

Prevalence of Congenital Hypothyroidism in Neonates

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ABSTRACT

Objective: To determine the frequency of congenital hypothyroidism in neonates.

Patients and Methods: A total number of 200 neonates born through SVD and cesarean section after 37 weeks of gestation aged 48 hours of birth to 2 weeks of neonatal age (as serum of neonates less than 48 hours of life contain mother's serum level of TSH were included). The study was conducted from 05-Nov-2020 to 04-May-2021. Data regarding neonates' age, gender, weight, and height was taken. 2cc venous blood was taken for serum TSH and T4 level. Neonates having TSH hormone level more than 20mIU/mL were labelled as having congenital hypothyroidism (CH).

Results: Mean age of neonates was 9.63±3.94 days. There were 113 (56.50%) females and 87 (43.50%) male neonates. Mean weight of neonates was 2970.28±516.32 grams. Mean height of neonates was 52.23±2.11 cm. Mean gestational age was weeks. Mean occipitofrontal circumference of neonates was 34.69±1.29 cm. Mean TSH level were 6.64±7.92 mIU/L. Mean T4 level was 23.70±4.25 µg/dL. CH was diagnosed in 4 (2.0%) neonates.

Conclusion: Congenital hypothyroidism (CH) is common among neonates. In present study CH was diagnosed in 2.0% neonates who were screened for CH. So CH screening should be done in all neonates to diagnose CH at early stages and to prevent complication of CH in these neonates.

Keywords: Thyroid Stimulating Hormone (TSH), Congenital Hypothyroidism (CH).

INTRODUCTION

The thyroid is a small, butterfly shaped gland located in neck¹, arises from an outpouching of the foregut at the base of tongue (foramen cecum)². It secretes triiodothyronine (T3) and thyroxine (T4), necessary for neurodevelopmental growth and metabolism. Its secretion is regulated by Thyroid Stimulating Hormone (TSH) secreted by anterior pituitary². Neonates depend on endogenous thyroid production that reaches term levels at approximately 18 to 20 weeks gestation¹.

Studies have proven that thyroid hormone plays a crucial role in neurodevelopmental events necessary for development of subcortical and cortical posterior regions of the brain, including the cerebellum, striatum, hippocampus, and corpus callosum. Therefore, insufficient thyroid hormone levels in utero and early neonatal life may result abnormal brain development resulting in cognitive impairment, stunted growth & developmental delay.^{3,4}

The most prevalent form of hypothyroidism is primary congenital hypothyroidism (CH), which is caused by thyroid gland developmental anomalies (thyroid agenesis or dysgenesis) or delays in thyroid hormone production (thyroid dysmorphogenesis). TSH deficiency owing to pituitary insufficiency or anatomical abnormalities of the pituitary gland or hypothalamus causes secondary congenital hypothyroidism (central CH).⁵

CH occurring in approximately 1:2000 to 1:4000 newborns, is one of the most common preventable causes of intellectual disability.⁶ Diagnosis and treatment of CH before 3 months are mandatory to avoid cretinism¹. Variations in the incidence of CH have been found in various communities around the world.⁴ The timing of diagnosis is crucial, later the treatment is started the worse the neuro developmental outcome and IQ will be¹, the best outcome is when treatment is started by two weeks of age¹. If treatment is not started, or significantly delayed, clinical features include widely open anterior and posterior fontanelle, prolongation of physiologic jaundice, feeding difficulties, constipation, respiratory difficulties due to large tongue, somnolence, umbilical hernia, facial puffiness, hoarse cry, hypothermia, skin mottling, bradycardia, lethargy and later on retardation of physical and mental development if remains untreated.^{6,7}

Clinicians depends on neonatal screening programs for diagnosis of CH². Many countries have implemented CH screening programmes. In North America, the first screening programmes for CH were launched in 1972.⁴ For this potentially deadly condition to be diagnosed early and for treatment to begin before considerable harm is done, screening programmes are essential.⁶ One study

conducted at tertiary care hospital of Karachi 16.3% of newborns screened were positive for CH.⁸

The rationale of this study is to reinforce the guidelines regarding early diagnosis of CH to prevent the neurocranial damage. We considered samples before 2 weeks of life as brain damage increases with each passing day if treatment is not initiated. Though data from Pakistan is available, Still we haven't been able to formulate a uniform policy to diagnose CH for every healthy neonate delivered in public sector hospitals, where bulk of patients also come from rural areas. That's why we conducted this study to emphasize the importance of this very effective tool against a treatable cause of mental retardation and to make thyroid screening as an essential part of early newborn screening.

PATIENTS AND METHODS

A total of 200 neonates born through spontaneous vaginal delivery and cesarean of both genders of age 48 hours of birth to 2 weeks of life were included. Study had been approved by ethical committee of the hospital. All neonates were evaluated for enrolment in the study after informed consent from parents about screening. Premature neonates, those born to known hypothyroid mothers/hyperthyroid mothers, admitted for sepsis and other congenital anomalies were excluded.

A 2cc venous blood was taken for serum TSH and T4 level. After collection of samples, they were sent to laboratory. The TSH level above 20 m IU/L was considered for congenital hypothyroidism. It does not caused extra financial burden on parent's pocket. TSH levels was done in the hospital free of cost.

For the statistical analysis SPSS v.16.0 used. Quantitative variables e.g. age, weight, head circumference and height were expressed as mean and standard deviation. CH and gender were expressed in frequency and percentages. To control the effect modifier data was stratified through gender, age, weight, and height and occipito frontal circumference of neonate.

