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POPULATION-BASED NEWBORN SCREENING BY DRIED BLOOD SPOT METHOD: A PROSPECTIVE OBSERVATIONAL STUDY FROM NORTH KARNATAKA

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ABSTRACT

Background: Newborn screening (NBS) using dried blood spot (DBS) methodology is a well-established public health strategy for early detection of treatable congenital disorders. Despite its proven benefits, implementation in resource-limited settings remains variable, necessitating region-specific feasibility studies.

Objective: To evaluate the feasibility, screening yield, and associated maternal and demographic factors of a mandatory DBS-based newborn screening program in a tertiary care setting in North Karnataka.

Methods: This prospective observational study included 1,000 newborns screened between January and December 2024 at S. Nijalingappa Medical College, Bagalkote. DBS samples collected between 24–72 hours of life were analyzed for phenylketonuria (PKU), glucose-6-phosphate dehydrogenase (G6PD) deficiency, galactosemia, congenital hypothyroidism (TSH), and hemoglobinopathies using standardized laboratory protocols. Maternal and sociodemographic data were recorded. Statistical analysis was performed using chi-square tests, with $p < 0.05$ considered significant.

Results: The overall screen-positive rate was 8.7% (87/1,000). Congenital hypothyroidism was the most common abnormality (3.5%), followed by G6PD deficiency (2.2%), galactosemia (1.2%), hemoglobinopathies (1.0%), and PKU (0.8%). A statistically significant association was observed between consanguinity and screen-positive outcomes ($p = 0.002$), while maternal age, socioeconomic status, residence, and maternal medical history showed no significant association. No significant correlation was found between screening positivity and sex or place of residence. Implementation of repeat DBS testing improved diagnostic accuracy and reduced false-positive referrals.

Conclusion: Mandatory DBS-based newborn screening is feasible and effective in a resource-limited setting, with a high screening yield and strong relevance in populations with prevalent consanguinity. Expansion of such programs can significantly reduce preventable morbidity and mortality through early diagnosis and intervention.

KEYWORD: Newborn screening, dried blood spot, congenital hypothyroidism, G6PD deficiency, metabolic disorders, consanguinity, India.

INTRODUCTION

Newborn screening (NBS) is a population level public health initiative designed to identify, shortly after birth, infants at high risk for a set of serious, often treatable congenital disorders so that early interventions can prevent irreversible morbidity or death. Over the past several decades NBS has evolved from single-disorder tests to expanded panels that screen for metabolic, endocrine, hematologic, and emerging genetic conditions; programs that use dried blood spot (DBS) microsamples account for the vast majority of contemporary screening worldwide and have demonstrated substantial population-level health benefits. [1]

Newborn screening (NBS) is a population-level public-health intervention that aims to identify asymptomatic neonates at elevated risk for selected congenital disorders for which early diagnosis and treatment substantially reduce mortality, prevent irreversible disability, or improve long-term outcomes. Modern NBS programs therefore shift the clinical paradigm from reactive treatment of symptomatic disease to early identification and preemptive management, with benefits measured at both individual and population levels (reduced neurodevelopmental impairment, fewer emergency admissions, lower lifetime disability burden).[2]

The dried blood spot (DBS) technique — collection of capillary heel-prick blood onto standardized filter paper, drying, and centralized laboratory testing — is the dominant specimen method worldwide because it is minimally invasive,

cost-efficient, compatible with high-through output automation, and robust to transport and storage conditions compared with liquid samples. DBS cards can be archived for confirmatory testing and quality assurance, and are compatible with multiple analytic platforms (tandem mass spectrometry, enzyme assays, immunoassays, PCR-based molecular tests). These properties make DBS especially suitable for national and regional programs, including resource-constrained settings, while supporting expansion of screening panels.[3]

A standard expanded DBS-based panel screens for disorders across several categories:

- Endocrine: congenital hypothyroidism (CH), congenital adrenal hyperplasia (CAH).
- Metabolic: phenylketonuria (PKU), aminoacidopathies, organic acidurias, fatty-acid oxidation disorders (detectable by MS/MS).
- Hematologic / hemoglobinopathies: sickle cell disease and severe thalassemia genotypes.
- Emerging molecular targets: severe combined immunodeficiency (SCID), spinal muscular atrophy (SMA), selected lysosomal storage disorders.

