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ORIGINAL RESEARCH

PREVALENCE OF CONGENITAL HEART DISEASE AMONG INFANTS REPORTING IN TERTIARY CARE HOSPITAL

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ABSTRACT

Congenital heart disease (CHD) represents the most prevalent group of structural birth defects and is a leading cause of infant morbidity and cardiac mortality worldwide. Despite its substantial public health significance, population-specific prevalence data for the Punjab region of northern India are lacking. Such data are essential for planning appropriate cardiac care resources and informing regional health policy. A cross-sectional descriptive study was conducted over 18 months in the Department of Pediatrics, Rajindra Hospital and Government Medical College, Patiala. Infants aged 0-12 months comprising both consecutive live newborns delivered in the hospital and those attending outpatient or inpatient services were screened. Infants with clinical features suggestive of CHD were evaluated by comprehensive echocardiography (two-dimensional, M-mode, colour flow Doppler, and spectral Doppler) using a Sonosite Micromaxx device with an 8 MHz probe. Echocardiograms were interpreted by a trained consultant cardiologist in accordance with the American Society of Echocardiography guidelines. Of 14,236 infants screened, CHD was confirmed echocardiographically in 66, yielding a prevalence of 4.6 per 1,000 live births. CHD was more prevalent among preterm (n = 52; 78.8%) and male (n = 36; 54.5%) infants. The most common clinical presentations were pallor (36.4%), cyanosis (34.9%), and hepatomegaly (34.9%). Atrial septal defect (ASD) was the most frequent lesion (62.1%), followed by patent ductus arteriosus (PDA; 18.2%), combined ASD and PDA (7.6%), and ventricular septal defect (VSD; 4.5%). CHD was an incidental echocardiographic finding in 39.4% of cases. No statistically significant association was found between demographic variables and the type of CHD. The prevalence of CHD in this infant cohort from Punjab was 4.6 per 1,000 live births. ASD was the predominant lesion, consistent with studies predominantly recruiting infants. These findings underscore the need for systematic echocardiographic screening and strengthening of regional paediatric cardiac care infrastructure.

Keywords: Congenital heart disease; Neonates; Echocardiography; Atrial septal defect; Punjab

INTRODUCTION

Congenital heart disease (CHD) encompasses a broad spectrum of structural cardiac anomalies present at birth, including defects of the cardiac chambers, valves, septae, and the great intrathoracic vessels. These malformations arise during fetal cardiac morphogenesis and disrupt normal haemodynamics to varying degrees, ranging from clinically silent lesions to life-threatening haemodynamic compromise [1]. CHD accounts for approximately 28% of all major congenital anomalies, and when considered alongside neural tube defects collectively constitutes nearly two-thirds of all significant birth defects [2]. Its aetiology is multifactorial, involving interactions between genetic susceptibility and maternal environmental exposures during the first trimester of pregnancy, including gestational diabetes, febrile illness, exposure to teratogens, and autoimmune disorders [3].

CHD is the leading cause of birth defect-related morbidity and infant mortality across both high- and low-income countries. Globally, an estimated 1.35 million infants are born with CHD each year, with a birth prevalence of approximately 9 per 1,000 live births [4]. In India, epidemiological estimates have varied considerably across published studies, with reported prevalence figures ranging from 0.8 to 26.4 per 1,000 individuals (pooled mean: 7.10; 95% CI:

2.94–11.27 per 1,000) [4]. This variability reflects genuine differences in study methodology, echocardiographic access, age distribution of study populations, and healthcare settings. In terms of defect distribution, VSD and ASD are the most commonly reported anomalies across Indian studies, followed by tetralogy of Fallot (TOF), PDA, and transposition of the great arteries (TGA) [4].

India carries a disproportionately large share of the global CHD burden, with an estimated 200,000 or more infants born with CHD each year and access to corrective surgery limited to a minority of those who require it [5]. While several hospital-based and community-based studies have been published from different Indian states including Uttar Pradesh, Rajasthan, Maharashtra, and Karnataka comprehensive epidemiological data from the Punjab region of northern India remain notably absent from the literature. Regional data are essential for accurate resource planning, training of specialist pediatric cardiologists, and directing health policy investments toward improving diagnostic and surgical capacity in under-served areas.

Echocardiography has supplanted chest radiography and clinical auscultation as the diagnostic standard for CHD, enabling precise structural characterisation, haemodynamic quantification, and lesion severity grading with high sensitivity and specificity in the hands of trained operators [1]. The present study was therefore conducted to determine the echocardiographically confirmed prevalence and clinical pattern of CHD among infants presenting to a tertiary care hospital in Patiala, Punjab, and to contribute regional data that may inform cardiac care planning in this part of northern India.

MATERIALS AND METHODS

Study Design, Setting, and Duration: This was a cross-sectional descriptive study conducted over 18 months in the Department of Pediatrics, Rajindra Hospital and Government Medical College, Patiala, Punjab. The hospital provides tertiary-level healthcare services to infants and children from across Punjab and Haryana, receiving both locally delivered (inborn) and referred (outborn) patients.

Ethical Approval and Informed Consent: Institutional ethics committee approval was obtained prior to commencement of the study. Written informed consent was sought from the parents or legal guardians of all potential participants prior to enrolment. A detailed participant information sheet, outlining the study objectives, procedures, potential benefits, and the voluntary nature of participation, was provided in the local language.

Study Population: The study population comprised infants aged 0–12 months in two groups: (i) consecutive live newborns delivered in Rajindra Hospital and admitted to the Department of Pediatrics; and (ii) infants attending the outpatient department (OPD) or inpatient wards for other clinical complaints or post-discharge follow-up, who were clinically suspected to have CHD.

