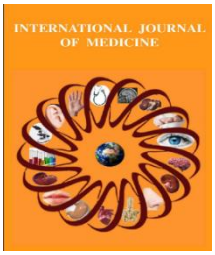
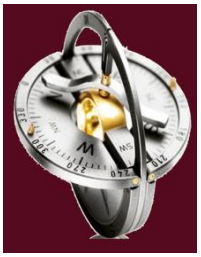


International Journal of Medicine
ISSN (P): 1468-3814; ISSN (e): 2667-7008

https://www.theinternationalmedicine.com



Volume: 7(12) 2025

Original Research

Silent Threats At Birth: Screening For Prevelance Of Congenital Hypothyroidism And G6PD Deficiency In The Malwa Region Of Punjab

Ms Isha Arora[1], Dr. Gesu Singla[2*], Dr. Harijot Bhattal[3], Dr. Saranpal Singh[4], Dr. Shweta Singla[5], Dr. Sanjana Devi[6]
[1]*MBBS Student, Adesh Institute of Medical Sciences and Research, Bathinda*
[2]*Professor and Head, Department of Biochemistry, Adesh Institute of Medical Sciences and Research, Bathinda*
[3]*Professor and Head, Department of Paediatrics, Adesh Institute of Medical Sciences and Research, Bathinda*
[4]*Professor, Department of Biochemistry, Adesh Institute of Medical Sciences and Research, Bathinda*
[5]*Professor, Department of Pharmacology, Adesh Institute of Medical Sciences and Research, Bathinda*
[6]*Assistant Professor, Department of Anatomy, Guru Gobind Singh Medical college & Hospital, Faridkot, Punjab*

Received: 20 Nov 2025 / Accepted: 19 Dec 2025

Abstract

Background: New Born Screening (NBS) is the practice of testing every newborn for harmful or potentially fatal disorders that are otherwise not apparent at birth. Early detection and prompt treatment can make the difference between healthy development or lifelong impairment and possible death. Congenital hypothyroidism is defined as thyroid hormone deficiency present at birth. Glucose-6-phosphate-dehydrogenase deficiency is one of the commonest human enzymopathies, caused by inherited mutations of the X- linked gene G6PD. Being X-linked it is more common in males. ***Materials Used***: The newborns born in the Labour Room/Operation Theatre of tertiary care hospital and all newborns admitted to the Paediatric Department of the tertiary care centre less than 14 days were included in the study with proper informed consent. Data was collected for 4.5 months. Blood samples of the newborns born & admitted neonates were collected. Data was analysed using Microsoft excel. ***Results***: Overall prevalence (percentage) of G6PD deficiency is 0.7% and it was detected only in males. Overall prevalence (percentage) of congenital hypothyroidism is 0.7% and it was detected only in females. ***Conclusions***: The study found out the prevalence of Congenital Hypothyroidism to be 0.7% and G6PD deficiency to be 0.7%. Newborn screening of these disorders has identified these conditions that can affect a child's long-term health or survival. Early detection, diagnosis, and intervention of these disorders can prevent death or disability and enable children to reach their full potential.

Keywords: *Congenital hypothyroidism, G6PD deficiency, neonates, new born screening, prevalence*

Introduction

New Born Screening (NBS) is the practice of testing every infant for dangerous or potentially fatal diseases that are else not apparent at birth. Beforehand discovery and prompt treatment can make the difference between healthy development or lifelong impairment and possible death. Utmost common New Born diseases in India are Prematurity, Respiratory Dysfunction, natural Deformations, Neonatal Infections & Haemolytic conditions of Newborn.[1]

Congenital Hypothyroidism (CHT) is defined as thyroid hormone deficiency present at birth, which could be moreover due to absent or underdeveloped thyroid gland (Dysgenesis) or because of deficient hormone conflation in a well- developed thyroid gland (Dyshormonogenesis). CHT must be diagnosed instantly because detention in treatment can lead to unrecoverable neurological poverties. NBS styles have led to earlier opinion and treatment of CHT performing in bettered neurodevelopmental issues. The thyroid gland secretes the hormones T3 & T4 under the influence of TSH. The thyroid hormones play an essential part in energy metabolism, growth and neurodevelopment. Specifically, it acts on neuronal isolation, synapsis development, & myelination in antenatal & invigorated period, regulating CNS development.[2]

Address for Correspondence Dr. Gesu Singla, Department of Biochemistry, Adesh Institute of Medical Sciences and Research, Bathinda.