Early identification of CH and CAH alone prevents readily-measurable adverse outcomes such as intellectual disability and life-threatening adrenal crisis respectively; metabolic disorders detected by tandem MS/MS can prevent acute metabolic decompensation and improve neurodevelopmental outcomes with dietary or medical interventions.[4]

Incidence- recent data

Quantitative incidence estimates help prioritize conditions for screening and model clinical and economic impact. Selected recent estimates (presented as per 10,000 live births here for easy comparison) are:

- Congenital hypothyroidism (India meta-analysis): ~9.69 per 10,000 (approx. 1 in 1,031 live births). This meta-analysis suggests India's CH prevalence is higher than typical global averages and variable across regions.
- Congenital adrenal hyperplasia (India reporting): ~1.74 per 10,000 (approx. 1 in 5,762) in large Indian cohorts — substantially higher than some global point-estimates; CAH screening can prevent early mortality (salt-wasting crisis) and reduce morbidity.
- Phenylketonuria (PKU; recent screening study): ~1.33 per 10,000 live births in a contemporary screened cohort. Though lower incidence than CH, PKU is a classical NBS target because effective lifelong dietary management prevents severe intellectual disability.
- Combined inherited metabolic disorders (example regional program — Udupi, Karnataka): ~45.45 per 10,000 (1 in 220) for any one of the screened Inborn Errors of Metabolisms(IEMs) in a regional pilot, illustrating how regional/ethnic clustering and targeted panels can reveal much higher combined incidence for treatable IEMs in certain populations. Such local data strongly support pilot and regional screening programs that reflect local epidemiology. (Figure 1,2)

These values illustrate two important points: (1) single-condition incidences vary widely (so national panel selection must be evidence-driven and flexible), and (2) combined incidence for groups of IEMs in a screened panel can be high enough to justify screening from a population-health perspective in many regions.[5]

