



Knowledge about Complication of Congenital Hypothyroidism among Parents in Aseer Region, Saudi Arabia

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Authors' contributions

This work was carried out in collaboration among all authors. All authors read and approved the final manuscript.

Article Information

DOI: 10.9734/JPRI/2021/v33i53B35779

Open Peer Review History:

This journal follows the Advanced Open Peer Review policy. Identity of the Reviewers, Editor(s) and additional Reviewers, peer review comments, different versions of the manuscript, comments of the editors, etc are available here: <https://www.sdiarticle5.com/review-history/83730>

Original Research Article

Received 01 November 2021

Accepted 07 December 2021

Published 08 December 2021

ABSTRACT

Background: A lack of thyroid hormone present at birth is described as Congenital Hypothyroidism (CH). A difficulty with thyroid development or thyroid hormone production (dysgenesis) is most frequent in thyroid birth hormone insufficiency. It is one of the main causes of avoidable mental delay in infants This study aims to assess knowledge and awareness of Saudi parents towards complication of congenital hypothyroidism in Aseer region, KSA.

Methods: A cross sectional study was conducted of general population of Aseer region. Data was collected by means of online questionnaire distributed online to be self-filled by parents. The study included 1086 participants. The collected data was entered and analyzed using the Statistical

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Package for the Social Science (SPSS Inc. Chicago, IL, USA) version 23. Descriptive statistics was performed.

Results: All participants were from Aseer region, 39.5% of participants reported consanguinity between parents, 34.5% of all participants had heard of CH. 18.2% identified iodine deficiency during pregnancy as risk factor for CH. 22.6% identified excessive sleeping, 15.2% jaundice and 16% facial swelling. 60.2% did not know CH complications while 22.1% reported poor growth, resulting in short stature as a complication, followed by 14.4% Goiter, 14.1% delayed puberty, 13.5% mental retardation, 12.9% osteoporosis, 9.9% heart defects, and 4.6% bone fractures. 11.9% of all participants identified optimal time to start treatment to prevent complications as the first two weeks of the baby's life

Conclusion: Parental knowledge of congenital hypothyroidism in Aseer is relatively unsatisfactory. Health campaigns and conferences to raise awareness is necessary looking to catastrophic complications caused by delayed diagnosis and management of the disease.

Keywords: Congenital hypothyroidism; knowledge; complications CH.

1. INTRODUCTION

A lack of thyroid hormone present at birth is described as Congenital Hypothyroidism (CH). A difficulty with thyroid development or thyroid hormone production (dysgenesis) is most frequent in thyroid birth hormone insufficiency. It is one of the main causes of avoidable mental delay in about 1:2,000 to 1:4,000 infants [1]. The published statistics show comparatively high incident CH in certain Arab nations, including Lebanon (1 in 1823), Bahrain (1, in 2967), Palestine (1 in 2133), Oman (1 in 2200), Saudi Arabia (1 in 2931), and Egypt with comparatively high occurrences of the CH in several Arab nations (Alexandria: 1 in 397412) [2].

CH is classified into permanent and transient forms, which in turn can be divided into primary, secondary, or peripheral etiologies. Thyroid dysgenesis accounts for 85% of permanent, primary CH, while inborn errors of thyroid hormone biosynthesis (dyshormonogeneses) account for 10-15% of cases [3]. Secondary or central CH may occur with isolated TSH deficiency, but more commonly it is associated with congenital hypopituitarism. Transient CH most commonly occurs in preterm infants born in areas of endemic iodine deficiency. Babies in whom severe fetomaternal hypothyroidism was present in utero tend to be the most symptomatic at birth. Similarly, babies with athyreosis or a complete block in thyroid hormonogenesis tend to have more signs and symptoms at birth than infants with an ectopic thyroid, the most common cause of CH [4].

