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# Period Prevalence of Inborn Metabolic Errors in Neonates Admitted in NICU in Uttarakhand: A Cross-sectional Period Prevalence Study

**Sunakshi Bhatt[1], Anita Sharma[2], Chinmay Chetan[3]**

[1]Medical Undergraduate, [2]Department Biochemistry, [3]Department of Neonatology,
Himalayan Institute of Medical Sciences, Swami Rama Himalayan University, Medical Undergraduate,
Himalayan Institute of Medical Sciences, Swami Rama Himalayan University, Dehradun, Uttarakhand,
India- 248016

Corresponding Author: Dr. (Professor) Anita Sharma

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## ABSTRACT

**Background:** Inborn errors of metabolism in neonates represent a critical subset of genetic disorders that can lead to life-threatening complications if not promptly diagnosed and managed. Early identification through newborn screening enables timely therapeutic intervention, significantly reducing morbidity and mortality associated with metabolic crises and neurodevelopmental impairment.
**Objectives:** To determine the prevalence of inborn errors of metabolism in neonates admitted in NICU in Garhwal, Uttarakhand, India. This pilot study assessed the regional prevalence of specific congenital metabolic disorders in newborns through universal screening of all neonates after stabilization just prior to discharge and age ranged from 3 to 48 days admitted to hospital in underserved area of Uttarakhand.
**Methods:** A cross-sectional period prevalence study was conducted at a private medical college in the state of Uttarakhand. A total of 410 neonates were enrolled in this study. Glucose-6-Phosphate Dehydrogenase deficiency, Congenital Hypothyroidism, Congenital Adrenal Hyperplasia, Biotinidase Deficiency, Maple Syrup Urine Disease, Phenylketonuria, and Galactosemia. Data was analysed statistically with SPSS software.
**Results:** During initial screening, a total of 17 neonates tested positive. Out of 17, 9 neonates showed G6PD deficiency, 4 showed CAH, and 2 neonates each were diagnosed with CH and BTD. Following diagnostic confirmation, G6PD deficiency exhibited the highest prevalence, accounting for 4 cases (0.98%). In contrast, CAH and CH were each identified in a single case (0.24%), indicating comparatively lower incidence rates.
**Conclusions:** The findings of this cross-sectional period prevalence pilot study underscore the clinical implication of use of universal newborn screening for congenital metabolic disorders in a tertiary care setting. The observed prevalence rates highlight the importance of early detection strategies in identifying asymptomatic neonates at risk, thereby facilitating timely therapeutic interventions and informing public health policy for expanded screening programs in resource-limited regions such as Uttarakhand.

***Keywords:*** New born screening, Neonates, Glucose-6-Phosphate Dehydrogenase, Congenital Hypothyroidism, Congenital Adrenal Hyperplasia, Biotinidase, Maple Syrup Urine Disease, Phenylketonuria, Galactosemia

## INTRODUCTION

Inborn errors of metabolism are defined as defects in metabolic pathways resulting from deficiencies in enzymes, their coenzymes, or transporters. These defects lead to the accumulation of upstream substrates and a deficiency of downstream products in the metabolic pathway[1]. This atypical accumulation of precursors along with absence of requisite intermediates resulted in metabolic process abnormality. Alternatively, this may also induce creation of alternative metabolic pathways, which can result in generation of abnormal compounds. These metabolic alterations can occur during metabolic and/or energy synthesis or catabolism, inducing metabolic disorders [2]. To date, approximately 500 inborn errors of metabolism have been identified, and this number continues to grow. The frequency of these disorders varies significantly between countries and even among different regions within a country [1]. Although, prevalence of these metabolic disorder is individually rare (less than 1:100,000 cases), due to occurrence of multiple diverse disorders, the composite occurrence ranges between 1:800 – 1:2500 in live-born children [3].

Consequently, certain countries like, Germany, Netherlands, USA, UK, have made a standard schedule to carry out neonatal screening for metabolic diseases like, phenylketonuria (PKU), Congenital Hyperthyroidism (CH), deficiency of medium chain acyl-Co A dehydrogenase etc, although the battery of tests differ in each country [4-6].

