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Original Research Article

Clinico-Biochemical Profiling through Newborn Screening (NBS) to Diagnose Congenital Hypothyroidism (CH): A Cross-Sectional Study

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Abstract

Background: Congenital hypothyroidism (CH) is regarded as the commonest cause of preventable mental retardation. With the introduction of newborn screening, early diagnosis and timely initiation of thyroxine replacement therapy have improved the outcomes of affected children. Study was conducted to find out the incidence of congenital hypothyroidism in term and preterm babies and to find out the epidemiological correlation of congenital hypothyroidism (incidence iodised salt and non-iodised salt consuming population.

Materials and Methods: Cord blood samples were collected with the help of a 5cc syringe from the cord stump of the babies satisfying the afore mentioned inclusion and exclusion criteria at the time of birth in the labour room or Operation Theatre in a clot vial and sent for analysing TSH levels within four hours of collection. The babies were re-evaluated on 4th day for term babies and 5th day for preterm babies by heel prick method. A short history and clinical examination about the babies were conducted. Babies with elevated TSH levels (>20mcU/L) this time also were re-evaluated at the age of one month with a venous sample for TSH levels. Those with persistently raised TSH levels (>20mcU/L) at the age of one month were considered to be suffering from congenital hypothyroidism. The data thus collected were analysed by statistical methods to find out the incidence of congenital hypothyroidism in the institution.

Results: From the present study we can understand that the 85% of those diagnosed to have congenital hypothyroidism by cord blood TSH estimation have transient hypothyroidism. Also, the rate of positivity of TSH levels decline as the numbers of days of life of the child increases. Thus, transient hypothyroidism must be ruled out before treating a child for congenital hypothyroidism. In our study mean TSH for preterm is 5.3±10.6 and 4.09±6.4mIU/L for term babies. Our study shows significant correlation between preterm delivery and congenital hypothyroidism (p value- 0.0006). The present study shows that prolonged jaundice, hypotonia, difficult oral feeding and open posterior fontanelle have statistically significant association with congenital hypothyroidism The normative range cord blood TSH for babies as found in my study population is 2.2 to 11.09 (3^{rd} to 97^{th} percentile as obtained from the study population). The normative range heel prick TSH for babies as found in my study population is 0.9 to 7.54 (3^{rd} to 97^{th} percentile as obtained from the study population).

Conclusion: Congenital hypothyroidism has a high incidence in our country. Its inclusion in neonatal screening protocol has been rightly done. This can help us prevent an important cause of mental retardation. Transient hypothyroidism is a common entity. So treatment of hypothyroidism should not be started on the basis of cord blood Thyroid Stimulating Hormone (CBTSH). The 4-5^{th} heel prick TSH and 28 day venous TSH should be done before diagnosing a case of congenital hypothyroidism.

Keywords: Congenital hypothyroidism, Transient hypothyroidism, Newborn screening (NBS), Thyroid Stimulating Hormone (TSH), Pregnancy, Vaginal Delivery, Caesarean Section.

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Introduction

Congenital hypothyroidism (CH) is the leading cause of preventable mental retardation, which is currently not universally screened in India. Congenital Hypothyroidism (CH), if diagnosed and treated early, is one of the most preventable and treatable causes of mental retardation in newborns and infants. Due to the passage of maternal thyroid hormone through the placenta to some extent, clinical features of CH remain subtle rendering newborns undiagnosed at birth [1]. Studies from various countries indicated treatment of CH should be started no later than the first 2 weeks of life [2, 3].

The prevalence of congenital hypothyroidism in India (1:1000) is higher than the other countries (1:3000–4000). A recent meta-analysis reported an overall CH prevalence of 0.97 per thousand (1:1031) that ranged from <1:4057 to 1:23. India is a diverse country with varying genetic and environmental influences. Hence, understanding the prevalence across different states or geographical regions is essential [4, 5].

Congenital hypothyroidism (CH) is one of the most important causes of preventable mental retardation. The goal of neonatal CH screening programs is early diagnosis and treatment [6] . The delayed diagnosis made only on the basis of clinical findings may result in irreversible complications such as mental retardation and deafness [7, 8] . The difficulty in recognizing CH and the serious consequences of delayed therapy have led to the introduction of screening programs for hypothyroidism in newborns by measuring thyroxine (T4) or thyroid-stimulating hormone (TSH or thyrotropin) in spots of blood collected via heel stick during the first few days of life [9].

Aetiology of congenital hypothyroidism:

1. Permanent congenital hypothyroidism is a type of congenital hypothyroidism, a thyroid hormone deficiency present from birth [10] and may be due to primary or secondary (central) causes. Primary causes include defects of thyroid gland development, deficiencies in thyroid hormone production, and hypothyroidism resulting from defects of TSH binding or signal transduction.