Congenital hypothyroidism is common and can cause severe neurodevelopmental morbidity. The clinical manifestations are often subtle or not

present at birth. Common symptoms include decreased activity and increased sleep, feeding difficulty, constipation, and prolonged jaundice. On examination, common signs include myxedematous facies, large fontanelles, macroglossia, a distended abdomen with umbilical hernia, and hypotonia [5].

Most newborns with congenital hypothyroidism (CH) have no or few clinical manifestations at birth. In countries with newborn screening programs in place, infants with CH are diagnosed after detection by screening tests [6]. There is an increased incidence of other congenital malformations in children with congenital hypothyroidism (CH), particularly cardiac malformations, including septal defects, renal abnormalities, and the risk of neurodevelopmental disorders [7].

Prompt diagnosis and treatment of congenital hypothyroidism (CH) is critical for the optimal neurodevelopmental outcome and requires interprofessional communication, and care coordination by newborn screen laboratory, primary care physicians, and pediatric endocrinologists are important [8]. Careful neurodevelopmental and neurosensory evaluations should be started early in life and repeated at important critical developmental phases, taking into account disease severity at diagnosis and providing appropriate interventions as required. Universal newborn screening is an important tool for detecting congenital hypothyroidism, but awareness of its limitations, repeated screening in high-risk infants, and a high index of clinical suspicion are needed to ensure that all affected infants are appropriately identified and treated [9].

Parents need to be educated on the diagnosis of congenital hypothyroidism (CH), the importance of early and adequate treatment that will prevent poor neurodevelopmental outcomes. Efforts should focus on educating both patients and caregivers to ensure that adequate treatment is continued into adulthood as parental adherence is associated with successful therapy [10]. Therefore, it is important to know the level of information of family members about the possible damage that CH, when not treated early, can cause in the auditory system, for efficient strategies for the prevention and promotion of hearing health, intervention and monitoring [11].

1.1 Aim of the Study

To assess knowledge and awareness of Saudi parents in Aseer region towards complication of congenital hypothyroidism.

2. METHODS & PARTICIPANTS

2.1 Study Design and Setting

A cross sectional study design was adopted.

2.2 Study Area and Setting

The study was carried out in Aseer, Saudi Arabia.

2.3 Study Period

The data was collected during a period of two months from June 1st 2021 to August 31st, 2021.

2.4 Study Population

General population in Aseer was eligible for inclusion in the study, provided they fulfill the inclusion criteria.

Inclusion criteria and Exclusion criteria: The study will include all Saudi parents who can read and write was included in the study. Non-Saudi subjects, adults who don't have children and illiterate subjects was excluded.

Sample size: The minimum sample size for this study has been decided according to Swinscow, as follows:

$$n = Z^2 \times P \times Q/D^2$$

Where,
n: Calculated sample size

Z: The z-value for the selected level of confidence $(1 - \alpha) = 1.96$.

P: An estimated prevalence of having knowledge as 50% since there is no specific figure for that

Q: $(1 - 0.50) = 50\%$, i.e., 0.50

D: The maximum acceptable error = 0.05.

So, the calculated minimum sample size was:

$$n = (1.96)^2 \times 0.50 \times 0.50 / (0.05)^2 = 384$$

2.5 The Sampling Technique

Random sampling technique was adopted to select the sample size.

2.6 Data Collection Tool

A pre-designed questionnaire was used for data collection. Questions regarding socio-demographic characteristics of the participants (age, educational level, occupation, number of children and residence) as well as knowledge about complication of congenital hypothyroidism along with risks and management of it. The questionnaire will include a brief introduction explaining the idea of the research to participants.

2.7 Data Collection Technique

The researchers distributed the questionnaire online as the questionnaire was distributed online on social media sites (WhatsApp- Facebook- Twitter) to be filled out personally. The questionnaire had a brief introduction explaining the nature of the research and confidentiality of the information that given to participants.

2.8 Data Management and Analysis Plan

All data was entered and analyzed using SPSS 23 with using appropriate statistical methods for description and analysis. P-value less than 0.05 was considered for statistical significance.

