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ORIGINAL ARTICLE

Screening for congenital hypothyroidism in Charikot using cord blood: a single center retrospective study

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Abstract

Introduction: Congenital hypothyroidism is a major preventable cause of mental retardation in newborns. According to the screening protocol, neonates with abnormal TSH are recalled for confirmatory tests. According to the American Academy of Pediatrics, the appropriate recall rate after primary thyroid stimulating hormone (TSH) screening is approximately 0.05%. The recommended recall rate was 0.05%, but there is around 0.01 to 13% recall rate worldwide. Our study aims to evaluate the value of cord blood TSH for screening rather than the ideal use of TSH at 72 hours after birth. It would be essential to improve our screening programs with cost-feasible and noninvasive.

Method: A descriptive retrospective study was done in Charikot Dolkha to analyze the recall rate for a repeat TSH when the cord sample is positive for congenital hypothyroidism. Descriptive analysis was performed with a chisquare test for inferential statistics (p 0.05 considered significant).

Result: The recall rate or positive screening rate was 1.8%, the positive prevalence rate was 18.2%, and the overall prevalence rate was 0.33%. This result is higher than the American Academy of Pediatrics estimated but within the range of recall rates worldwide.

Conclusion: The recall rate is significant, and our study's prevalence rate is low. A higher cutoff of >30 mIU/L can be considered, but more extensive populationbased studies are required to establish a higher normative value.

Keywords: Cord Blood, Congenital Hypothyroidism, Thyroid Stimulating Hormone

INTRODUCTION

Congenital hypothyroidism is a major preventable cause of mental retardation in newborns. According to various studies worldwide and neonatal screening programs, it has an incidence of 1 in 4000 births.[1] Neonatal screening programs for detecting congenital hypothyroidism in the neonatal period have been done in various parts of developed countries for decades. They are also gaining momentum in the developing world.[2] In most screening programs, blood samples are collected at 72 hours to 7 days of life.[3] Still, with many babies being discharged within 24 hours in our context, cord blood samples are being used and widely studied in Nepal and other developing countries.[3] Indication for cord blood TSH (cTSH) screening in developed countries is usually done on those who have maternal thyroid disorders or a family history of Congenital hypothyroidism.[4] In our country and peripheral setup like ours, it is very difficult to call back babies once discharged due to the difficult transport system and social system. Thus, cord blood remains a practical alternative for screening purposes in our case and many other developing countries like ours. Newborn screening (NBS) by heel-prick-dried blood spot (DBS) has become an integral part of the public health system in developed countries for pre-symptomatic detection of specific metabolic, endocrine, and hematologic disorders.[5] Single TSH measurement, in either cord blood or heel prick DBS on the fifth day of life, is an easy procedure and has a low false positive rate but will not detect central hypothyroidism.[6] It has a high false negative rate in preterm and low birth weight due to hypothalamic immaturity. [7] The true positive rate for congenital hypothyroidism (CH) in NBS is 1 in 26 (3.9%) of screen positive. Failure to screen for hypothyroidism results in a huge burden to the country with children with mental disabilities.[8] According to the screening protocol, neonates with abnormal TSH are recalled for confirmatory tests.[9] According to the American Academy of Pediatrics, the appropriate recall rate after primary thyroid stimulating hormone (TSH) screening is approximately 0.05%. The recommended recall rate was 0.05%, but there is around 0.01 to 13% recall rate worldwide.[10] The recall was done on the 3rd to 7th days of life. [9] In our study, the main aim is to evaluate the value of cord blood TSH for screening rather than the ideal use of TSH at 72 hours after birth, for which we will use recall rates of cord blood TSH values. Cord blood used for screening programs of congenital hypothyroidism will be the most cost-effective, noninvasive, and have a low follow-up loss rate.[3]

METHOD

A descriptive cross-sectional retrospective study was done in Charikot from Jun 2022 to Jun 2023. The ethical approval was taken from the National Health Research Council with reference no 1299. All newborns were delivered to the Pashupati Chaulagain Memorial Hospital, whose cord blood was collected for screening for congenital hypothyroidism and taken as a sample that doesn't have any exclusion criteria. The exclusion criteria are prematurity and maternal hypothyroidism, which data is not available in the database hospital and where direct venous TSH is recorded. (figure 1) The cutoff of TSH of more than 20mIU/L is taken for screening for congenital hypothyroidism. Recall is done if the cord TSH value is more than 20mIU/L from day 3 to day 7 of life for repeating TSH. (Figure 1)