Peripheral hypothyroidism results from defects in thyroid hormone transport, metabolism, or resistance to thyroid hormone action. Secondary or central causes include defects of thyrotropin releasing hormone (TRH) formation or binding and TSH production [11, 12].

2. Transient hypothyroidism is defined as transient abnormality in newborn which revert back to normal after varying period of time and are identified in neonatal screening tests. It may be caused by maternal or neonatal factors. Maternal factors include antithyroid medications, trans placental thyrotropin receptor blocking antibodies and exposure to iodine deficiency or excess. Neonatal factors include, neonatal iodine deficiency or excess, congenital liver haemangioma and mutations in the genes encoding for DUOX and DUOXA2 [6, 7].

3. Overt hypothyroidism-Overt hypothyroidism is defined as a clinical syndrome of hypothyroidism associated with elevated TSH and decreased serum levels of T4 or T3 [8]

4. Subclinical Hypothyroidism- Subclinical hypothyroidism, also called mild thyroid failure, is diagnosed when peripheral thyroid hormone levels are within normal reference laboratory range but serum thyroid-stimulating hormone (TSH) levels are mildly elevated [9].

The critical points in the identification of CH, and its prevalence, are as follows:

- CH is due to a deficiency in circulating thyroxin.
- Untreated CH is a cause of severe mental retardation.
- Early detection and treatment that starts within the first month of life can prevent mental retardation due to CH.
- World data have shown that the birth prevalence of CH is approximately1:3000.

Aims and Objectives

Primary Objective: To find out the incidence of congenital hypothyroidism in term and preterm babies in the institute.

Secondary objectives

- To find out the correlation between congenital hypothyroidism and various disease processes like perinatal asphyxia, sepsis, maternal risk factors like antithyroid drugs, thyroid supplementation etc.
- To find out the correlation between congenital hypothyroidism and gestational maturity of the baby.
- To find out the epidemiological correlation of congenital hypothyroidism (incidence iodised salt and non-iodised salt consuming population)

Materials and Methods

Study Area: Babies born by vaginal delivery and Caesarean section in the Labour Room-Operation Theatre complex of Department of Obstetrics and

Gynaecology in N.R.S. Medical College and Hospital

Study Population: 1000 term and preterm babies born in N.R.S. Medical College and Hospital by vaginal delivery or caesarean section.

Sample Size: 1000

Study Period: 1 year

Study Design: Prospective observational study

Inclusion Criteria: Term or preterm babies born by normal delivery or caesarean section.

Exclusion Criteria

1. Preterm babies of gestational age less than 28 weeks.
2. Term and preterm babies with congenital anomalies.
3. Either parent of the baby refusing to participate in the study.

Study Techniques: Cord blood samples were collected with the help of a 5cc syringe from the cord stump of the babies satisfying the afore mentioned inclusion and exclusion criteria at the time of birth in the labour room or Operation Theatre in a clot vial and sent for analysing TSH levels within four hours of collection. The babies were re-evaluated on 4th day for term babies and 5th day for preterm babies by heel prick method. A short history and clinical examination about the babies were conducted. Babies with elevated TSH levels (>20mcU/L) this time also were re-evaluated at the age of one month with a venous sample for TSH levels. Those with persistently raised TSH levels (>20mcU/L) at the age of one month were considered to be suffering from congenital hypothyroidism. The data thus collected were analysed by statistical methods to find out the incidence of congenital hypothyroidism in the institution.

Parameters to be studied: The babies included in the study will be analysed in a methodical manner as follows-

1. **Cord blood TSH-** The cord blood collected were sent for analysis by chemiluminescence method. and evaluated by Beckman Coulter ACCESS machine

2. **Heel prick sample for TSH on 4-5 day of life:** The babies will be tested by heel prick method on day 4 for term neonates and day 5 for preterm neonates using a filter paper which will be sent to the laboratory for estimation of TSH by elution technique by the same machine

3. **Short history and clinical examination:** This will include a short maternal history including age, complications, address, type of delivery, paternal history, environmental history and examination of the baby including gestational age, birth weight, sex, post maturity, poor feeding, hypotonia, hoarse cry, macroglossia, size of posterior fontanelle, persistent jaundice, US:LS, head circumference etc.

4. **Venous sample for TSH levels:** The babies with raised TSH levels in heel prick method will be re-evaluated at one month of age for TSH levels in venous blood. Those found with raised TSH levels this time also will be considered to be suffering from hypothyroidism (overt hypothyroidism). The rest who was found to have TSH less than <20mIU/L were considered to have transient hypothyroidism.

Analysis of Data: Data of all the cases was entered into SPSS version 17 program and analyzed for statistical package. Socio-demographic data such as mother's age, weight, gestational age, was presented as mean and standard deviation.

The qualitative data such as sex, history of any thyroid disease in mother, use of anti-thyroid drugs by mother during pregnancy and TSH results were presented as frequency tables. Chi-square test was applied to find out association of CH with mother's age, newborn's sex, weight and gestational age, history of any thyroid disease in mother and use of anti-thyroid drugs by mother during pregnancy.