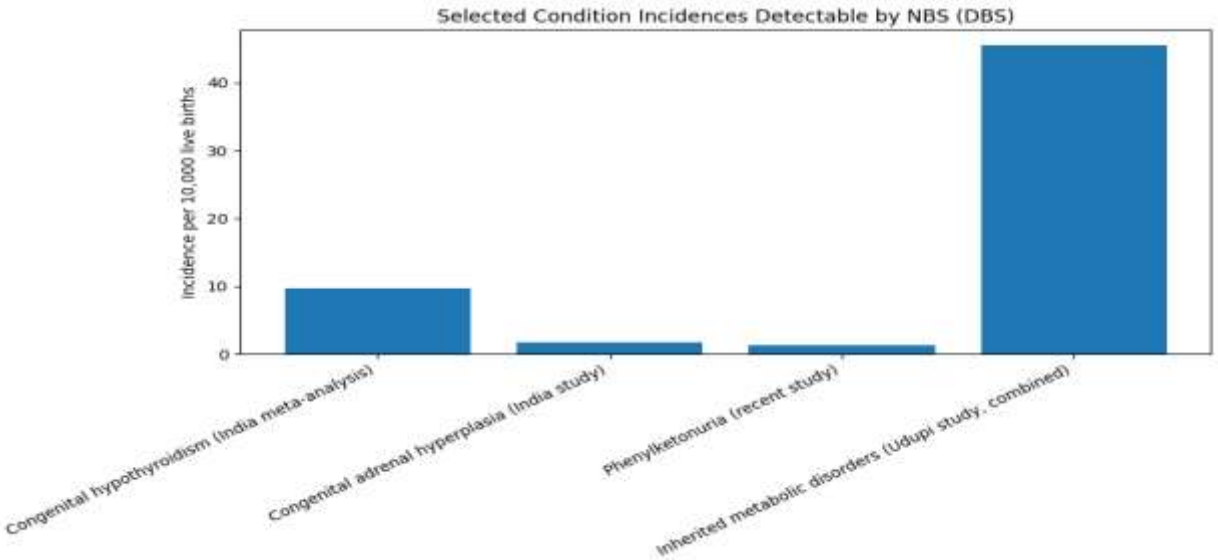

Figure 1. (plotted above) — "Selected Condition Incidences Detectable by NBS (DBS)" shows the per-10,000 incidence for the selected conditions/regions described above (CH, CAH, PKU, combined IEMs). The plot is based on the cited studies and illustrates why DBS-based NBS panels can capture substantial disease burden even when single-disease incidence is low.

Condition (source)	Incidence	Approx. ratio
Congenital hypothyroidism — India meta-analysis.[8]	9.69 per 10,000	1 : 1,031
Congenital adrenal hyperplasia — India cohort. [9]	1.74 per 10,000	1 : 5,762
Phenylketonuria — recent screened cohort.[10]	1.33 per 10,000	1 : 7,519
Combined IEMs — Udupi pilot program (regional).[11]	45.45 per 10,000	1 : 220

Figure 2. Bar chart — Selected Condition Incidences Detectable by NBS (DBS).

Objectives of the Study:

Primary objective:

1. To evaluate the feasibility and screening yield of a mandatory dried blood spot (DBS)-based newborn screening program implemented at the tertiary care centers from north Karnataka.

Secondary Objectives:

2. To determine the frequency and distribution of screen-positive cases for the mandatory minimum newborn screening panel, including:
   - Phenylketonuria (PKU)
   - Glucose-6-phosphate dehydrogenase (G6PD) deficiency
   - Galactosemia
   - Congenital hypothyroidism (TSH estimation)
   - Hemoglobinopathies
3. To assess the association between demographic variables (sex, rural/urban residence) and screen-positive outcomes.
4. To evaluate the association between maternal factors (maternal age, consanguinity, medical history, and socioeconomic status) and abnormal newborn screening results.
5. To analyze the effectiveness of repeat DBS testing and follow-up protocols in distinguishing true-positive cases from transient or marginal biochemical elevations.

MATERIALS AND METHODS

Study design and institutional policy

This study was designed as a prospective, laboratory-based observational newborn screening program, conducted in accordance with national recommendations for newborn screening. As part of an institutional social responsibility initiative, newborn screening (NBS) was made mandatory for all live-born neonates delivered at the study hospital during the study period, irrespective of socioeconomic status or clinical indication. The screening was provided as a minimum essential NBS panel aimed at early detection of common, treatable congenital disorders with proven public-health benefit.

Study setting and period

The study was conducted in the Department of Genetics and Biochemistry in collaboration with Neonatal Intensive Care Unit (NICU) - Department of Pediatrics and Obstetrics and Gynecology(OBG), S. Nijalingappa Medical College & HSK Hospital, Bagalkote, Karnataka, India, over a one-year period from January 2024 to December 2024.

Maternal and Sociodemographic Data Collection

Maternal and sociodemographic information was collected at the time of newborn screening using a structured proforma. Data included maternal age, consanguinity status, relevant maternal medical history, place of residence (urban/rural), and socioeconomic status. Information was obtained from medical records and direct interview with the mother or parent/guardian.

Maternal age was categorized into predefined age groups. Consanguinity was recorded as present or absent based on parental history. Maternal medical history included documented endocrine disorders, metabolic conditions, anemia, pregnancy-induced hypertension, diabetes mellitus, thyroid disorders, and other chronic illnesses. Socioeconomic status was classified according to routinely used institutional or national socioeconomic scales (Table 1).