DOI: 10.61336/im/25-12-47

G6PD insufficiency- Glucose-6-phosphate-dehydrogenase insufficiency is one of the commonest mortal enzymopathies, caused by inherited mutations of the X- linked gene G6PD. Being X-linked it is more common in males. G6PD insufficiency makes the red blood cells largely vulnerable to oxidative damage, and increased vulnerability to haemolysis. utmost people remain asymptomatic throughout their continuance; still, numerous may develop acute and occasionally veritably severe. Haemolytic Anaemia, when 'touched off' by ingestion of Fava sap, or by several medicines (for e.g. Primaquine, Rasburicase) or infrequently by infection. Acute Haemolytic Anaemia can be managed effectively handed it is instantly diagnosed. [3] Symptoms of Acute Haemolysis associated with G6PD Deficiency include Jaundice, Abdominal/ Back Pain, Fatigue, Haemoglobinuria, Haemolytic Anaemia. [4] This study is planned to screen the babies for these 2 diseases, that is, CHT and G6PD Deficiency & give a detailed description of the results attained. We hope this study can give meaningful information for unborn exploration and eventually contribute to timely and better clinical operation of newborn babies harbouring these diseases.

Materials and Methods

This study is a type of descriptive cross-sectional Study. It was done by Department of Biochemistry in collaboration with the Department of Paediatrics in a tertiary care centre in Malwa region of Punjab. All newborns born in the Labour Room/Operation Theatre of tertiary care hospital and all newborns admitted to the Paediatric Department of the tertiary care centre less than 14 days were included with proper informed consent. Incidence of these disorders was calculated based on the results obtained between the timeline of 4.5 months. Newborns who have received blood transfusion were excluded in the study. Fresh Venous Blood Samples of the newborns were collected. For thyroid profile estimation- plain vials were used and for G6PD estimation- EDTA vials were used.[5] Venous Sample were taken from Day 3 – Day 5 of life.[6] In newborns who were admitted on greater than 5th day and less than 14th day in the hospital their venous Samples were taken on the respective date of admission. Blood samples were processed in the Biochemistry laboratory for the following parameters:

Name Of The Parameters/ Method(S) Used/ Instruments Used

- T3, T4 -COMPETITIVE CHEMILUMINESCENCE IMMUNOASSAY [7] on instrument MAGLUMI SNIBE
- TSH- SANDWICH CHEMILUMINESCENCE IMMUNOASSAY [8] on instrument MAGLUMI SNIBE
- G6PD LEVELS (QUANTITATIVE)- KINETIC [9] on instrument LABSYSTEM DIAGNOSTICS CLINICAL CHEMISTRY ANALYSER [9]

The ones who were screened positive for Congenital hypothyroidism were confirmed by a Confirmatory test by collecting their venous sample within 2 weeks.[10] The results obtained were assessed quantitatively for any possible deficiency (out of the 2 mentioned i.e. CHT and G6PD Deficiency) for each newborn. The parents were counselled regarding the purpose of the study, to find out CHT & G6PD deficiency and hence early management of the same. Proper confidentiality of reports was maintained with respect to each newborn enrolled.

Statistical Analysis: Descriptive analysis of various parameters mentioned in the study was done and the percentage deficiency of the two disorders was calculated via Microsoft Excel and Epi info.

Results

A total of **258 newborns** were enrolled in the study over a period of **4.5 months**. Blood samples were collected for quantitative estimation of **T3, T4, TSH, and G6PD**, and relevant demographic and perinatal data were obtained from the mother or accompanying attendant. After completion of data collection, the compiled data were analyzed and the following results were obtained.