Results

A total number of 1640 babies born in NRSMCH in the year Dec 2016-Sep 2017were screened for congenital hypothyroidism. the sociodemographic profile of the newborn is as follows:

Table 1: Sex distribution of the newborn screened

Sex	Number (%)	Number of positive on Day 28	P value
Boy	946 (57.6%)	1	
Girl	694 (43.4%)	1	0.8

This shows that out of total 1640 newborn screened 946(57.6%) are boys and 694(43.4%) were girls [Table 1]. This above table shows that sex of the baby is not statistically significant with congenital hypothyroidism.

Table 2: Gestational age distribution in the population

Gestational Age	Number	Mean Gestational Age	Mean Birth Weight
Term (> 37 weeks)	1504 (81.7%)	38.5±3.4	2.87±0.87
Preterm (<37 weeks)	136 (8.3%)	33.3±9.6	1.7±0.7

Table 3: Distribution of preterm birth

Preterm	Number (%)	Mean GA	Mean Birth Weight
< 34 Weeks	69 (50.7%)	31.3±2.6	1.42±0.4
> 34 Weeks	67 (49.2%)	36.5±1.8	2.45±0.6

The above tables show that out of 1640 babies 1504 (91.7%) were term babies with mean GA of 38.5±3.4 and mean birth weight of 2.87±0.87 and 136 (8.3%) were preterm (mean GA-33.3±9.6, mean B.W.- 1.7±0.7) of which 49.2% were of GA >34 wks (mean GA 36.5±1.8, mean B.W. 2.47±0.6) and 50.7% were of <34wks (mean GA 31.3±2.6, mean B.W. 1.4±0.4) [Table 2, 3].

Table 4: Gestational age wise distribution of cord blood and heel prick TSH

Gestational Age	Mean CBTSH	Mean Heel Prick TSH	More than 20 in CBTSH	More than 20 in Heel Prick TSH	Positive in 28 day venous sample	P value
Term	4.09±6.4	2.63±4.2	8 (0.7%)	5 (0.4%)	0	0.0006
Pre-term	5.3±10.6	3.5±8	6 (4.4%)	4 (2.9%)	2	
<34 wks	5.25±10.4	3.75±9,0	3	3	2	0.48
>34 wks	4.15±6.6	2.6±4.2	3	1	0	

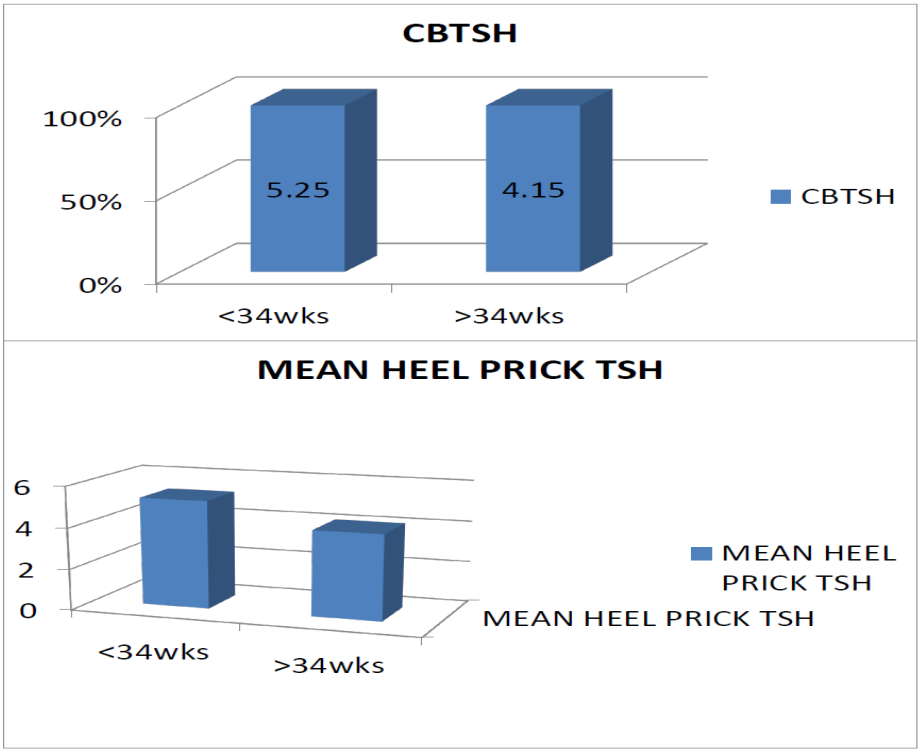

Figure 1: CBTSH and Mean Heel Prick TSH

From the above tables we can conclude that cord blood and heel prick TSH levels are more in preterm than in term babies. Among the preterm it is more in <34wks of gestational age. Also, it has been found that correlation between preterm babies and congenital hypothyroidism is statistically significant.