3. RESULTS

In Table (1); all participants were from Aseer region in KSA, 29.6% of all fathers aged 45 years old or more while 28.7% of mothers aged between 18- 25 years old. 59.1% of both parents were highly educated (University or more). 39.5% of parents reported consanguinity between them and 34.3% had 4 children or more.

As illustrated in Table (2); 34.5% of all participants had heard of CH. 18.2% identified

iodine deficiency during pregnancy as risk factor for CH. 64.1% of participants didn't know the causes of CH, 24.3% reported hormonal disturbance as a cause of CH, 12.2% reported absence of thyroid gland, and 9.9% reported drugs during pregnancy as cause of CH. 21.2% of all participants identified constipation as CH symptom, 22.6% identified excessive sleeping, 15.2% jaundice and 16% facial swelling. 60.2% did not know CH complications while 22.1% reported poor growth, resulting in short stature as a complication, followed by 14.4% Goiter, 14.1% delayed puberty, 13.5% mental retardation, 12.9% osteoporosis, 9.9% heart defects, and 4.6% bone fractures.

According to Table (3); only 4.4% of study participants reported having a child diagnosed with CH. 1.7% reported having 4 children or

more with CH, 1.7% reported 3 children, 0.6% reported 2 children and 1.9% reported having one child with CH. 3% of all study participants reported that doctor gave information about the disease at the time of child's birth.

The most commonly cited source of information was internet social networking site in 31.5% followed by family and friends 13.3%, doctors and nurses 12.2%, and TV or the Radio in 6.6%. only 11.9% of all participants identified optimal time to start treatment to prevent complications as the first two weeks of the baby's life As in Table (4).

Table (5) shows no significant association between neither parents age or consanguinity between parents with occurrence of CH among children.

Table (1). Sociodemographic characteristics, consanguinity between parents and number of children of participants (n=362)

Parameter		No.	Percent
Aseer region	• Yes	362	100.0%
Social status (married or not)	• Yes	355	98.1%
	• No	7	1.9%
Age of Father	• 18 - 25 years old	44	12.2%
	• 26 - 35 years old	135	37.3%
	• 36 - 45 years old	96	21.0%
	• 45 and over	107	29.6%
Age of mother	• 18 - 25 years old	104	28.7%
	• 26 - 35 years old	99	27.3%
	• 36 - 45 years old	74	20.4%
	• 45 and over	118	23.5%
Education level of father	• Illiterate	17	4.7%
	• primary	23	6.4%
	• Intermediate	22	6.1%
	• secondary	86	23.8%
	• University or more	214	59.1%
Education level of Mother	• uneducated	26	7.2%
	• primary	25	6.9%
	• Preparatory	19	5.2%
	• secondary	78	21.5%
	• University or more	214	59.1%
Consanguinity between parents	• Yes	143	39.5%
	• No	219	60.5%
Number of children	• No Children	77	21.3%
	• 1	49	13.5%
	• 2	64	17.7%
	• 3	48	13.3%
	• 4 and more	124	34.3%

Table (2). Knowledge of participants of CH and its risk factors, causes, symptoms and complications (n=362)