Table I: Distribution Of Neonates On The Basis Of Various Gestational Parameters

Category	Subcategory	Count	Percentage
Gender	Male	124	48%
	Female	134	52%
Gestational Age	Preterm	88	34%
	Term	170	66%
	Postterm	0	0%
Birth Weight	Low	87	33.7%
	Normal	166	64.34%
	High	5	1.93%
Mode of Delivery	NVD	132	51%
	LSCS	126	49%

The screening was done randomly (prior number was not fixed number for males and females) and total of 258 newborns were screened and on analysis it showed that there were 48% males and 52% females. The gestational age was asked to look for any relation whether these disorders are more in preterm born babies as compare to term babies. But no such pattern was seen. Birth weight was also measured to see whether these babies have low or high birth weight. But maximum number of newborns were having normal birth weight which showed that these congenital disorders have no relation with birth weight. The mode of delivery also had no relation with the congenital disorders.

Table II: Mean ±Sd Of Various Thyroid Parameters And G_6PD

Parameter	Mean ± SD
T3	1.33 ± 0.467
T4	10.59 ± 4.11
TSH	3.37 ± 3.55
G6PD	14.33 ± 2.75

T3, T4, TSH, and G6PD levels were analyzed in all neonates, and their **mean ± standard deviation** values were calculated. The majority of neonates had values within the reference range for age.

Table III: Distribution of Neonates (Number and Percentage) Having G6PD Deficiency and Congenital Hypothyroidism on the Basis of Gender

Disease	Males N (%)	Females N (%)	Total (out of 258)
G6PD Deficiency	2 (0.7%)	-	2
Congenital Hypothyroidism	-	2 (0.7%)	2

Discussion

Neonatal screening for common metabolic and inherited disorders is a cornerstone of neonatal care, as early identification and timely intervention can prevent intellectual and physical disabilities and potentially life-threatening complications. The spectrum of disorders screened varies across countries based on prevalence, available resources, and public health priorities. In many high-income countries, including the United States and parts of Europe, universal newborn screening for 40-50 metabolic and genetic disorders is mandatory. Although resource-intensive, the benefits of such programs far outweigh the costs by significantly reducing neonatal morbidity and mortality [11].

In India, neonatal screening is recommended for selected conditions, including congenital hypothyroidism (CH), congenital adrenal hyperplasia (CAH), and glucose-6-phosphate dehydrogenase (G6PD) deficiency [12-19]. Several Indian studies and programs have demonstrated the feasibility and importance of such screening. Augustine et al. [12] and Paul [13] reported the effectiveness of neonatal hearing screening programs in Southern India, emphasizing early detection to prevent long-term deficits. The ICMR Multicentric Study highlighted challenges and successes in screening for CH, CAH, and high-risk infants across India, providing a framework for national-level implementation [14]. Gopalakrishnan et al. [15] demonstrated that screening for CH, galactosemia, and biotinidase deficiency in Uttar Pradesh identified infants requiring early intervention, underscoring the regional need for structured programs. Kaur [16] and Mukhopadhyay & Balachandran [17] emphasized the critical importance and challenges of universal newborn screening in India, including infrastructure, resource allocation, and parental awareness. Verma et al. [18] conducted a prospective study on G6PD deficiency in neonates, reporting variable prevalence based on region and ethnicity. Rama Devi & Naushad [19] reviewed newborn screening programs in India, stressing the urgent need to expand coverage and improve follow-up mechanisms.

In the present study, 258 neonates were screened, comprising 124 males (48.1%) and 134 females (51.9%). Data on gestational age, birth weight, and mode of delivery were collected, but no apparent association was observed between these perinatal factors and the occurrence of CH or G6PD deficiency. This is consistent with the nature of inborn metabolic disorders, which are primarily determined by genetic and enzymatic factors, rather than perinatal characteristics.