Table 5: Day wise positivity of TSH level in the babies screened

Day when tested	Number found positive (>20 mcU/L)
Day 1 (CB-TSH)	14
Day 4 (Heel prick TSH)	9
Day 28 (Venous blood TSH)	2

From the above charts we can understand that the 85% of those diagnosed to have congenital hypothyroidism by cord blood TSH estimation have transient hypothyroidism. Also, the rate of positivity of TSH levels decline as the numbers of days of life of the child increases. Thus, transient hypothyroidism must be ruled out before treating a child for congenital hypothyroidism.

Table 6: Association between mode of delivery and TSH levels

Type of Delivery	Number	Number Positive by CB-TSH	Number Positive by Heel Prick TSH	Number of Babies found positive (>20 mcU/L in Day 28)	P value
Vaginal Delivery	827	6	5	1	0.99
Caesarean Section	813	8	4	1	

From the above table we can conclude that mode of delivery has no influence over the TSH levels of the child. The correlation has been found to be statistically insignificant.

Table 7: Association between mode of delivery and TSH levels

Maternal History	Number	Number Positive by CB-TSH	Number Positive by Heel Prick TSH	Number of Babies found positive (>20 mcU/L in Day 28)	P value
Anaemia	250	1	0	0	0.5
Antepartum Haemorrhage	16	0	0	0	0.8
Gestational Diabetes	10	0	0	0	0.9
Hypertension	33	2	2	0	0.8
Hypothyroidism	25	2	2	0	0.8
Fibroid Uterus	2	0	0	0	0.9
Increased Age	2	2	2	1	0.0001
NAD	1304	7	3	1	0.8
Antithyroid Drugs	0	0	0	0	0

From the above table we can conclude that increased age of mother(>30yrs) has statistical correlation with the TSH level of the baby. other complications were not found to be statistically significant with increased TSH levels.

Table 8: Association of maternal age with TSH levels

Maternal Age	Number	Number Positive by CB-TSH	Number Positive by Heel Prick TSH	Number of Babies found positive (>20 mcU/L in Day 28)	P value
≤ 25 Yrs	1422	13	8	1	0.62
>25 yrs	218	1	1	1	

From the above table we can conclude that the correlation between TSH levels of the baby and mother's age is not statistically significant.

Table 9: Association of type of salt consumed with TSH levels of baby

Type of salt consumed	Number	Number Positive by CB-TSH	Number Positive by Heel Prick TSH	Number of Babies found positive (>20 mcU/L in Day 28)	P value
Iodised	1612 (98.2%)	3.97±5.4	2.56±3.6	1 (0.06%)	0.01
Non-iodized	28 (1.7%)	17.027±20.0	11.08±15.32	1 (3.5%)	

From the above table we can conclude that maternal consumption of non-iodised salt and raised TSH levels in the babies have a correlation that is statistically significant.

Table 10: Association of neonatal complications with hypothyroidism

Complications	Number	Number Positive by CB-TSH	Number Positive by Heel Prick TSH	Number Positive Venous TSH	P value
Sepsis/Meningitis	12	0	0	0	0.9
Perinatal Asphyxia	8	0	0	0	0.9
Macroglossia	0	0	0	0	0
Prolonged Jaundice	3	3	3	1	0.0001
Hypotonia	3	2	2	1	0.0001
Post Maturity	11	3	3	0	0.9
Difficult Oral Feeding	3	2	2	1	0.0001
Hoarse cry	0	0	0	0	0
Posterior Fontanelle Widening	1	1	1	1	0.0001
US:LS Ratio	2	2	2	2	0.0001

The above table shows that prolonged jaundice, hypotonia, difficult oral feeding and open posterior fontanelle have statistically significant association with congenital hypothyroidism The normative range cord blood TSH for babies as found in my study population is 2.2 to 11.09 (3rd to 97th percentile as obtained from the study population). The normative range heel prick TSH for babies as found in my study population is 0.9 to 7.54 (3rd to 97th percentile as obtained from the study population).

Discussion

Thyroid hormone with an important role in the development of a central nervous system, thyroid hormone deficiency at birth is called CH.3 As the clinical features of CH as subtle or absent at birth, many newborn babies remain undiagnosed [13]. Pilot screening programs for CH were developed in many part of the world by using newborn heel prick filter paper dried blood samples, and have now been established in most parts of the world [14].Newborn Screening (NBS) for CH has been considered a major achievement in the field of preventive medicine and is widely accepted as a tool in primary health care for infants like breastfeeding, immunisation and oral rehydration [15].

Screening for congenital hypothyroidism (CH) is widespread for the last two decades. We have not been able to implement it in India because of several factors, like cost, lack of reliable laboratories on a large scale, and nonavailability of baseline data in our population. Use of cord blood TSH as a screening tool is an attractive preposition because of its simplicity and accessibility. Fuse, et al.[17] had shown that mixed cord blood is a good sampling technique for screening for CH. Walfish [18]concluded that cord TSH had a better specificity and sensitivity as compared to cord or filter paper T4 at 3-5 days of age.

