



SCREENING FOR CONGENITAL HYPOTHYROIDISM WITH UMBILICAL CORD BLOOD TSH LEVEL 'AN EXPERIENCE FROM A TERTIARY CARE CENTER'

Pediatrics

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ABSTRACT

BACKGROUND: Congenital hypothyroidism (CH) is the most common preventable cause of mental retardation. Screening for congenital hypothyroidism can be helpful in preventing mental retardation among general population. Umbilical cord blood TSH (CBTSH) estimation has been universally accepted and is one of the most cost effective screening programs in the field of preventive medicine and public health. **AIMS AND OBJECTIVES:** This study was aimed to find the effectiveness of cord blood TSH as a screening tool for congenital hypothyroidism. **MATERIALS AND METHODS:** This hospital based cross sectional study was conducted at GCS Medical College Hospital & Research Centre, Ahmedabad, over a period of one year in 1687 neonates. All newborns delivered at the institute were subjected to cord blood TSH level estimation and a repeat Serum TSH estimation was done at or after 72 hours of life. Diagnosis of congenital hypothyroidism is confirmed postnatally by standard Serum TSH value cut offs as per age. Data collected and statistically analysed. **RESULTS:** Out of 1687 neonates born during the study period, 1548 formed the study group. 805 (52%) were males and 743 (48%) were females. The birth weight of babies ranged from 850 gms to 4300 gms. The value of CBTSH varied from 1.3mIU/L to 101.4mIU/L with mean CBTSH of 7.21mIU/L. 28(1.8%) babies had CBTSH levels >20mIU/L. Out of which four were later diagnosed to have congenital hypothyroidism. All four had CBTSH levels >20mIU/L. All other neonates with CBTSH levels less than 20mIU/L were found to have normal screening later postnatally. **CONCLUSION:** The current study concludes that cord blood TSH is a sensitive marker to screen for congenital hypothyroidism in neonates. A cut off value of cord blood TSH >20mIU/ml can be used for screening purpose.

KEYWORDS

INTRODUCTION:

Congenital hypothyroidism is the commonest preventable cause of mental retardation. The worldwide incidence is 1: 3000-4000 live births and the estimated incidence in India is 1:2500-2800 live births [1, 2]. More than 95% of new born infants with congenital hypothyroidism have a few, if any clinical manifestations. In view of paramount importance of early diagnosis and treatment various screening programs have been initiated [3, 4]. Congenital hypothyroidism screening is considered one of the best tools for the prevention of long term devastating complications like mental retardation, which can occur in untreated cases. The global intelligence quotient has improved after implementation of screening programs, the mean IQ in cases with congenital hypothyroidism who underwent early treatment was 107 compared to 82-88 in pre-screening era [5]. The first screening program for congenital hypothyroidism was established in Canada in 1974. Under this program, the number of cases discovered increased with worldwide variation between 1:3500 and 1:5000 live births [6]. Cord blood TSH (CBTSH) estimation has the advantages of being easy to collect, non-invasive and low rates of follow up loss as the results would be available before the mother leaves the hospital, enabling the repeat sampling if needed at the earliest, which is critical for early institution of treatment if necessary. It is the responsibility of every paediatrician to take initiative and start screening all the babies born under his/her care. Hence we carried this study to determine the usefulness of CBTSH levels in neonatal screening of congenital hypothyroidism.

AIMS AND OBJECTIVES:

- To determine the effectiveness of cord blood TSH as a screening tool for congenital hypothyroidism.

MATERIALS AND METHODS:

This hospital based cross sectional study was carried out from June 2019 to June 2020 at GCS Medical College Hospital & Research Centre, Ahmedabad, Gujarat in 1687 neonates. Prior approval from the institutional ethical committee was sought before starting the study.

Inclusion criteria:

- All inborn neonates

Exclusion criteria:

- Neonates with gross congenital abnormalities.
- Neonates requiring resuscitation at birth.
- Neonates requiring NICU admission at birth

Cord blood samples of all babies were collected and sent for TSH level estimation. Blood samples were collected from the umbilical vein with 2cc syringe, from 15 to 20 cm length of umbilical cord severed at the time of birth of the baby. Newborns were examined in detail after birth. Socio-demographic and other data of both mothers and newborns viz. gestational age, weight, sex, maternal medications and thyroid status were documented in predesigned performa. All neonates were subjected to a repeat Serum TSH estimation from peripheral blood at or after 72 hours of life. Confirmed diagnosis of congenital hypothyroidism was made from serum levels of TSH estimated postnatally as per standard cut offs for age.