The prevalence of congenital hypothyroidism in this study was 0.7% (2/258 neonates), while G6PD deficiency was also 0.7% (2/258 neonates). These findings differ from the study by Verma et al., which reported a birth prevalence of 1 in 2,000 for CH, 1 in 2,500 for CAH, and 1 in 125 for G6PD deficiency [20]. Similarly, Kumari et al. screened 369 neonates and identified one case of elevated TSH, six males with raised 17-OHP, and no cases of G6PD deficiency, highlighting regional variation in prevalence [21]. Patel et al. reported elevated TSH in 1.6% of neonates and G6PD deficiency in 2.6%, with confirmatory tests identifying 4 cases of CH (3.1 per 1,000) and 8 cases of G6PD deficiency (6.2 per 1,000) [22]. Compared to these studies, the prevalence of CH observed in the current study is slightly higher, potentially reflecting regional differences, iodine deficiency, or environmental and genetic factors.

One possible explanation for the elevated prevalence of CH in this region is iodine insufficiency, which persists despite the Universal Salt Iodization Program. Low iodine content in the soil and locally cultivated vegetables contributes to suboptimal intake, increasing the risk of thyroid dysfunction. Additional contributing factors include autoimmune thyroid disorders, genetic predisposition, maternal and environmental stress, and lack of awareness, which may delay diagnosis and treatment.

Although the study detected only a small number of affected neonates, it underscores the importance of early neonatal screening. Timely identification allows for prompt interventions, such as L-thyroxine therapy for CH and avoidance of precipitating drugs or foods in G6PD-deficient neonates, thereby preventing long-term complications like developmental delays, cognitive impairment, or hemolytic crises. Parental counseling regarding disease management and lifestyle precautions is a critical component of screening programs.

Drawbacks of the Study
1. The sample size was relatively small, limiting generalizability. Future studies should include larger, multicentric cohorts.
2. Screening was limited to CH and G6PD deficiency; inclusion of additional metabolic and enzymatic disorders could provide a broader understanding of neonatal health in the region.

Conclusion

India continues to bear a significant burden of congenital hypothyroidism and G6PD deficiency, yet awareness and screening coverage remain limited, leading to preventable complications. The prevalence of G6PD deficiency varies among ethnic groups, emphasizing the need for region-specific research to guide targeted screening and management. Expanding newborn screening programs and developing affordable, scalable strategies, especially in rural and underserved areas, can significantly reduce long-term healthcare costs. Strong epidemiological data are essential to drive policy changes, integrate universal neonatal screening into national health programs, and ensure early diagnosis and treatment. Further research should also evaluate long-term developmental outcomes, dietary and environmental risk factors, and maternal health influences to improve preventive strategies and treatment protocols in India.

Implications of the Study

Early identification of CH and G6PD deficiency allows for timely interventions, preventing long-term complications such as developmental delays, intellectual disability, or life-threatening hemolytic anemia. Screening programs increase community awareness, empower parents and caregivers, and provide guidance on disease management, including dietary precautions and medication avoidance. For CH, proper L-thyroxine administration and follow-up monitoring are essential to ensure favorable neurodevelopmental outcomes. Overall, neonatal screening enhances child health, reduces preventable morbidity, and improves the quality of life for affected infants.