The present study population consisted of 1640 babies of which 946 (57.6%) were male and 694(43.4%) were females, 1504 (91.7%) babies were term and 136(8.3%) were preterm, 1138(69.3%) were Hindus, 497(30.3%) were Muslims, and 5(0.3%) were Christians. Similar demographic statistics were found in studied conducted by Manglik et al [19] and Anand et al [20]. Our study shows that 5.12% (84) babies had a cord blood TSH more than 10mIU/L. This is comparable to figures from Ethiopia. Our mean value was 4.19±6.8mIU/L while Feleke, et al.[21] observed value of 9.6 ± 7.8 mIU/L in 4206 newborns. Thus, our values are much lower compared to other studies. Our 97th percentile is 11.09mIU/L and Kung, et al.[22] have quoted a 95th percentile of 16 mIU/L. However, our TSH values were somewhat lower than found by Khadilkar, et al.[23] who, in a study of 203 neonates found a mean cord blood TSH value of 12.3 ± 4.9 mIU/L. Thus, we have found a lower TSH baseline values compared to that found in other studies worldwide. but our normative data is comparable to other Indian studies.

We had two babies with CH out of a cohort of 1640 establishing an incidence of 1:820 which is much higher than the world figure of 1 in 4000 [24], but other Indian data too have quoted higher incidences as 1 in 248 [25] and 1 in 1700 [26] and study done by Manglik et al [19] also shows an incidence of 1:600. A recent Iranian study found an incidence of 1 in 914 [27]. Probably geographic and ethnic differences are responsible and of course, this cohort of 1640 samples are too small to assess incidence. In our study there is no statistical correlation between sex of the baby and congenital hypothyroidism (p value- 0.8). But the incidence of congenital hypothyroidism has been found to be higher in females (1:694) than males (1:946). These results are in accordance to Nelson textbook of paediatrics [24] which states that the incidence of congenital hypothyroidism is much higher in females (ratio is 2:1). Rastogi et al[1] also found higher incidence in females. The study done by Anjum et al [28] however found a male: female ratio is1.1:1. This could be due to higher number of males in the population. In other studies, mean TSH is 6.4mIU/L for males and 6.2mIU/L for females. The mean TSH for males in our study is 4.33±5.6mIU/L and for females the mean is 4.32±7.0 The results are similar to study by Gopalkrishnan et al [29]. In our study vaginal delivery babies have a mean CBTSH of 4.15±6.8 and caesarian section babies have a mean CBTSH of 4.23±6.8. The mean CBTSH 6.5mIU/L for vaginal delivery and 5.6mIU/L for caesarian section.

The correlation of type of delivery and congenital hypothyroidism has been found to be statistically insignificant (p value-0.9). however, Miyamoto et al [30] in his study found significant correlation between vaginal delivery and congenital hypothyroidism. This might be due to his smaller population size. Seth et al [31] conducted a study in which 130 neonates were screened. Her population size was smaller, but she also found results similar to our study. This difference might be due to different ethnic background. There is no literature available to show the possible cause of this correlation between vaginal delivery and congenital hypothyroidism. Which is also consistent with our results. The mean TSH for 6.3mIU/L for term and 6.3mIU/Lfor preterm in other studies. In our study mean TSH for preterm is 5.3±10.6 and 4.09±6.4mIU/L for term babies. Our study shows significant correlation between preterm delivery and congenital hypothyroidism (p value- 0.0006).

This result is consistent with Nelson Textbook of Paediatrics [24] which states that preterm infants have immature hypothalamo-pituitary-thyroid axis, immature thyroid metabolism and immature hormone synthesis. Mao et al [32], Rastogi et al [1] and Manglik et al [19] in their study have also concluded similar results. In the present study the recall rate based on CBTSH of more than 20mIU/L is 0.9% which is 14 out of 1640 which did not compare well with of the large study of

Wu et al.[33] whose large cohort of 11,000 neonates had a recall rate of 2.27%. But a 5-year prospective study from Thailand [34] used a cut-off value of 30 to begin with and had a recall rate of 1.1% in a large sample size of 35,390 neonates which is well compared with our study. It also closely matches with recall rates of study done by Manglik et al[19] which is 1.83%. Thus, this states that a cut off level of 20mIU/L can be standardised for our country.