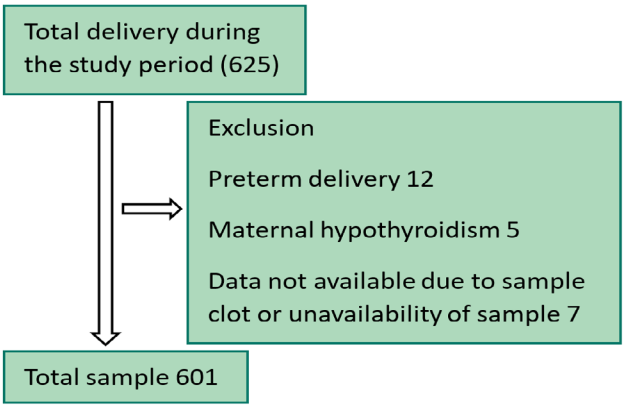

Figure 1. Exclusion of sample

The variables included in the study were maternal age, week of gestation, gender of child, mode of delivery, TSH, and repeat TSH. The collected data was entered in Microsoft Excel and exported into SPSS (Statistical Package for Social Science) software for statistical analysis using version SPSS 25. For descriptive analysis, proportion, percentage, mean, median, and Standard deviation were calculated.

RESULT

A total of 601 cord blood samples were tested for TSH. Out of 601, 283(47.1%) were female baby and 318(53.9%) were male baby (Figure 2).

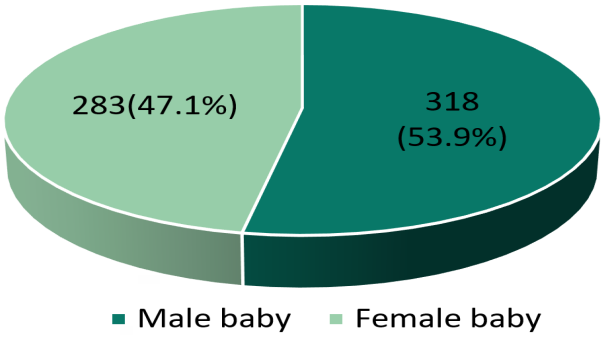

Figure 2. Distribution of male and female babies

The birth weight ranges from 1.7 kg to 4.05kg, with a mean weight 2.99 (standard deviation 0.43). The mean TSH was 9 (standard deviation 5.43), with a minimum TSH of 1.05 and a maximum TSH of 61.51. TSH value corresponding to 3rd, 10th ,25th ,50th, 75th, 95th,97th, 99th are 2.41, 5.10, 6.20, 8.20,10.8, 15.2, 16.2 and 38.15 mIU/L respectively. The range of TSH value is shown in table 1. Among 601, 11 patients were

recalled to confirmed for congenital hypothyroidism, which makes the recall rate to be 1.8%. Among 11, there was one dropout from follow-up, and 2 of them had TSH more than 20 during follow-up on the 7th day, so they were referred to Kanti Children's Hospital for further evaluation. It concluded that the positive predictive value of the study was 18.2% with a TSH cutoff value of 20mIU/L, and the prevalence rate of congenital hypothyroidism was 0.33%.

Table 1. Range of TSH

Range of TSH (mIU/L)	TSH N (%)
0 to 9.99	430 (71.5%)
10 to 19.99	160 (26.6%)
20 to 39.99	7 (1.2%)
40 to 59.99	3 (0.5%)
60 to 100	1(0.2%)

DISCUSSION

Congenital hypothyroidism screening is always an important topic of discussion and is always viewed with great importance as it is linked with preventable mental retardation.[1] Screening for congenital hypothyroidism remains one of the most cost-effective tools for preventing mental retardation, as the nationwide program also exists in our country. [7] Ideally, universal screening is done at 72 hours of birth to detect congenital hypothyroidism.[2] Walfish et al [10] concluded that cord TSH had a higher specificity and sensitivity than tests done in 3 to 5 days. Our study found a higher recall rate of 1.8% than the recommended recall rate by the American Academy of Pediatricians. However, looking at the worldwide recall rate of up to 13%, our recall rate seems good.[3,4] Different cutoff values are used in different studies done worldwide, which can also decrease the recall rate in our study. However, a found and proven recommendation was made to keep the cutoff of 20. [8,10] The prevalence rate in another part of Nepal was 2.4%, and in our study, it was 0.33%, which was found to be much less.[9] Reasons behind different prevalence rates may be due to different geographical regions. In our country and peripheral setup like ours, it is very difficult to call back babies once discharged due to the problematic transport system and financial burden. Cord blood screening could prevent mental retardation caused by congenital hypothyroidism. Screening for congenital hypothyroidism with a cord blood sample remains one of the most cost-effective tools for preventing mental retardation, as the nationwide program also exists in our country. [7]

CONCLUSION

The screening positive rate is significant. However, the prevalence is relatively low. A cutoff of cord blood TSH value of >20 mIU/L was taken for screening for congenital hypothyroidism. A higher cutoff of >30 mIU/L can be considered. Large population-based studies are required to establish normative values for cord blood TSH in our country.

DECLARATIONS

Acknowledgment

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Conflict of Interest

None

Funding

None

Ethical Clearance

The ethical clearance was taken from the National Health Research Council with reference no 1299.

Consent of study

The consent was taken from the pregnant women whose cord blood was used in the study.

Consent of publication from the author

All the authors and participants consented to the publication of the findings.

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