Table 1. Baseline maternal and sociodemographic characteristics of the study population (n = 1,000)

Parameter	Category	Number (n)	Percentage (%)
Maternal age (years)	< 20	85	8.5
	20–25	350	35
	26–30	480	48
	31–35	80	8.0

		> 35	05	0.5
	Consanguinity	Present	350	35
		Absent	650	65
	Maternal medical history	No significant illness	680	68
		Diabetes mellitus	115	11.5
		Thyroid disorder	95	9.5
		Pregnancy-induced hypertension	65	6.5
		Other chronic illness*	45	4.5
	Place of residence	Rural	600	60.0
		Urban	400	40.0
	Socioeconomic status	Lower	350	35
		Middle	400	40
		Upper	250	25
	Total		**1,000**	**100**

*Other chronic illnesses include anemia, epilepsy, cardiac disease, or other documented medical conditions.

Mandatory minimum NBS panel

In alignment with feasibility considerations and regional disease burden, the hospital implemented a minimum mandatory NBS panel, comprising the following disorders:

- Phenylketonuria (PKU) by HPLC
- Glucose-6-phosphate dehydrogenase (G6PD) deficiency by HPLC
- Galactosemia (total galactose estimation) by HPLC
- Congenital hypothyroidism (thyroid-stimulating hormone, TSH) by HPLC
- Hemoglobinopathies (including sickle cell disease and major thalassemia syndromes) by enzyme immunoassay.

These conditions were selected based on their clinical severity, availability of effective early interventions, feasibility of DBS-based detection, and relevance to the regional population.

DBS sample collection and timing

DBS samples were collected using a heel-prick method following standard aseptic precautions. Capillary blood was spotted onto standardized DBS filter paper cards, ensuring uniform saturation of pre-marked circles without overlapping or layering. Sample collection was preferentially performed between 24 and 72 hours of life, as recommended by national newborn screening guidelines.

Drying, storage, and transport

Following collection, DBS cards were air-dried horizontally at room temperature for 3–4 hours, protected from heat, humidity, and direct sunlight. Dried cards were individually packaged with desiccants and transported to the Genetics and Genomics laboratory under controlled conditions. All samples were logged and tracked using unique identifiers.

Laboratory analysis

DBS samples were processed using validated screening assays for each analyte included in the mandatory panel. Uniform punches were obtained from each DBS card and analyzed according to established laboratory protocols. Internal quality controls and calibrators were included in each batch. Samples showing abnormal or borderline results were subjected to repeat testing or referred for confirmatory evaluation as per standard operating procedures (Figure 3,4)(Table 2).