In our study we have found an increased incidence of hypothyroidism found with increased age of the mother (p value- 0.0001). Studies done by Rastogi et al [1], Manglik et al [19] also show similar results. The reason might be that increased maternal age especially > 30yrs complications pregnancy with placental insufficiency leading topreterm delivery, and also increased chances of non-disjunction causing various genetic mutations .However when maternal age of less than 25 yrs and more than 25 yrs is compared it is found to be insignificantly correlated to congenital hypothyroidism These results are in accordance with the study by Adeniran et al. [35] in which most of the mothers were above 25 years of age. Similar results were also reported previously in Pakistan by Karam et al. [36]

Maternal consumption of non-iodised salt has been found to be significantly correlated to congenital hypothyroidism. (p value-0.04) Nelson textbook of Paediatrics [24] states that maternal goitre due o iodine deficiency is the most common cause of congenital hypothyroidism. Decreased iodine intake by mother causes decreased iodine uptake by fetus. causing increased TSH and low FT4 in the baby.

Maternal hypothyroidism has been found to be insignificantly ralated to congenital hypothyroidism in our study (p value- 0.8). Manglik et al [19] however found statistical significance between the two. The results are similar to that of Anjum et al [28] who also found significance of maternal hypothyroidism which however goes against our study. Our study shows increased incidence of congenital hypothyroidism among Muslims, but it is not statistically significant (p value-0.1). No literature however has also been found to second this correlation. The present study shows significant correlation of hypotonia, prolonged jaundice, difficulty in oral feeding, posterior fontanelle widening and deranged US:LS ratio with congenital hypothyroidism. The studies of Rastogi et al [1], Macgillivray et al[37] and Kurtoglu et al[38] showed positive correlation between prolonged jaundice and congenital hypothyroidism due to poor bile duct and intestinal motility. Rastogi et al[1] and Smith et al[39] also show correlation of wide posterior fontanelle with congenital hypothyroidism.

Present study has certain limitations. We could not perform T4 level along with TSH for neonatal screening. This would not detect central hypothyroidism. This would not precisely able to differentiate between true or physiological rise in TSH. However, since Neonatal screening for CH is recommended by the 5th day of the child's life at the latest, we have screened the babies on 1st and 4th day of life, the rise in TSH in the present study might be true We were unable to do maternal TSH levels, maternal anti-TSH receptor antibody and urinary iodine levels which would have been useful for determination of etiology in cases of transient CH.

The prevalence of CH was estimated separately for neonates in endemic and non-endemic regions, neonates born to mothers with thyroid disease and preterm neonates. Meta-analysis of 54 studies that included term neonates from non-endemic regions (55 datasets, and 819 559 neonates), gave a pooled prevalence of 0·97 per 1 000 neonates (95% confidence interval/CI: 0·9 to 1·04), which is about 1 in 1 031 neonates [4].

Conclusion

Congenital hypothyroidism has a high incidence in our country. Its inclusion in neonatal screening protocol has been rightly done. This can help us prevent an important cause of mental retardation. Transient hypothyroidism is a common entity. So treatment of hypothyroidism should not be started on the basis of Cord blood Thyroid Stimulating Hormone (CBTSH). The 4-5th heel prick TSH and 28 day venous TSH should be done before diagnosing a case of congenital hypothyroidism. The children with CH grow generally in a normal tempo but the disappearance of bone age retardation is individual and may be protracted until 10 years of age. The incidence of hypothyroidism in India is different in different studies. Larger and multicentric studies are required to correctly document the actual incidence.

References

1. Rastogi MV, LaFranchi SH. Congenital hypothyroidism. Orphanet J Rare Dis. 2010 Jun 10;5:17. doi: 10.1186/1750-1172-5-17. PMID: 20537182; PMCID: PMC2903524.