Parameter		No.	Percent	
Heard of congenital hypothyroidism	• Yes	125	34.5%	
	• No	237	65.5%	
Risk factors for congenital hypothyroidism	• Iodine deficiency during pregnancy	66	18.2%	
	• Increased iodine during pregnancy	39	10.8%	
	• I do not know	237	71.0%	
Symptoms of congenital hypothyroidism	• constipation	77	21.2%	
	• facial swelling	58	16.0%	
	• dry skin	68	18.8%	
	• Protruding tongue	50	13.8%	
	• Low activity of child	22	6.1%	
	• excessive sweating	47	12.9%	
	• excessive sleeping	82	22.6%	
	• Swelling under a child's jaw	35	9.3%	
	• Yellowing of the skin and whites of the eyes (jaundice)	55	15.2%	
	• Fever	27	7.5%	
	• Insomnia	51	14.1%	
	• I do not know	219	60.5%	
	Complications of the disease in children and adolescents	• Delayed growth of permanent teeth	50	13.8%
		• Poor growth, resulting in short stature	80	22.1%
• Delayed puberty		51	14.1%	
• Mental retardation		49	13.5%	
• Osteoporosis		47	12.9%	
• Goitre		52	14.4%	
• Heart defects		36	9.9%	
• Bone fractures		17	4.6%	
• Thyroid toxicity		37	10.2%	
• Don't know		218	60.2%	
Causes of congenital hypothyroidism		• Hormonal disturbance	88	24.3%
		• Absence of thyroid gland	44	12.2%
		• Birth defect	28	7.7%
	• Complication during pregnancy	27	7.4%	
	• Iodine deficiency	38	10.5%	
	• Hypopituitarism	27	7.5%	
	• Drugs	108	9.9%	
	• Partial formation of the thyroid gland	36	7.5%	
	• Migratory thyroid gland	18	4.9%	
	• Don't know	232	64.1%	

Table (3). Prevalence of diagnosed CH in one or more child of participants family (n= 362)

Parameter		No.	Percent
If participant have a child or more diagnosed with congenital hypothyroidism	• Yes	16	4.4%
	• No	346	95.6%
If the last question is yes, had doctor gave information about the disease at the time of child's birth	• Yes	11	3.0%
	• No	36	9.9%
	• Don't have children with CH	315	87.0%

Table (4). Source of information of participants about CH and other knowledge determinants (n= 362)

Parameter		No.	Percent
Optimal time to start treatment to prevent complications	The first two weeks of the baby's life	43	11.9%
	After the first year of the child's life	19	5.2%
	The second month of the baby's life	19	5.2%
	I do not know	281	77.6%
Source of information about congenital hypothyroidism	• Internet social networking sites	114	31.5%
	• family and friends	48	13.3%
	• TV or the Radio	24	6.6%
	• Doctor or nurse	44	12.2%
	• other	132	36.5%
There is an increased awareness of the community about congenital hypothyroidism and its complications	• Yes	305	84.3%
	• No	57	15.7%

Table (5). Association between parents age and consanguinity between parents with occurrence of CH among children

		If participant have a child or more diagnosed with congenital hypothyroidism		Total (n= 362)	P value
		Yes	No		
Age of Father	18 - 25 years old	2 12.5%	42 12.1%	132 12.2%	0.339
	26 - 35 years old	5 31.3%	130 37.6%	405 37.3%	
	36 - 45 years old	5 31.3%	71 20.5%	228 21.0%	
	45 and over	4	103	321	

		If participant have a child or more diagnosed with congenital hypothyroidism		Total (n= 362)	P value
		Yes	No		
Age of Mother	18 - 25 years old	25.0%	29.8%	29.6%	0.006
		5	99	312	
	26 - 35 years old	31.3%	28.6%	28.7%	
		1	98	297	
		6.3%	28.3%	27.3%	
36 - 45 years old	5	69	222		
	31.3%	19.9%	20.4%		
45 and over	5	80	255		
	31.3%	23.1%	23.5%		
Consanguinity between parents	Yes	7	136	429	0.538
		43.8%	39.3%	39.5%	
	No	9	210	657	
		56.3%	60.7%	60.5%	

4. DISCUSSION

Most congenital hypothyroidism (CH) is not avertable; however, the adverse effects of CH are preventable with early detection and treatment. It has become evident that, in some patients with persistent mental retardation and neurological symptoms, defects in transcription factors which are expressed in the thyroid gland as well as in the central nervous system (CNS) during embryonic development cause both defective thyroid and CNS development. However, mild or subtle deficits in verbal skills, attention, memory, or motor development may be observed, particularly in those with severe CH. The severity of CH and pretreatment T4 level are important predictors of adverse cognitive and motor outcomes [12]. There is an increased incidence of other congenital malformations in children with congenital hypothyroidism (CH), particularly cardiac malformations, including septal defects, renal abnormalities, and the risk of neurodevelopmental disorders. A thorough clinical examination, including a hearing screen, should be performed [13].