RESULTS

Mean age of neonates was 9.63±3.94 days. There were 113 (56.50%) females and 87 (43.50%) male neonates. Mean weight of neonates was 2970.28±516.32 grams. Mean height of neonates was 52.23±2.11 cm. Mean gestational age was weeks. Mean occipitofrontal circumference of neonates was 34.69±1.29 cm. Minimum occipitofrontal circumference was 31.48 cm and maximum occipitofrontal circumference was 37.20 cm. Mean TSH level were 6.64±7.92 mIU/L. Mean T4 level was 23.70±4.25 µg/dL (Table 1).

CH was diagnosed in 4 (2.0%) neonates (Figure 1).

Table 1. Data of Baseline Study Variables.

Age (Days)	9.63±3.94
Height (cm)	52.23±2.11
Weight (Grams)	2970.28±516.32
Gestational Age (weeks)	39.06±1.71
Occipitofrontal Circumference (cm)	34.69±1.29
TSH Levels (mIU/L)	6.64±7.92
T4 levels (µg/dL)	23.70±4.25

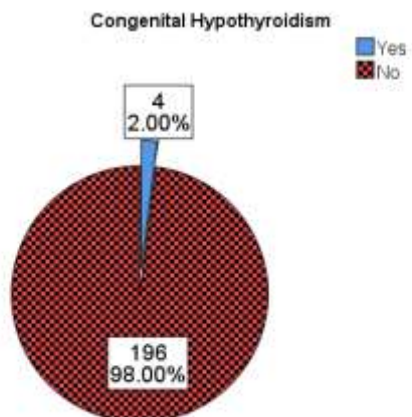


Figure 1. Frequency of CH.

DISCUSSION

Congenital hypothyroidism is a prevalent cause of mental retardation and growth retardation in children. 94 Maternal thyroid hormone helps the fetus's brain development during pregnancy.⁹ It begins to synthesise TSH and T4 hormones on its own in the 20th week of pregnancy. Because of the rapid rise in thyroid stimulating hormone and subsequent fall in T4 levels within the first 3-4 days after birth, variations in thyroid function tests can be seen during the first week of life. TSH levels peak in the first 30 minutes after birth and then gradually decline over the following 3-4 days. After the fourth day of life, most screening programmes recommend taking samples. Since the majority of newborns are released from tertiary care hospitals within 48-72 hours of birth, blood is typically collected at that time. The neonate's age is taken into consideration while interpreting TSH levels.^{10,11}

Mental retardation can be prevented in several cases, but one of the most common is CH. One in every 4000 infants in the globe is born with CH, according to recent research. Hispanics and Native Americans have a higher frequency than blacks (1 in 20,000). (1 per 2000).¹² In the United States and many other nations, neonatal TSH screening programmes are conducted using blood from the cord and heel. Race and ethnicity play a role in the prevalence of disease. One in every 4500 Americans, one in every 700 Red Indians, one in every 3500 Europeans, and one in every 7500 Japanese are infected with CH. 99 In addition, 1 in 2759 Saudi Arabians, 1 in 6246 Thais, and 1 in 2860 Estonians have been reported to have CH. According to Ghaffor et al., from Pakistan, of the 1357 neonates screened for the blood TSH level, 2 had CH.¹³ In 2008, Afroze et al. evaluated the prevalence of CH screening in Pakistani children. Only 10 kids in this hospital-based investigation were diagnosed with congenital hypothyroidism. To get an idea of the true incidence rate, they examined 2008 data over a period of ten months. The final incidence rate was 1 in 1600 live births, according to the researchers.¹⁴

In present study, we determined the frequency of congenital hypothyroidism in neonates of age ≤ 15 days. In present study, CH was diagnosed in 2.0% neonates who were screened for congenital hypothyroidism. This study results are contrary to the results of the study conducted by Salim et al. in Karachi Pakistan reported congenital hypothyroidism in 16.3% patients who were referred for determination of thyroid dysfunction.¹

In newborns, an elevated serum TSH level implies a lack of thyroid hormone delivery to the developing brain. Neonatal TSH has been added as one of the indicators for assessing iodine deficiency illnesses by international organisations such as the World Health Organization and the United Nations Children's Fund.^{15,16} The immunoassay assessment of various combinations of thyroid hormones is used to detect CH in newborn screening programmes. T4 total is not the same as T4 free. Thyroid hormone can be measured in free T4 form, which is a more precise measurement. The level of free, unbound thyroxine T4 in the bloodstream is measured by free T4, which is frequently lower in hypothyroidism. However, due to its simplicity and low false positive rate compared to combination techniques, many newborn screening programmes, especially those in Australasia, use a single TSH test. Due to hypothalamic immaturity, this technique will not detect central hypothyroidism, and low birth weight and premature neonates are a potential cause of false negative screenings, necessitating a second sample.¹⁷ We didn't include newborns who were critically ill or born prematurely.

Before being discharged from the hospital, every newborn must have a blood sample taken for testing. The first 3–5 days following birth are the best. The blood TSH level can yield a misleading positive result when specimens are taken within the first 24–48 hours of life.¹⁸ As a result, this test was performed in the first 3–15 days following birth in our study.

The neonatal TSH screening approach is appropriate for early detection of this crucial condition. As a result, TSH screening programmes should be implemented in all countries. When CH is detected and treated early, there is invariably a positive outcome with no long-term consequences. In our hospital, TSH screening tests for newborns are done on a regular basis. It is critical that all neonates have this screening test performed.

CONCLUSION

Congenital hypothyroidism (CH) is common among neonates. In present study CH was diagnosed in 2.0% neonates who were screened for CH. So, CH screening should be done in all neonates to diagnose CH at early stages and to prevent complication of CH in these neonates.

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