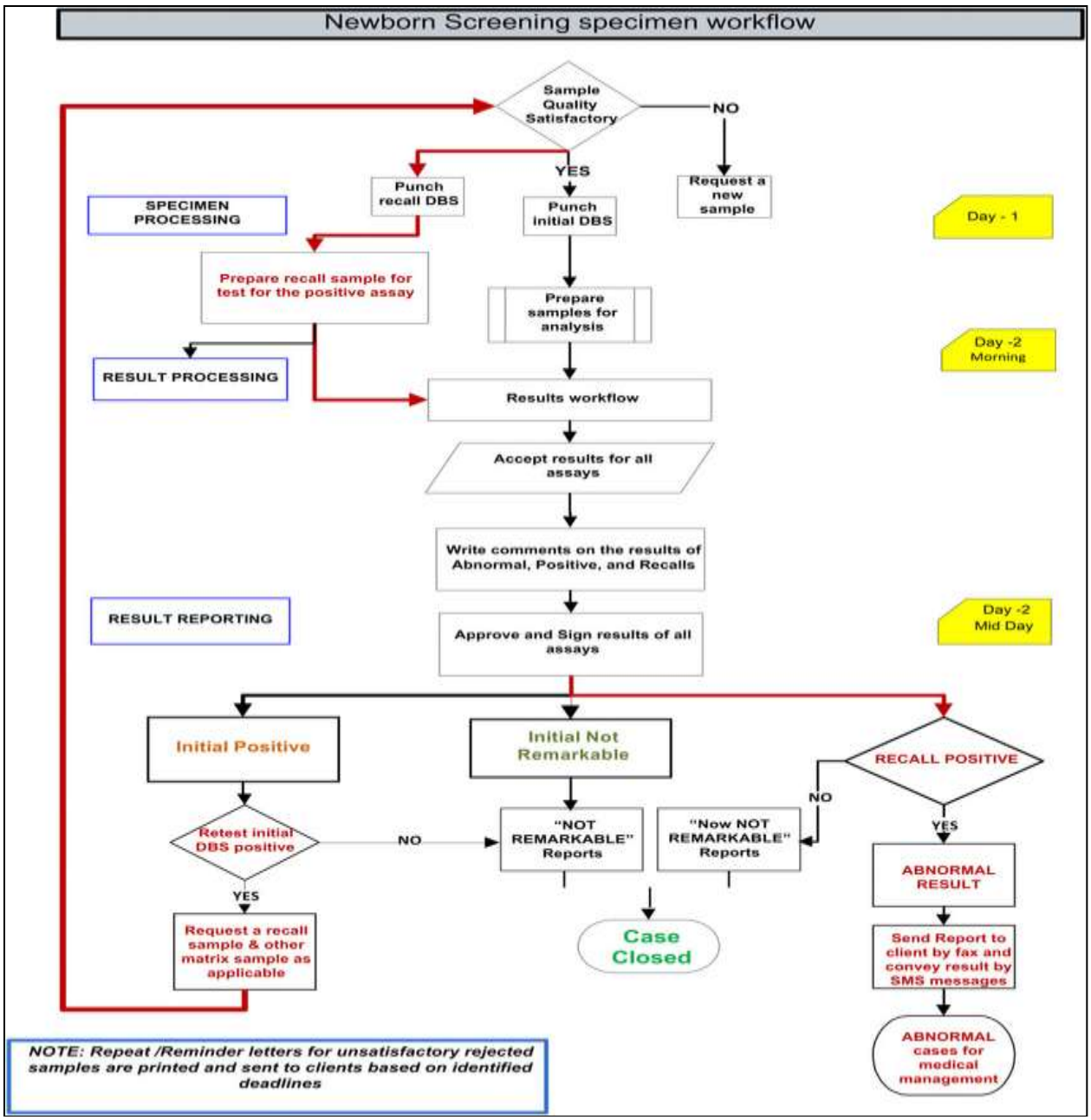

Figure 3. Schematic representation of the DBS-based newborn screening workflow.

Figure 4: Newborn identification and consent, heel-prick blood collection, DBS card drying and storage, transport to screening laboratory, primary screening analysis, repeat/confirmatory testing for abnormal results, and clinical referral and follow-up.

Table 2. DBS Sample Rejection Criteria (for quality assurance)

Sl. No.	Rejection criterion	Reason
1	Insufficient blood volume	Incomplete saturation affects analyte quantification
2	Layered or clotted blood	Causes uneven analyte distribution
3	Contaminated DBS (water, antiseptic, powder)	Analytical interference
4	Improper drying or moisture exposure	Degradation of analytes
5	Heat-damaged or discolored cards	Loss of assay reliability
6	Improper labeling or missing identifiers	Sample traceability compromised
7	DBS collected without documented consent	Ethical non-compliance

Inclusion and exclusion criteria

Inclusion criteria

- Newborns enrolled under the mandatory institutional NBS program
- Adequately collected and labeled DBS samples
- Written informed consent from parent or legal guardian

Exclusion criteria

- Inadequate blood volume or improperly collected DBS samples
- Contaminated, clotted, overheated, or improperly dried cards
- Samples lacking consent documentation

Ethical considerations

The study protocol and Written informed consent was obtained from the parent or legal guardian (patient attendee) of each newborn prior to DBS collection. The mandatory nature of screening was communicated as part of institutional policy, while ensuring confidentiality, ethical compliance, and the right to counseling and follow-up.

Statistical analysis

Screening data were entered into a secure database and analyzed using standard statistical software IBM SPSS. Descriptive statistics were used to summarize screening outcomes, and results were expressed as frequencies, percentages, and incidence rates where applicable.

Statistical note

Categorical variables were expressed as frequencies and percentages. Mean values were calculated per year. Associations between screening outcomes and demographic variables were assessed using the chi-square test. A p-value < 0.05 was considered statistically significant.