2. Léger J, Olivieri A, Donaldson M, Torresani T, Krude H, van Vliet G, Polak M, Butler G; ESPE-PES-SLEP-JSPE-APEG-APPES-ISPAE; Congenital Hypothyroidism Consen sus Conference Group. European Society for Paediatric Endocrinology consensus guidelines on screening, diagnosis, and management of congenital hypothy roidism. J Clin Endocrinol Metab. 2014 Feb;99(2):363-84. doi: 10.1210/jc.2013-1891. Epub 2014 Jan 21. PMID: 24446653; PMCID: PMC4207909.
3. Mishra B, Keshary Bhatta N, Chandra Regmi M, Kumar Lal Das B, Ahmed Khan S, Gelal B, Niraula A, Lamsal M. Congenital Hypothyroidism among Infants Undergoing Thyroid Function Test in a Tertiary Care Centre: A Descriptive Cross-sectional Study. JNMA J Nepal Med Assoc. 2022 Jun 1;60(250):503-506. doi: 10.31729/jnma.7505. PMID: 35690974; PMCID: PMC9275452.
4. Anne RP, Rahiman EA. Congenital hypothyroidism in India: A systematic review and meta-analysis of prevalence, screen positivity rates, and etiology. Lancet Reg Health Southeast Asia. 2022 Jul 14;5:100040. doi: 10.1016/j.lansea.2022.100040. PMID: 37383660; PMCID: PMC10305877.
5. Sarathi V, Nikith S. Prevalence of Congenital Hypothyroidism in India: Mapping and Critical Appraisal. Indian J Endocrinol Metab. 2024 Jan-Feb;28(1):98-99. doi: 10.4103/ijem.ijem_73_23. Epub 2024 Feb 26. PMID: 38533278; PMCID: PMC10962764.
6. Büyükgebiz A. Congenital hypothyroidism clinical aspects and late consequences. Pediatr Endocrinol Rev. 2003 Dec;1 Suppl 2:185-90; discussion 190. PMID: 16444157.
7. Bellman SC, Davies A, Fuggle PW, et al. Mild impairment of neuro-otological function in early treated congenital hypothyroidism. Arch Dis Child 1996; 74 (3): 215– 8.
8. Rouet J, Walker W, Bliss B, et al. Long term sequel of hearing impairment in congenital hypothyroidism. J Pediatr 1996; 128 (6): 776–83.
9. Veisani Y, Sayehmiri K, Rezaeian S, Delpisheh A. Congenital hypothyroidism screening program in iran; a systematic review and metaanalysis. Iran J Pediatr. 2014 Dec;24(6):665-72. Epub 2014 Oct 6. PMID: 26019769; PMCID: PMC4442825.
10. Permanent congenital hypothyroidism [homepage on internet]. [updated 2010 Aug]. Available from: http://www.orpha.net/cgi-bin/OC_Exp. [Accessed on April 10, 2026]
11. Bhavani N. Transient congenital hypothyr oidism. Indian Journal of Endocrinology and Metabolism 2011; 15(Suppl2): S117-S120. https://doi.org/10.4103/2230-8210.83345.
12. Khandelwal D, Tandon N. Overt and subclinical hypothyroidism: who to treat and how. Drugs. 2012 Jan 1;72(1):17-33. doi: 10.2165/11598070-000000000-00000. PMID: 22191793.
13. Bernal J. Action of thyroid hormone in brain. J Endocrinol Invest. 2002 Mar;25(3):268–88.
14. Grant DB, Smith I, Fuggle PW, Tokar S, Chapple J. Congenital hypothyroidism detected by neonatal screening: relationship between biochemical severity and early clinical features. Arch Dis Child. 1992 Jan;67(1):87–90. doi: 10.1136/adc.67.1.87.
15. Dussault JH, Coulombe P, Laberge C, Letarte J, Guyda H, Khoury K. Preliminary report on a mass screening program for neonatal hypothyroidism. J Pediatr. 1975 May;86(5):670–4.
16. Buyukgebiz A. Newborn screening for congenital hypothyroidism. J Clin Res Pediatr Endocrinol. 2013;5(Suppl 1):8–12.
17. Fuse Y, Wakae E, Nemoto Y, Uga N, Tanaka M, Maeda M, Tada H, Miyachi Y, Irie M. Influence of perinatal factors and sampling methods on TSH and thyroid hormone levels in cord blood. Endocrinol Jpn. 1991 Jun;38(3):297-302. doi: 10.1507/endocrj1954.38.297. PMID: 1794336.
18. Walfish PG. Evaluation of three thyroid-function screening tests for detecting neonatal hypothyroidism. Lancet. 1976 Jun 5;1(7971):1208-10. doi: 10.1016/s0140-6736(76)92159-0. PMID: 58257.
19. Manglik AK, Chatterjee N, Ghosh G. Umbilical cord blood TSH levels in term neonates: a screening tool for congenital hypothyroidism. Indian Pediatr. 2005 Oct;42(10):1029-32. PMID: 16269841.
20. Anand MR, Ramesh P, Nath D. Congenital Hypothyroidism Screening with Umbilical Cord Blood: Retrospective Analysis. Indian Pediatr. 2015 May;52(5):435-6. doi: 10.100 7/s13312-015-0652-8. PMID: 26061935.
21. Feleke Y, Enquoselassie F, Deneke F, Abdulkadir J, Hawariat GW, Tilahun M, Mekbib T. Neonatal congenital hypothyroidism screening in Addis Ababa, Ethiopia. East Afr Med J. 2000 Jul;77(7):377-81. PMID: 12862157.
22. Kung AW, Lao TT, Low LC, Pang RW, Robinson JD. Iodine insufficiency and neonatal hyperthyrotropinaemia in Hong Kong. Clin Endocrinol (Oxf). 1997 Mar;46(3):315-9. doi: 10.1046/j.1365-2265.1997.1310960.x. PMID: 9156041.
23. Khadilkar V, Khadilkar A, Cowasji H. Neonatal thyroid screening program using filter paper method. Cape News 2002; 6:1 70. Ordookhani A, Mirmiran P, Najafi R, Hedayati