In our study; 34.5% of all participants had heard of CH. This was higher than a figure reported in Pakistan in which only (20%) of study participants had heard of CH [14]. The etiological classification of CH is based on clinical and biochemical evaluation. Useful tests are measurement of serum TSH, thyroxine (T4), triiodothyronine (T3), and thyroglobulin (TG); thyroid ultrasound and scintigraphy, using 99mTc pertechnetate or, preferably, 123I and when indicated, the perchlorate (ClO₄⁻) discharge test [15]. A previous study reported that parents cited viral/bacterial infections as the most common etiology 83% [14].

Symptoms of CH may include quiet and long sleep duration through the night, hoarse cry and constipation. Neonatal hyperbilirubinemia for more than three weeks is common [16]. A previous study reported the most common symptoms were prolonged jaundice, lethargy, feeding difficulty and constipation [17]. In our study, 21.2% of all participants identified constipation as CH symptom, 22.6% identified excessive sleeping, 15.2% jaundice and 16% facial swelling.

Regarding risk factors and causes of CH, previous studies reported significant association of CH with birth defects, female gender, gestational age >40 weeks, and gestational

diabetes. An increased risk for CH was detected in twins by a multivariate analysis [18]. 18.2% identified iodine deficiency during pregnancy as risk factor for CH. 64.1% of participants didn't know the causes of CH, 24.3% reported hormonal disturbance as a cause of CH, 12.2% reported absence of thyroid gland, and 9.9% reported drugs during pregnancy as cause of CH.

According to our results; 60.2% did not know CH complications while 22.1% reported poor growth, resulting in short stature as a complication, followed by 14.4% Goiter, 14.1% delayed puberty, 13.5% mental retardation, 12.9% osteoporosis, 9.9% heart defects, and 4.6% bone fractures. In comparison to a previous study, (28%) of participants were aware that CH was a cause of intellectual disability. Mothers who had attended the sessions were 11 times more likely to know that CH can cause intellectual disability and 13 times more likely to know that it causes physical growth impairment [14].

In our study, the most commonly cited source of information was internet social networking site in 31.5% followed by family and friends 13.3%, doctors and nurses 12.2%, and TV or the Radio in 6.6%. A previous study in Pakistan reported source of information was friends and family (44%), whereas after the intervention most women cited health sessions (73%) as their source of information [14].

In general, neurodevelopmental outcomes in congenital hypothyroidism (CH) are excellent. Early and adequate therapy initiation, prior to 2nd week of life, will result in an appropriate global intelligence. In our study, only 11.9% of all participants identified optimal time to start treatment to prevent complications as the first two weeks of the baby's life.

5. CONCLUSION AND RECOMMENDATIONS

Parental knowledge of congenital hypothyroidism in Aseer is relatively unsatisfactory. Health campaigns and conferences to raise awareness is necessary looking to catastrophic complications caused by delayed diagnosis and management of the disease. Parents need to be educated on the diagnosis of congenital hypothyroidism (CH), the importance of early and adequate treatment that will prevent poor neurodevelopmental outcomes.

DISCLAIMER

The products used for this research are commonly and predominantly use products in our area of research and country. There is absolutely no conflict of interest between the authors and producers of the products because we do not intend to use these products as an avenue for any litigation but for the advancement of knowledge. Also, the research was not funded by the producing company rather it was funded by personal efforts of the authors.

ETHICAL APPROVAL

Approval was obtained by the Research Ethics Committee of Aseer University, Aseer, KSA. Data was anonymous for patient confidentiality. Use of these anonymous data in this research project was reviewed and approved by the research ethics committee. The collected data was kept safely in a password protected computer.

CONSENT

As per international standard or university standard, Participants' written consent has been collected and preserved by the author(s).

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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Peer-review history:
The peer review history for this paper can be accessed here:
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