Results:

Demographic distribution and screening yield

Among the 1,000 screened newborns, the mean number of screened newborns per year during the study period (Jan 2024–Dec 2024) was 333.3 ± 57.7. Male newborns constituted 560 (56%) and females 440 (44%), while 600 (60%) newborns were from rural areas and 400 (40%) from urban areas.

The overall screen-positive rate for the mandatory NBS panel was 8.7% (87/1,000), corresponding to a mean screen-positive yield of 2.9 ± 1.4 cases per disorder per year.

Association of Maternal Factors with Screen-Positive Outcomes

The association between maternal and sociodemographic variables and newborn screening outcome (screen-positive vs screen-negative) was evaluated using the Chi-square test of independence. A p-value < 0.05 was considered statistically significant.

Table 3. Association of maternal and sociodemographic factors with screen-positive outcomes (n = 1,000)

Variable	Category	Screen-positive n (%)	Screen-negative n (%)	χ^2 (df)	p-value
Maternal age (years)	< 20	10 (11.8)	75 (88.2)	4.12 (4)	0.39
	20–25	30 (8.6)	320 (91.4)		
	26–30	38 (7.9)	442 (92.1)		

	31–35	7 (8.8)	73 (91.2)		
	> 35	2 (40.0)	3 (60.0)		
Consanguinity	Present	42 (12.0)	308 (88.0)	9.62 (1)	**0.002**
	Absent	45 (6.9)	605 (93.1)		
Maternal medical history	No significant illness	50 (7.4)	630 (92.6)	8.01 (4)	0.09
	Diabetes mellitus	14 (12.2)	101 (87.8)		
	Thyroid disorder	11 (11.6)	84 (88.4)		
	PIH	8 (12.3)	57 (87.7)		
	Other chronic illness	4 (8.9)	41 (91.1)		
Place of residence	Rural	55 (9.2)	545 (90.8)	0.83 (1)	0.36
	Urban	32 (8.0)	368 (92.0)		
Socioeconomic status	Lower	38 (10.9)	312 (89.1)	3.21 (2)	0.20
	Middle	32 (8.0)	368 (92.0)		
	Upper	17 (6.8)	233 (93.2)		

Interpretation of statistical findings

- Consanguinity showed a statistically significant association with screen-positive outcomes ($\chi^2 = 9.62$, df = 1, $p = 0.002$), indicating a higher proportion of abnormal screening results among newborns born to consanguineous parents.
- Maternal age, maternal medical history, place of residence, and socioeconomic status did not show statistically significant associations with screen-positive outcomes ($p > 0.05$).
- These findings suggest that while demographic and socioeconomic variables did not significantly influence screening positivity, parental consanguinity remains an important risk factor detectable through universal newborn screening (Table 3).

Association of screening positivity with sex and residence

- Chi-square test was applied to assess the association between screen-positive status and sex as well as place of residence. No statistically significant association was observed (Table 4).

Table 4. Association of screen-positive results with demographic variables

Variable	Screen-positive (n)	Screen-negative (n)	p-value
Gender of baby			
• Male (n = 560)	50	510	0.41
• Female (n = 440)	37	403	
Residence			
• Rural (n = 600)	55	545	0.36
• Urban (n = 400)	32	368	

p-value calculated using chi-square test; $p < 0.05$ considered statistically significant.

Disorder-wise screening results with mean values

Table 5. Disorder-wise screening outcomes with mean distribution (n = 1,000)

Disorder	Screen-positive cases (n)	Percentage (%)	Mean per 1,000 ± SD
Phenylketonuria (PKU)	8	0.8	2.7 ± 0.6
G6PD deficiency	22	2.2	7.3 ± 1.5
Galactosemia	12	1.2	4.0 ± 1.0
Congenital hypothyroidism (TSH)	35	3.5	11.7 ± 2.1
Hemoglobinopathies	10	1.0	3.3 ± 0.8
Total	87	8.7	29.0 ± 4.6

Mean values represent average number of screen-positive cases per year across the study period.

