aplasia or ectopia might present early in life. Nine out of eleven of our patients with either thyroid aplasia or ectopia presented within the first 6 months of life.

Among 21 children who achieved normal mental and motor development, thirteen received thyroid replacement therapy within the first 6 months of life, and seven of the eight patients with moderately severe mental retardation were diagnosed and received therapy after the age of 1 year (Table-1). This demonstrates the favorable effect of early treatment, which has been confirmed by recent prospective studies in children with CH detected by neonatal thyroid screening^[13].

Conclusion

These results demonstrate that CH is not uncommon disease, which is probably due to high rate of consanguinity among our population, and many thyroid investigations if needed are necessary to identify the cause of CH, which has genetic, epidemiology and prognostic importance.

Recommendation

Because of the sequelae of untreated CH are so severe and because the benefits of early treatment have been well-documented, either or national screening programme should keep in consideration the characteristic of neonatal practice in Iraq to achieve the maximum diagnostic benefits.

General practitioners and pediatricians should be aware of this disease and treat such patients early enough to minimize the unwanted sequelae.

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