In India, the overall prevalence of inborn errors of metabolism is estimated to be around 1 in 2,497 live births, with the highest incidence observed for Glucose-6-Phosphate Dehydrogenase (G6PD) deficiency and CH, with reported incidences of 2–7.8% and 2.1 per 1,000 live births, respectively [7,8]. Individuals with metabolic disorders may present with abnormal growth, developmental delays, cardiac abnormalities, neurological disorders, cataracts, organic acidurias, and feeding difficulties in infancy. However, many of these complications can be prevented or mitigated with early diagnosis and timely treatment using appropriate supplementation [9,10]. Hence, screening of metabolic disorders at an early age will significantly curtail morbidity and mortality. Neonatal screening for metabolic disorders is an effective method for early detection and intervention, which can significantly improve the quality of life in affected individuals [11,12].

To date, no study has been conducted in the state of Uttarakhand to assess the prevalence of congenital metabolic disorders. Therefore, this cross-sectional period prevalence pilot study was designed to estimate the prevalence of selected seven inborn metabolic errors in more than 400 newborns through universal screening of all neonates admitted to a tertiary care hospital in Uttarakhand over a period of one year from August 2022 to September 2023. After getting ethical clearance, data was collected retrospectively till August 2022 and prospectively till September 2023. This will help in elucidating the prevalence of metabolic disorders in the underserved area of Uttarakhand region lacking awareness on metabolic disorders in neonates. This will facilitate to establish the necessity of neonatal screening in order to address the health challenges of the region and developing country thereby. Further, the findings of this study may serve as a foundation for future research with a larger sample size.

## MATERIALS & METHODS

### Ethical Approval

This cross-sectional period prevalence study was conducted at a private medical college; Swami Rama Himalayan University, Dehradun, in the state of Uttarakhand, India. Data was collected over a one-year period, from August 2022 to September 2023. The study was approved by the Institutional Ethics Committee (IEC No: SRHU/HIMS/PHARMA/E-1/2023/7, dated 25-01-2023).

**Subjects details**

Neonates (age: 3 to 48 days) admitted to the Neonatal Intensive Care Unit (NICU) during the study period were included. As part of the NICU protocol, counselling regarding the basic newborn screening panel comprising seven selected metabolic disorders including G6PD deficiency, CH, CAH, BTD, MSUD, PKU, and Galactosemia was provided to parents prior to discharge. After obtaining informed parental consent, blood samples were collected and dried on Whatman 903 filter paper. The dried samples were stored carefully at ambient temperature and analyzed using ELISA or rapid testing kits as per the methods specified with the kits.

**Screening using colorimetric assays**

All the analyses were performed on BornSafe® NBS LAB Microplate Reader (Tulip Diagnostics Private Ltd, Goa, India), a microprocessor-controlled photometric system capable of reading and calculating assay results from microtiter plates. The principles of analysis and the procedures are given below.

***G6PD testing***

BornSafe® G6PD kit was used for the assay and procedure was performed as per the instructions supplied with the kit.[13] The neonatal G6PD screening was done on dried blood spots specimens (cellulose paper) eluted in a buffer. After the elution step, the eluate containing G6PD is incubated with a reagent containing G6P, which in the presence of NADP, catalyses the oxidation of G6P to 6-phosphogluconate. The NADPH produced reacts with a colour reagent in which a tetrazolium salt gets reduced producing a distinct colour. The developed color, directly proportional to the concentration of G6PD was measured at 550 nm on BornSafe® NBS LAB Microplate Reader (Tulip Diagnostics Private Ltd, Goa, India).

***CH testing***

CH neonatal screening was based on measurement of elevated levels of Thyroid-Stimulating Hormone (TSH) in a dried blood spot sample using enzyme immunoassay kit of BornSafe® Neonatal TSH Screening ELISA [13]. The procedure was performed as per the instructions supplied with the kit. Sandwich ELISA testing involved mixing of dried blood spots with Elution Reagent which results in reaction between the eluted antigen and immobilized anti-TSH antibody to form an antigen-antibody complex. After suggested incubation period, unreacted reactants and remnants were washed off via wash step. Another antibody (directed at a different epitope) labelled with an enzyme was added. Another interaction occurs to form an enzyme labelled antibody-antigen-antibody complex on the surface of the wells. Excess enzyme was washed off via a wash step. A suitable substrate was added to produce color measured on BornSafe® NBS LAB Microplate Reader (Tulip Diagnostics Private Ltd, Goa, India). The enzyme activity on the well was directly proportional to the TSH concentration in the dried blood spot.