M, Azizi F. Congenital hypothyroidism in Iran.Indian J Pediatr 2003; 70: 625-628.
24. LeFranchi S. Hypothyroidism. In: Kliegman RM, Stanton BF, St. Geme III JW, et al., eds. Nelson Textbook of Pediatrics . 20th ed. Philadelphia, Pa.: Elsevier Saunders; 2011;chap 559.
25. Rama Devi AR, Naushad SM. Newborn screening in India. Indian J Pediatr. 2004 Feb;71(2):157-60. doi: 10.1007/BF02723099. PMID: 15053381.
26. Desai MP, Colaco MP, Ajgaonkar AR, Mahadik CV, Vas FE, Rege C, Shirodkar VV, Bandivdekar A, Sheth AR. Neonatal screening for congenital hypothyroidism in a developing country: problems and strategies. Indian J Pediatr. 1987 Jul-Aug;54(4):571-81. doi: 10.1007/BF02749056. PMID: 3653961.
27. Ordookhani A, Mirmiran P, Najafi R, Hedayati M, Azizi F. Congenital hypothyroidism in Iran. Indian J Pediatr. 2003 Aug;70(8):625-8. doi: 10.1007/BF02724251. PMID: 14510082.
28. Anjum A, Afzal MF, Iqbal SM, Sultan MA, Hanif A. Congenital hypothyroidism in neona tes. Indian J Endocrinol Metab. 2014 Mar;18 (2):213-6. doi: 10.4103/2230-8210.129114. PMID: 24741519; PMCID: PMC3987273.
29. Gopalakrishnan V, Joshi K, Phadke S, Dabadghao P, Agarwal M, Das V, Jain S, Gambhir S, Gupta B, Pandey A, Kapoor D, Kumar M, Bhatia V. Newborn screening for congenital hypothyroidism, galactosemia and biotinidase deficiency in Uttar Pradesh, India. Indian Pediatr. 2014 Sep;51(9):701-5. doi: 10.1007/s13312-014-0485-x. PMID: 25228601.
30. Miyamoto N, Tsuji M, Imataki T, Nagamachi N, Hirose S, Hamada Y. Influence of mode of delivery on fetal pituitary-thyroid axis. Acta Paediatr Jpn. 1991 Jun;33(3):363-8. doi: 10.1111/j.1442-200x.1991.tb01567.x. PMID: 1785333.
31. Seth A, Rashmi M, Bhakhri BK, Sekri T. Neonatal thyroid screening: Relationship between cord blood thyroid stimulating hormone levels and thyroid stimulating hormone in heel prick sample on 4(th) to 7(th) day-of-life. Indian J Endocrinol Metab. 2014 Jan;18(1):125-6. doi: 10.4103/2230-8210.126599. PMID: 24701451; PMCID: PMC3968726.
32. Mao S, Wang Y, Jiang G, Zhao Z. Effects of levothyroxine therapy on left and right ventricular function in neonates with congenital hypothyroidism: a tissue Doppler echocardiography study. Eur J Pediatr. 2007 Dec;166(12):1261-5. doi: 10.1007/s00431-007-0417-z. Epub 2007 Feb 15. PMID: 17962990.
33. Wu LL, Sazali BS, Adeeb N, Khalid BA. Congenital hypothyroid screening using cord blood TSH. Singapore Med J. 1999 Jan;40(1):23-6. PMID: 10361481.
34. Mahachoklertwattana P, Phuapradit W, Siripoonya P, Charoenpol O, Thuvasethakul P, Rajatanavin R. Five-year thyrotropin screening for congenital hypothyroidism in Ramathibodi Hospital. J Med Assoc Thai. 1999 Nov;82 Suppl 1:S27-32. PMID: 10730514.
35. Adeniran KA, Okolo AA, Onyiriuka AN. Thyroid profile of term appropriate for gestational age neonates in Nigeria: a forerunner to screening for congenital hypothyroidism. J Trop Pediatr. 2010 Oct;56(5):329-32. doi: 10.1093/tropej/fmp143. Epub 2010 Jan 19. PMID: 20085966.
36. Karam GA, Hakimi H, Rezaeian M, Gafarzadeh H, Rashidinejad H, Khaksari M. Thyroid function in mothers who gave birth to neonates with transient congenital hypothyroidism. Pak J Med Sci. 2009;25:568–2.
37. MacGillivray MH. Pediatric Endocrinology. In: Pescowitz OH, Eugster Ea, editors. congenital hypothyroidism. Philadelphia PA: Lippincott Williams Wilkins; 2004. pp. 490–507.
38. Kurtoğlu S, Akın MA. Konjenital hipotiroidizm. In: Kurtoğlu S, editor. Yenidoğan dönemi endokrin hastalıkları. İstanbul: Nobel Matbaacılık; pp. 449–73. 2011.syf.
39. Smith DW, Klein AM, Henderson JR, Myrianthopoulos NC. Congenital hypothyroid dism--signs and symptoms in the newborn period. J Pediatr. 1975 Dec;87(6 Pt 1):958-62. doi: 10.1016/s0022-3476(75)80918-8. PMID: 1237554.