References

1. Gupta HD, Choudhury RG. Neonatal disorders and obstetricians. J Indian Med Assoc. 2001 May;99(5):262-4, 266. PMID: 11676112.
2. Bowden SA, Goldis M. Congenital Hypothyroidism. 2022 Jun 11. In: Stat Pearls [Internet]. Treasure Island (FL): Stat Pearls Publishing; 2022 Jan-. PMID: 32644339.
3. Luzzatto L, Ally M, Notaro R. Glucose-6-phosphate dehydrogenase deficiency. Blood. 2020 Sep 10;136(11):1225-1240. Doi: 10.1182/blood.2019000944. PMID: 32702756.
4. Belfield KD, Tichy EM. Review and drug therapy implications of glucose-6-phosphate dehydrogenase deficiency. Am J Health Syst Pharm. 2018 Feb 1;75(3):97-104. doi:10.2146/ajhp160961. Epub 2018 Jan 5. PMID: 29305344.
5. Leonard JV, Morris AAM. Diagnosis and early management of inborn errors of metabolism presenting around the time of birth. Acha Pediatrica 2006; 95:6-14.
6. Sudhanshu S, Riaz I, Sharma R, Desai MP, Parikh R, Bhatia V. Newborn Screening Guidelines for Congenital Hypothyroidism in India: Recommendations of the Indian Society for Paediatric and Adolescent Endocrinology (ISPAE)- Part 2: Imaging, Treatment and Follow up. Indian J Pediatr.2018;85:448-53.
7. Bowen R (2010-07-24). "Physiological Effects of Thyroid Hormones ". Colorado State University. Retrieved 2013-09-29.
8. Grossman M, Weintraub BD, Szkundlinski MW: Novel Insights into the Molecular Mechanisms of Human Thyrotropin Action: Structural, Physiological and Therapeutic Implications for the Glycoprotein Hormone Family. Endocr Rev 1997;18(4): 203-221.
9. WHO, Tech. Rep. Ser No. 366,1967.
10. Sheikhbahaei S, Mahadaviani B, Abdollahi A, Nayeri F, Serum thyroid stimulating hormone, total and free T4 during the neonatal period; Establishing regional reference intervals. Indian J Endocrinol Metab.2014; 18(1): 39itz
11. Boyle CA, Bocchini Jr JA, Kelly J. Reflections on 50 years of newborn screening. Pediatrics. 2014; 133:961-3.
12. Augustine AM, Jana AK, Kuruvilla KA, Danda S, Lepcha A, Ebenezer J, et al. Neonatal hearing screening – experience from a tertiary care hospital in Southern India. Indian Pediatr. 2014;51 :179-83.
13. Paul AK. Early identification of hearing loss and centralized newborn hearing screening facility - The Cochin experience. Indian Pediatr. 2011; 48:355-9.
14. ICMR Multicentric Study: Newborn Screening for Congenital Hypothyroidism and Congenital Adrenal Hyperplasia and High Risk Screening of Infants. National Task Force of Inborn Metabolic Disorders. Draft report. 2014
15. Gopalakrishnan V, Joshi K, Phadke S, Dabadghao P, Agarwal M, Das V, et al. Newborn screening for congenital hypothyroidism, galactosemia and biotinidase deficiency in Uttar Pradesh, India. Indian Pediatr. 2014; 51:701-5.

16. Kaur M. Newborn screening: The critical importance. Indian Pediatr. 2014;51: 698-9.
17. Mukhopadhyay K, Balachandran B. Universal newborn screening – Is it going to be a reality in India? Indian Pediatr. 2014;51: 697-8.
18. Verma M, Singla D, Crowell SB. G6PD deficiency in neonates: prospective study. Indian J Pediatr. 1990;57: 385-8.
19. Rama Devi AR, Naushad SM. Newborn screening in India. Indian J Pediatr. 2004;71: 157-60.
20. Verma J, Roy P, Thomas DC, Jhingan G, Singh A, Bijarnia-Mahay S, Verma IC. Newborn Screening for Congenital Hypothyroidism, Congenital Adrenal Hyperplasia, and Glucose-6-Phosphate Dehydrogenase Deficiency for Improving Health Care in India. J Pediatr Intensive Care. 2020 Mar;9(1):40-44. doi: 10.1055/s-0039-1698424. Epub 2019 Oct 14. PMID: 31984156; PMCID: PMC6978160.
21. Kumari, Bandana; Raj, Khushboo; Sharma, Sadhana; Kumar, Sushil; Chowdhry, Bhabesh K; Kumar, Amit. Newborn screening for congenital hypothyroidism, congenital adrenal hyperplasia and glucose-6-phosphate dehydrogenase deficiency in Bihar: A pressing priority in today's time. Journal of Family Medicine and Primary Care 12(12): p 3332-3338, December 2023. | DOI: 10.4103/jfmpc.jfmpc_1029_231.
22. Patel S, Priya R, Padhi P, et al. Prevalence of Congenital Hypothyroidism and G6PD Deficiency in Newborns in a Tertiary Care Hospital of Central India. Journal of Neonatology. 2021;35(1):5-9. doi:10.1177/0973217920987713.