***BTD testing***

Biotinidase screening, an enzymatic assay for the quantitative determination of biotinidase activity in neonates was performed using blood spot samples dried on Whatman 903 filter paper. The procedure was performed as per the instructions supplied with the BornSafe® Biotinidase screening kit. Biotinidase from cellulose paper (dried blood spot samples) was extracted with a buffer containing the artificial substrate biotin 4-amidobenzoic acid (B-PAB). The biotinidase enzyme catalyses the release of free 4-amidobenzoic acid (PABA) from B-PAB. The reaction was stopped by trichloroacetic acid which denatured the biotinidase. The PABA produced reacted with sodium nitrite to form a diazotized compound (the nitrite in excess was neutralized with the ammonium

sulfamate); after that the diazotized compound reacted with the NED to form an azo-dye. The purple colour produced was measured colorimetrically on BornSafe® NBS LAB Microplate Reader (Tulip Diagnostics Private Ltd, Goa, India) at 550 nm.

***PKU testing***

BornSafe® Neonatal PKU screening was performed using an enzymatic assay for the quantitative determination of phenylalanine concentrations in neonates using blood spot samples dried on Whatman 903 filter paper [13]. The Phenylalanine from cellulose paper (dried blood spot samples) was extracted with trichloroacetic acid containing elution buffer supplied with the kit. After extraction, the eluted sample was combined with the enzyme reagent Phenylalanine dehydrogenase. This enzyme reagent catalysed the NAD-dependent oxidative deamination of Phenylalanine to phenylpyruvate and ammonia. The NADH produced, reacted with a colour reagent in which tetrazolium salt got reduced producing a distinct colour. This colour was measured colorimetrically with a photometer at 550 nm on BornSafe® NBS LAB Microplate Reader (Tulip Diagnostics Private Ltd, Goa, India).

***Galactosemia testing***

We used BornSafe® Neonatal Total Galactose Screening kit based on enzymatic assay for the quantitative determination of total galactose (galactose and galactose-1-phosphate) concentrations in neonates using blood spot samples dried on Whatman 903 filter paper [13]. The total galactose (galactose and galactose-1-phosphate) from cellulose paper (dried blood spot samples) was extracted with trichloroacetic acid containing elution buffer supplied with the kit. After extraction, the eluted sample was combined with the enzymes-coenzyme reagent containing alkaline phosphatase, galactose dehydrogenase and NAD. Galactose-1-phosphate got converted to galactose by alkaline phosphatase. Galactose dehydrogenase oxidized the galactose to galactonolactone reducing NAD to NADH. The NADH produced was reacted with a colour reagent in which tetrazolium salt gets reduced producing a distinct colour which was measured colorimetrically with a photometer at 550 nm on BornSafe® NBS LAB Microplate Reader (Tulip Diagnostics Private Ltd, Goa, India).

***MSUD testing***

BornSafe® Neonatal MSUD Screening assay is based on enzymatic colorimetric quantitative detection of MSUD in new born using blood spot samples dried on Whatman 903 filter paper [13]. This test measures the L-branched-chain amino acid concentrations (BCAA). Leucine and BCAAs from cellulose paper (dried blood spot samples) extracted with trichloroacetic acid containing elution buffer supplied with the kit. After extraction, the eluted sample was combined with the enzyme reagent Leucine dehydrogenase. This enzyme reagent catalysed the NAD-dependent oxidative deamination of Leucine and L-BCAAs to α-ketoisocaproate acid. The NADH produced, reacted with a colour reagent in which tetrazolium salt gets reduced producing a distinct colour which was measured colorimetrically with a photometer at 550 nm on BornSafe® NBS LAB Microplate Reader (Tulip Diagnostics Private Ltd, Goa, India).

***CAH testing***

BornSafe® Neonatal 17 Alpha-Hydroxyprogesterone (17-OHP) ELISA based enzyme immunoassay kit was used for the quantitative determination of 17-OHP in blood samples dried on Whatman 903 filter paper [13]. The essential reagents required for the assay in the kit included antibody, enzyme- antigen conjugate and native antigen. Upon mixing the antibody with blood spot containing the antigen, the enzyme conjugate after specified period of time mentioned in the kit procedure. This delayed addition permitted increased

sensitivity for low concentration samples and better precision. Upon the addition of the enzyme conjugate, competition reaction results between the enzyme analog and the antigen in the sample for a limited number of antibody binding sites (not consumed in the first incubation). Simultaneously, the immune complex was immobilized through the interaction with Anti-Rabbit antibody coated to the well. Unbound reactants (17-OHP and 17-OHP-HRP) were removed at the end of the incubation time. A substrate was then reacted with the enzyme bound on the wall of the microwells followed by termination of reaction with addition of acid. The end product is measured at 450 nm on on BornSafe® NBS LAB Microplate Reader (Tulip Diagnostics Private Ltd, Goa, India).