Follow-up and outcome classification

All high-risk screen-positive cases underwent repeat DBS testing and family history assessment. Newborns with persistent elevation on repeat testing were referred for confirmatory diagnostic evaluation. Marginally elevated cases were advised repeat biochemical evaluation after 4 months, as transient neonatal physiological variations could not be excluded.

DISCUSSION

Newborn screening (NBS) using dried blood spot (DBS) methodology has emerged as a cornerstone of preventive neonatology, enabling early identification of treatable congenital disorders before clinical manifestation. The present study demonstrates the feasibility and effectiveness of implementing a mandatory, institution-based NBS program in a resource-limited setting, with a substantial overall screen-positive yield of 8.7%.

The observed screening yield in this study is comparatively higher than many international reports, which may be attributed to regional genetic heterogeneity, higher prevalence of metabolic and endocrine disorders, and inclusion of a targeted minimum panel relevant to the local population. Similar findings have been reported in Indian pilot studies, where combined incidence of inherited metabolic disorders (IEMs) was significantly higher than global averages, reinforcing the importance of region-specific screening strategies [12].

Among the screened disorders, congenital hypothyroidism (CH) was the most frequently detected condition (3.5%). This aligns with existing Indian data suggesting a relatively higher prevalence of CH compared to Western populations, possibly due to iodine status variations and genetic factors. Early detection of CH is critical, as timely thyroxine therapy prevents irreversible intellectual disability and growth failure [8,9].

G6PD deficiency (2.2%) was the second most common abnormality detected. Given its association with neonatal jaundice and risk of hemolytic crises, especially in populations with high prevalence, inclusion of G6PD screening in the mandatory panel is justified. Similar prevalence trends have been documented in South Asian populations [5].

The detection rates for phenylketonuria (PKU) (0.8%) and galactosemia (1.2%) highlight the clinical value of metabolic screening even in regions where these conditions are traditionally considered less common. Early dietary and medical interventions in these disorders significantly reduce morbidity and improve neurodevelopmental outcomes [4].

A key finding of this study is the statistically significant association between consanguinity and screen-positive outcomes ($p = 0.002$). This observation is consistent with well-established genetic principles, as consanguineous unions increase the likelihood of autosomal recessive disorders. Similar associations have been widely reported in Indian and Middle Eastern populations, emphasizing the need for genetic counseling and targeted screening in such high-risk groups [12].

In contrast, maternal age, socioeconomic status, residence, and maternal medical history did not show statistically significant associations with screening positivity. These findings suggest that universal screening remains essential, as reliance solely on risk-based screening may miss a significant proportion of affected newborns.

The study also highlights the operational robustness of DBS methodology. The ability to collect, transport, and analyze samples efficiently under routine clinical conditions demonstrates its suitability for large-scale implementation. DBS-based screening offers advantages including minimal invasiveness, stability during transport, and compatibility with multiple analytical platforms such as HPLC and immunoassays [8,10].

An important programmatic strength observed was the implementation of a repeat testing and follow-up protocol, which helped differentiate true-positive cases from transient biochemical abnormalities. This approach reduced unnecessary referrals and parental anxiety, thereby improving the overall efficiency of the screening program.

When compared to global NBS programs, which increasingly incorporate expanded panels including molecular screening (e.g., SCID, SMA), the current study supports a stepwise expansion model. Starting with a feasible minimum panel tailored to regional disease burden is a pragmatic approach for developing countries, with gradual scaling based on infrastructure and epidemiological data [9].

Despite its strengths, the study has certain limitations. Being a single-center study, the findings may not be fully generalizable to other regions. Additionally, confirmatory diagnostic outcomes and long-term follow-up data were not extensively analyzed, which are essential for assessing the true clinical impact of screening programs.

CONCLUSION

This study reinforces that mandatory DBS-based newborn screening is feasible, effective, and highly relevant in the Indian context, particularly in regions with high consanguinity and genetic disease burden. Expansion of such programs at regional and national levels can significantly reduce preventable morbidity and mortality, thereby contributing to a reduction in neonatal and infant mortality rates.

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The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

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