RESULTS:

Over the study period, 1687 neonates were born. We excluded 139(8.2%) neonates because of need of resuscitation, NICU admission and gross congenital anomalies. Of 1548 neonates included in the study, 805 were males and 743 were females [Table 1]. The birth weights ranged from 850 gms to 4300 gms. Table 2 depicts the weight wise distribution of entire cohort. 1062 were term neonates where as 486 were preterm neonates [Table 3].

Table 1: Gender wise distribution (n= 1548)

Gender	N (%)
Male	805 (52)
Female	743 (48)

Table 2: Distribution of weight at the time of birth (n=1548)

Birth weight (in kg)	N (%)
<1.0	2 (0.12)
1.0-1.49	8 (0.54)
1.5-1.99	43 (2.8)
2.0-2.49	322 (20.8)
2.5-2.99	765 (49.4)
>3.0	408 (26.4)

Table 3: Gestational age wise distribution (n= 1548)

Gestational age	N (%)
Term	1062 (68.6)
Preterm	486 (31.4)

The CBTSH levels ranged from 1.3 mIU/L to 101.4 mIU/L with mean CBTSH of 7.21 mIU/L. Table 4 shows distribution of CBTSH values. There was no dropout amongst the babies approached for repeat testing.

Table 4: Distribution of CBTSH values & comparison with results (n=1548)

CBTSH value (mIU/L)	N (%)	Confirmed CH
<10	1413 (91.3)	00
11-20	107 (6.9)	00
>20	28 (1.8)	04 (14.28%)

Four babies out of the entire cohort were diagnosed to have congenital hypothyroidism postnatally. All four had CBTSH value >20mIU/L. The CBTSH values in these neonates were 31mIU/L, 38mIU/L, 51mIU/L, and 101mIU/L. All the rest neonates with lesser CBTSH value were found to have negative postnatal screening for congenital hypothyroidism.

DISCUSSION:

Use of CBTSH as a screening tool is an attractive proposition because of its simplicity and accessibility. Fuse, *et al.* [7] had shown that mixed cord blood is a good sampling technique for screening of Congenital Hypothyroidism (CH). Walfish [8] concluded that CBTSH had better specificity and sensitivity as compared to filter paper T4 at 3-5 days of age. Various studies have used different cut offs for the CBTSH levels ranging from 20 to 90mIU/L. We found the cut off value for CBTSH >20mIU/L as abnormal which was comparable to a study by Devi AR and Noushad, *et al.* [9] where CBTSH < 10mIU/L is considered normal, 10-20mIU/L as borderline, >20mIU/L as abnormal. Our results show that only 8.7% samples showed CBTSH >10mIU/L which was comparable to figures from Ethiopia [10]. Our mean value was 7.28mIU/L while Feleke, *et al.* [11] observed value of 9.6mIU/L in 4206 newborns. There is no significant difference in CBTSH levels between male and female babies, similar to observation of Amit Gupta, *et al.* [12] 1.8% of total neonates screened had CBTSH >20mIU/L which was almost equal to 1.833% as reported by Manglik, *et al.* [13] in 1200 term newborns. Wu, *et al.* [14] reported 2.27% in 11,000 neonates. We had four babies with CH out of the cohort of 1548 giving, an incidence of 1 in 387 which is much higher than world figure of 1 in 4000 [15], but Indian data too have quoted higher incidences as 1 in 248 [1] and 1 in 1700 [9]. An Irani study by Ordookhani A, *et al.* [16] reported incidence of 1 in 914.

CONCLUSION:

The present study adds emphasis on the need for continuing screening, one of the important preventable causes of intellectual disability. In India incidence is very high, the necessity of screening programs is the need of the hour. In this study the incidence of CH was 1 in 387. Neonates diagnosed with congenital hypothyroidism had no signs and symptoms at birth or in early postnatal period. Our statistics show comparable trend as with normative data for CBTSH as reported by various workers across globe. We conclude that we may safely use cut off value of > 20mIU/L CBTSH as a screening tool for congenital hypothyroidism.

Still even larger population based studies may be done to achieve more credible guidelines and more so to gauge the epidemiology of CH.

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