Neonates who tested positive in the initial screening were recalled for follow-up. Venous blood samples were collected and analysed to confirm the diagnosis.

## Statistical Analysis

Data was collected and entered into Microsoft Excel 2018. Statistical analysis was performed using SPSS software version 22. Assuming an expected proportion of 50% (due to unknown prevalence), a minimum sample size of 100 newborns was required, calculated using the Chi-square test :

$N = [(Z_{1}\text{-}\alpha/2)^2 \times P(1–P)] / d^2$

Where:

$Z_{1}\text{-}\alpha/2 = 1.96$ for a 95% confidence interval

P = anticipated prevalence (0.5)

d = margin of error (10%)

Qualitative and categorical variables were analysed by calculating frequencies and percentages. The overall prevalence of metabolic disorders was also calculated as a percentage. One-tailed Binomial Test is applied to calculate p value, <0.05 value was considered significant.

## RESULT

Inborn errors of metabolism are a group of rare genetic disorders that, if left undetected, can lead to developmental delays, disabilities in learning, chronic illness, or even death in infants [14,15]. Several metabolic errors are asymptomatic at birth but can rapidly progress to serious conditions such as seizures, coma, developmental delays, or death. Early detection allows for timely intervention before these symptoms manifest [16]. While neonatal screening for inborn errors of metabolism has become routine in many developed countries, its implementation in India remains uneven, especially in smaller districts where healthcare infrastructure is limited and public awareness is also low. Although, India is making progress in healthcare sector, still face several challenges in implementing widespread neonatal screening for inborn errors of metabolism. Programs are often limited to tertiary hospitals or pilot initiatives. In the underserved areas of Garhwal region in Uttarakhand, the absence of early diagnosis would result in treatable conditions go unnoticed until irreversible damage has occurred. Early diagnosis of inborn errors of metabolism through newborn screening is important because it can significantly prevent the irreversible damage before symptoms even appear and improve the quality of life of children and also reduce mortality [16].

Our study is an initiative in one such region of India wherein medical infrastructure is limited and will help to set an example to introduce importance of neonatal screening. A total of 410 neonates were enrolled in the study. Amongst them seventeen were found to be screen-positive cases (Table 1, Figure 1). The screen-positive cases had a median age of 36.6 weeks (33.5 to 38.5 weeks), 82.4 % of them were males (Table 2). Majority of the screen positive cases had a median gestational age of 36.6 weeks (33.5 to 38.5 weeks). Almost 41.2% (n=7) had been delivered through vaginal delivery. The median birth weight of these 17 cases was 2133 grams (1698.5-2702.5 grams). Most of the screen-positive cases had appropriate weight for gestational age (n=9; 53%) and almost 41.2% (n=7) had small

weight for gestational age while one case had large weight for gestational age (n=1; 5.9%). Amongst them, there was a family history of primgravida in 23.5% (n=4) cases, 17.6% (n=3) had hypothyroidism and 5.9% (n=1) each of diabetes and hypertensive disorder (Table 3).

Post initial blood analysis, the cases that tested positive for potential metabolic disorder were further subjected to follow up venous blood tests after obtaining suitable consent from parents (Figure 1). Of the 17 screen-positive cases, 6 were further confirmed through follow-up diagnostic testing with venous samples (Table 1). The baseline demographic characteristics of both the neonates and mothers, corresponding to screen-positive and confirmed-positive infants, are comprehensively summarized in Tables 2 and 3, respectively. The median gestational age of these six neonates was found to be 35.9 weeks (33.4-40 weeks) and all of them were males. Only 33.3% of them had been delivered through vaginal delivery. While, the median birth weight of these cases was 2337.5 grams (1950-2670 grams). Amongst them, 66.6% (n=4) of them had appropriate weight for the gestational age while 33.3% (n=2) had small weight for gestational age. One each of the confirmed positive case had a maternal history of hypothyroidism and diabetes. The positive cases from the repeat tests were then referred to a specialist for improving health.

