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Prevalence of Hypothyroidism and Cardiac Diseases Among Libyan Down Syndrome Patients

Rawasy Etaleb[1], Ahmed Atia[2]*

[1]*Faculty of Pharmacy, Attahadi University of Medical Sciences, Tripoli, Libya.*
[2]*Department of Anesthesia and Intensive Care, Faculty of Medical Technology, University of Tripoli, Libya.*

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ABSTRACT

Hypothyroidism is among the commonest thyroid abnormality in patients with Down syndrome (DS). The purpose of this study was to determine the prevalence of hypothyroidism and cardiac defects among pediatric down syndrome patients attended Tripoli University Hospital. A retrospective study was conducted among children with DS seen at endocrine follow-up clinic in Tripoli University Hospital. Data were collected from patients' registration book and medical records. A total of 50 patients with DS were included in the study out of which 64% were females. Their median age at diagnosis was range between 10–14 years. Abnormal thyroid function was observed in 34 patients (68 %). Presence of chronic heart disease were seen in 30(60%) patients. Early diagnosis and management of thyroid abnormalities are important to decrease further impairment of cognition function in children with DS.

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INTRODUCTION

Hypothyroidism is a condition characterized by an underactive thyroid gland, leading to insufficient production of thyroid hormones. This hormonal imbalance can have widespread effects on the body, impacting metabolism, energy levels, and overall health. On the other hand, Down syndrome is a genetic disorder caused by the presence of an extra copy of chromosome 21. Individuals with Down syndrome are at an increased risk of various health conditions, including congenital heart defects and cardiac diseases [1].

Individuals with Down syndrome have a higher prevalence of hypothyroidism compared to the general population [2]. The exact mechanism behind this association is not fully understood, but it is believed to be related to the altered immune function and genetic factors present in individuals with Down syndrome [3]. Hypothyroidism can further exacerbate the existing health challenges faced by individuals with Down syndrome, impacting their cognitive development, growth, and overall well-being [4].

Congenital heart defects are common in individuals with Down syndrome, affecting nearly half of all babies born with this genetic condition [5]. These heart abnormalities can range from minor issues that may not require treatment to more serious conditions that necessitate surgical intervention. Additionally, adults with Down syndrome are also at an increased risk of developing cardiac diseases later in life, such as atrial septal defects, ventricular septal defects, and atrioventricular canal defects [6]. The presence of hypothyroidism in individuals with Down syndrome in Libya has rarely been reported previously. One study was conducted in 2018 found that over 183 DS children, 30% had overt thyroid diseases [7]. Hence, the current study was conducted to assess the prevalence of hypothyroidism and cardiac defects among pediatric down syndrome patients attended Tripoli University Hospital in Libya.

METHODS

Study design and area

A cross sectional study was carried out at Tripoli University Hospital between May and August 2023.

Data collection

A total of 50 DS patients were collected at pediatric endocrine outpatient department. The collected data included age, gender, thyroid function test

*Corresponding E-mail addresses: ah.atia@uot.edu.ly
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results and types of cardiac problems.

Data analysis

Data were gathered though Microsoft excel and presented as descriptive statistics using tables and figures.

RESULTS

Collected data were organized in table 1, sequence of patients was random based on first collected.

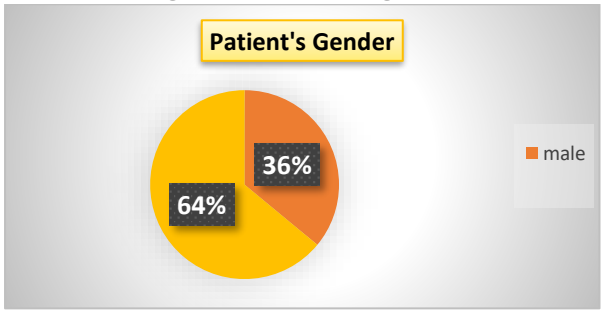
Patient's age was grouped into four groups, among all patients age, higher number (50%) were in age group from 10- 14 years old, followed by the age group from 5-9 years old with percentage 28%.

Table 1. Age related characteristics

Age group	No. of patients	Percentage
0-4 years	7	14%
5- 9 years	14	28%
10- 14 years	25	50%
15- 18 years	4	8%

Dawn syndrome is more common in female (64%) than male (36%).

Figure 2: Patient's gender



Based on collected data, 14% of DS patients were presented with normal thyroid function test (7 patients), 14% showed clinical hypothyroidism (5 patients), 4% with non-significant results were TSH results were normal while T3 or T4 were high (2 patients). While the majority of cases (54%) presented with high TSH level and T3 or T4 results were normal (Table 3).

Table 3. Thyroid function test of DS patients

TFT results	No. of patients	Percentage
Normal TSH and Normal T3 or T4	7	14%
High TSH and High T3 or T4	7	14%
Normal TSH and High T3 or T4	2	4%
High TSH and Normal T3 or T4	27	54%

For cardiac disease, 60% of our patient were diagnosis with cardiac disease and 40% of them were free of any cardiovascular diseases (Table 4).

Table 4. Association between CHD and down syndrome

Presence of CHD	No. of patients	Percentage
Yes	30	60%
No	20	40%

DISCUSSIONS

There is a well-established association between hypothyroidism and cardiac diseases. Hypothyroidism can lead to changes in heart function, including bradycardia (slow heart rate), diastolic dysfunction, and increased risk of atherosclerosis. These cardiac manifestations can further exacerbate the existing cardiac issues in individuals with Down syndrome [9].

In Libya, like in many other countries, individuals with Down syndrome face challenges related to access to healthcare services, early diagnosis, and management of associated health conditions [8]. Limited resources and awareness about the specific healthcare needs of individuals with Down syndrome can impact the detection and treatment of hypothyroidism and cardiac diseases in this population [10].

In the current study, thyroid abnormalities were detected in 34 patients (68 %), which was higher than studies done in other countries like South Africa (34.5%), California (32.5%) and Oregon (24%) [11-13]. Though, they recommend screening for thyroid abnormalities in DS patients at birth, 6 months, 12 months, and yearly then after, [14] in the present study, a significant number of thyroid abnormalities were detected between 10- 14 years of age. This might result in increased prevalence of thyroid abnormalities in the current study compared with the above studies. Other studies also recommend to have additional testing between 2 and 6 months of age [11]. Therefore, we propose additional screening in children with DS for thyroid abnormalities in order to detect the abnormalities earlier and to start treatment timely.

Studies focusing on the prevalence of hypothyroidism and cardiac diseases among individuals with Down syndrome in Libya are limited. In the current study, about 60% of DS patients were diagnosed with cardiac disease.

Similar study was conducted in UK revealed that 342 (42%) infants with Down's syndrome had a cardiovascular anomaly [15]. More research is needed to understand the specific patterns, risk factors, and management strategies for these conditions within the Libyan population with Down syndrome.

CONCLUSION

In conclusion, individuals with Down syndrome are at an increased risk of developing hypothyroidism and cardiac diseases. The association between these two conditions can pose additional challenges for those living with Down syndrome. In Libya, where healthcare resources may be limited, addressing the healthcare needs of individuals with Down syndrome, including screening for hypothyroidism and cardiac diseases, is crucial for improving their overall health outcomes.

Conflict of interest. Not declared

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