In the cohort, the overall prevalence of confirmed inborn errors of metabolism in the study was found to be 1.46%, with a highly significant p value of 0.00386. Primarily, three metabolic disorders were observed in the cohort, viz., G6PD deficiency, CAH, and BTD. The most prevalent metabolic disorders found were G6PD deficiency, and CAH. The deficiency of G6PD ($p < 0.0001$) and CAH ($p < 0.01$) was found to be significantly higher as compared to CH, BTD, PKU, Galactosemia, and MSUD (Table 1).

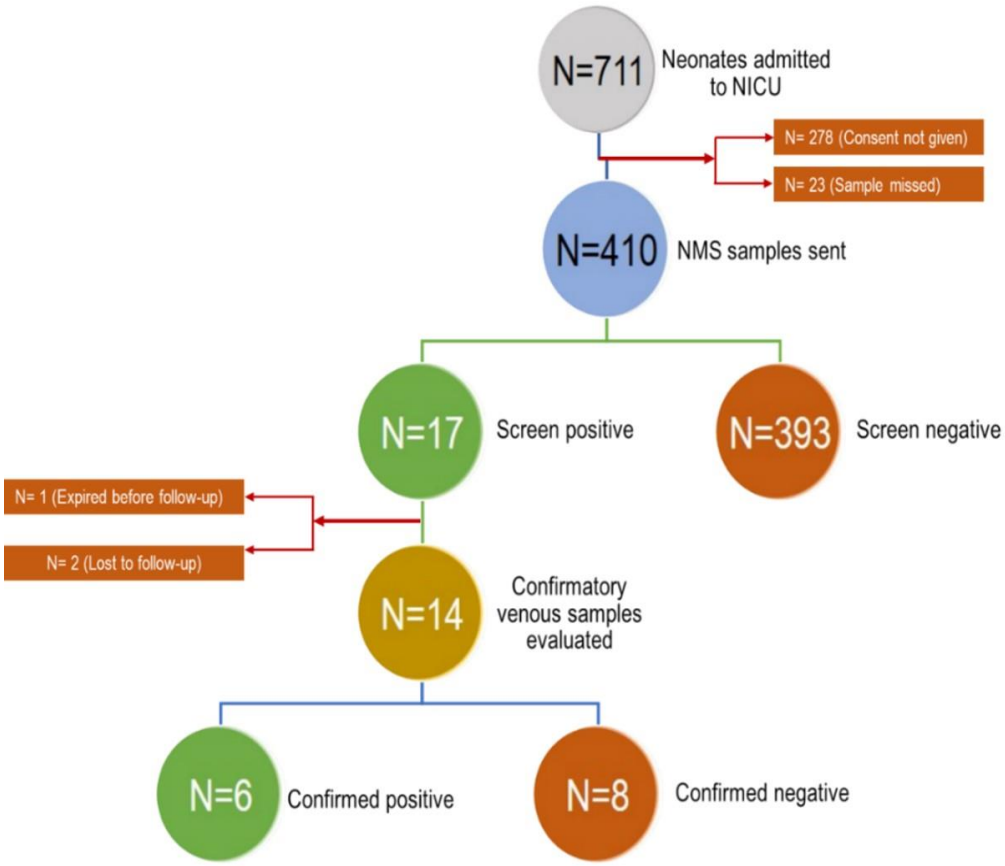


**Figure 1. Flow diagram illustrating the process of new born screening for metabolic disorders.**

**Table 1. Results of screening and the confirmatory tests for metabolic disorders in neonates (n=410).**

| Disease | Screening positive | Confirmatory sample | Prevalence (%) | *p* value* |
|---|---|---|---|---|
| G6PD Deficiency | 9 | 4 | 0.98 | 0.000194 |
| CAH | 4 | 1 | 0.24 | 0.0203 |
| CH | 2 | 1 | 0.24 | 0.127 |
| BTD | 2 | 0 | 0 | 0.993 |
| MSUD | 0 | 0 | 0 | 0.998 |
| PKU | 0 | 0 | 0 | 0.960 |
| Galactosemia | 0 | 0 | 0 | 0.993 |
| Total | 17 | 6 | 1.46 | 0.00386 |

***Values in bold type indicate significant (p <0.05)***

**Table 2. Demographic profile (Neonatal characteristics) of screen positive (n=17) and confirmed cases (n=6)**

| Neonatal characteristics | | Screen positive cases | Confirmed cases |
|---|---|---|---|
| Gestation age (weeks) | | 36.6 (33.5-38.5)* | 35.9 (33.4-40)* |
| Birth weight (grams) | | 2133 (1698.5 - 2702.5)* | 2337.5 (1950-2670)* |
| Male gender, n (%) | | 14 (82.4%) | 6 (100%) |
| Weight for gestation n (%) | Appropriate | 9 (53%) | 4 (66.6%) |
| | Small | 7 (41.2%) | 2 (33.3%) |
| | Large | 1 (5.9) | 0 |

***Values indicate Median (Inter Quartile Range)***

**Table 3. Demographic profile (Maternal characteristics) of screen positive (n = 17) and confirmed cases (n=6)**

| Maternal characteristics | Screen positive cases (%) | Confirmed cases (%) |
|---|---|---|
| Primigravida | 4 (23.5%) | 0 (0%) |
| Hypothyroidism | 3 (17.6%) | 1 (16.7%) |
| Diabetes | 1 (5.9%) | 1 (16.7%) |
| Hypertensive disorder | 1 (5.9%) | 0 (0%) |
| Vaginal delivery | 7 (41.2%) | 2 (33.3%) |

## DISCUSSION

Neonatal screening for enzymatic disorders has gained momentum in India in the recent years. It is driven by the need to detect manageable inborn errors of metabolism early stage of life to reduce the morbidity and mortality. However, many developing countries, including India, lack a universal newborn screening program for the early diagnosis of metabolic disorders. Despite over 30 years of pilot initiatives, only a few regions—namely Chandigarh, Goa, and Kerala—have successfully implemented and maintained such programs. Currently, only about 3% of newborns in India are screened, largely due to inadequate healthcare infrastructure, financial constraints, and limited diagnostic capabilities. This underscores the urgent need to incorporate newborn screening into India's national health programs [17]. Given the lack of prior studies in the Indian state of Uttarakhand, this pilot study was conducted to assess the prevalence of seven common metabolic disorders at a tertiary care center in Dehradun.

G6PD deficiency results from a lack of the enzyme glucose-6-phosphate dehydrogenase, which protects red blood cells from oxidative damage. A deficiency of G6PD can lead to acute hemolytic anemia, particularly during episodes of oxidative stress [18]. In our study, the prevalence of G6PD deficiency was 0.98%. This is consistent with previous findings—such as a 2017 study at Sir Ganga Ram Hospital in India reporting a prevalence of 0.8% (1 in 125) [19] , and a 2009 study from a Chandigarh medical college reporting 0.9% (1 in 112) [20]. However, a study in Andhra Pradesh showed a much lower rate of G6PD deficiency (1 in 2,200), with higher incidences of congenital hypothyroidism and CAH [21]. In tribal

populations of Central India, a study reported 8.9% prevalence of G6PD deficiency in neonates [22]. This regional disparity could be influenced by environmental and genetic factors unique to different regions of India.

Newborns with CH typically appear normal at birth, which can delay diagnosis. However, without timely detection, CH can result in severe developmental issues, including intellectual disability [23]. According to a study published in 2022 based on analysis performed in 2018, the prevalence of CH in India 72 cases per 100,000 live births equivalent to 1 in 1,388 (0.072%). Our study found a CH prevalence of 0.24% in Garhawal region, which is more 300% compared to earlier study of 2018 [24]. This further emphasises the regional differences observed in the prevalence of CH and the necessity for screening for metabolic disorders in neonates.

CAH, often caused by a deficiency in the enzyme 21-hydroxylase, leads to impaired cortisol synthesis and an accumulation of 17-hydroxyprogesterone (17-OHP). The global incidence of CAH ranges from 1 in 10,000 to 1 in 15,000 live births. Early screening is crucial, as CAH can be life-threatening and, in some cases, result in incorrect sex assignment in virilized female newborns [25]. In our study, the prevalence of CAH was also 0.24%, although more cases were detected initially through screening. Similarly, a study in Andhra Pradesh found CAH more common than CH, with incidences of 1 in 1,700 and 1 in 2,575, respectively [21].

An ICMR-supported study reported the prevalence of CH at 0.138% and CAH at 0.017%, but also noted significant regional variation in CAH prevalence [26]. A meta-analysis revealed that CH affects approximately 1 in 1,031 full-term neonates in non-endemic Indian regions. Factors such as iodine deficiency, environmental toxins, and autoimmune conditions may contribute to regional variation [27].

A review from Sweden observed a notable increase in CAH detection rates following the introduction of newborn screening. In India, a recent cohort study reported a CAH incidence of 1 in 5,762, with regional variation—from 1 in 2,036 in Chennai to 1 in 9,983 in Mumbai. In Andhra Pradesh, the rate was 1 in 2,600, while a six-year study in Bangalore found an incidence of 1 in 2,800 [28]. These findings highlight the influence of geography and environmental exposure on genetic disorders.

Biotinidase is an enzyme that recycles biotin, a coenzyme necessary for carboxylation reactions. Biotinidase deficiency is an autosomal recessive disorder that can cause developmental delays, skin rashes, hearing loss, and respiratory issues. Although the global incidence is approximately 1 in 60,000, early diagnosis is critical, as biotin supplementation can significantly improve dermatological symptoms and prevent complications [29,30]. In our study, no cases of biotinidase deficiency were identified, which is consistent with findings from Andhra Pradesh. In contrast, a study in the southwestern Netherlands reported biotinidase deficiency prevalence ranging from 1 in 6,100 to 1 in 8,200 [31], and an Italian study reported a rate of 1 in 5,996 [32].

Our study did not detect any cases of MSUD, PKU, and Galactosemia. Globally, also these disorders are rare. In line with this, a study in China reported an MSUD incidence of 1 in 219,472,[33] while an 11-year study in the Netherlands found it to be 1 in 491,000 [34]. Reported incidences for MSUD include 1 in 220,219 in the U.S., 1 in 119,573 in Germany, and 1 in 59,426 in Kuwait [35].

PKU results from a deficiency in the enzyme phenylalanine hydroxylase, disrupting the metabolism of phenylalanine. Left untreated, PKU can cause irreversible cognitive impairment, reduced motor control, and impaired verbal function. A global meta-analysis estimated PKU prevalence ranging from 0.3 to 38.13 per

100,000 live births, with notable regional differences—from 0.3 in Thailand to 38.13 in Turkey [36]. Another study estimated the global PKU prevalence at 0.00641%, with Italy having the highest rate (0.02736%) and Thailand the lowest in Asia (0.0004%) [37].

Galactosemia is a rare autosomal recessive disorder where infants cannot convert galactose from milk into glucose due to a galactose enzyme deficiency, and it can be detected at birth through newborn screening. Galactosemia is managed with a galactose- and lactose-free diet, but long-term complications may still occur [38].The global prevalence of classical galactosemia ranges from 1 in 30,000 to 1 in 75,000. In India, the true incidence remains unclear due to limited sample sizes. Recent screenings in Uttar Pradesh and Andhra Pradesh found no confirmed cases despite evaluating 13,500 and 10,300 newborns, respectively [39].

A pilot study from Udupi district in South India reported the following incidences: CH: 1 in 887, G6PD Deficiency: 1 in 118, CAH: 1 in 4,591, BTD: 1 in 531, Galactosemia: 1 in 4,236, PKU: 1 in 5,203. However, these estimates are based on limited data and may not accurately represent the true nationwide incidence [40]. Considering the overall results from studies conducted across the India, the way forward in is to prioritize regions identified endemic to particular inborn errors of metabolism and implement a compulsory neonatal screening program in these areas [41].

## CONCLUSION

The results of the newborn screening revealed a significant proportion of positive cases of metabolic disorders. This underscores the urgent need to implement newborn screening tests as a standard practice in the state. Early diagnosis and timely treatment of metabolic disorders in neonates can prevent serious health complications that may otherwise have long-term impacts on a child's life.

There is a clear opportunity to incorporate such screening protocols into the National Health Policy to enhance child health outcomes across the country. The health of all citizens—especially children—is a critical resource, as a nation's human capital is a key driver of its development.

### *Declaration by Authors*

**Ethical Approval:** Approved. The study was approved by the Institutional Ethics Committee (IEC No: SRHU/HIMS/ PHARMA/E-1/2023/7, dated 25-01-2023).

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**Conflict of Interest:** The authors declare no conflict of interest.

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