Inclusion criteria: All infants aged 0–12 months admitted to the pediatric ward or attending the OPD at Rajindra Hospital/Government Medical College, Patiala, irrespective of mode of delivery, gestational age, birth weight, or maternal health status, who demonstrated clinical features warranting echocardiographic evaluation.

Exclusion criteria: (i) critically ill or intubated infants who could not be safely transported to the echocardiography suite and for whom bedside echocardiography was unavailable; (ii) infants experiencing convulsions at the time of planned echocardiography; and (iii) infants whose parents or guardians declined to provide informed consent.

Clinical Screening: All enrolled infants underwent structured clinical assessment by a pediatrician. Infants were considered to have suspected CHD if they presented with one or more of the following: central cyanosis, a clinically significant cardiac murmur, respiratory distress unexplained by a primary respiratory aetiology, failure to thrive, signs of congestive cardiac failure, or other clinical features suggestive of a structural cardiac lesion. Demographic data, birth history, maternal history, gestational age, birth weight, mode of delivery, socioeconomic status, family history of CHD, presenting complaints, and clinical examination findings were recorded on a pretested, predesigned data collection proforma.

Echocardiographic Assessment: Each enrolled infant was examined by echocardiography after feeds, in a calm state (lying quietly or asleep), to minimise artefact and optimise image quality. All echocardiograms were performed using a Sonosite Micromaxx device with an 8 MHz probe. Standard echocardiographic modalities two-dimensional imaging, M-mode, colour flow Doppler, and spectral Doppler were applied, interrogating cardiac structures from apical, subcostal, parasternal, and suprasternal views. All echocardiograms were performed and reported by a consultant cardiologist with appropriate training and experience in pediatric and congenital echocardiography. Findings were classified in accordance with the criteria of the American Society of Echocardiography [1].

Statistical Analysis: Data analysis: Analysis of the report was done according the American Society of Echocardiography. All collected data was coded and analysed using IBM SPSS version 20.0. The prevalence of CHD was expressed as number per 1000 infants. The difference in prevalence between types of CHD was tested with the Z test. The difference in mean values such as ages of infants with different types of CHD was compared using the student's t-test or one way ANOVA. The level of significance was set at $p<0.05$.

RESULTS

Screening Outcomes and Prevalence

A total of 14,236 infants were assessed during the study period. Of these, 14,018 did not meet the inclusion criteria and were excluded, and 152 parents declined consent. Sixty-six infants were confirmed to have CHD on echocardiography, yielding a hospital-based prevalence of 4.6 per 1,000 live births (0.46%). Two critically ill infants presenting with severe cyanosis and oxygen desaturation were intubated and required emergency transfer to a higher-level cardiac care facility; they could not be evaluated on-site due to the absence of bedside echocardiography and are not included in the echocardiographic characterisation. The participant flow is illustrated in Figure 1.

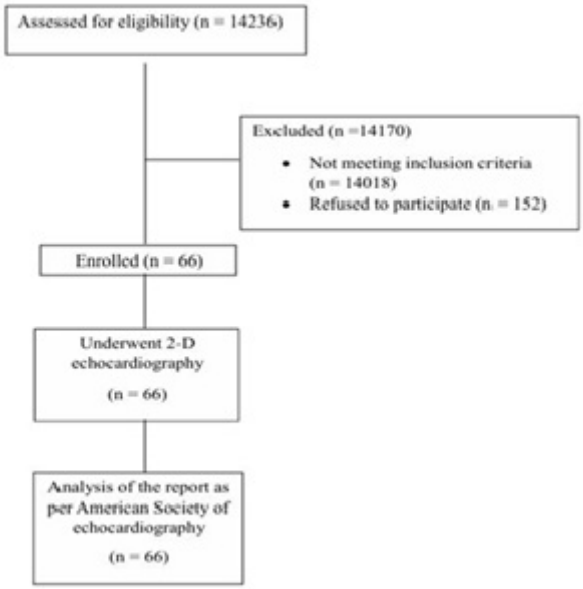

Figure 1. *Consort diagram showing the flow of participants through each stage of study*

Demographic and Perinatal Characteristics:

Of the 66 infants with confirmed CHD, 31 were inborn and 35 were outborn. The sample comprised 36 males (54.5%) and 30 females (45.5%). Fifty-two infants (78.8%) were preterm and 14 (21.2%) were born at term. The majority of infants (n = 38; 57.6%) had a birth weight between 1,500 and 2,499 g; the mean birth weight was 2,014 ± 362 g. Combining birth weight and gestational age, 46 infants (69.7%) were appropriate for gestational age (AGA), 20 (30.3%) were small for gestational age (SGA), and none were large for gestational age (LGA). Socioeconomic status was classified as lower-

middle in 42 infants (63.6%). Maternal age at delivery was 26–30 years in 41 cases (62.1%), with a mean maternal age of 27.8 ± 2.73 years. Forty-six infants (69.7%) were delivered by lower segment Caesarean section (LSCS) and 20 (30.3%) by normal vaginal delivery (NVD). A family history of CHD was documented in 17 infants (25.8%). The mean peripheral oxygen saturation (SpO_2) at presentation was 94.98 ± 2.34%. These characteristics are summarised in Table 1.

Table 1. Distribution of Infants with Confirmed CHD by Perinatal and Demographic Variables (n = 66)

Variable		Number	Inborn (n=31)	Out born (35)
Gestation	Preterm	52	21	31
	Term	14	10	4
Sex	Male	36	15	21
	Female	30	16	14
Birth Weight	1000-1499g	7	3	4
	1500-2499g	38	17	21
		21	11	10
Birth weight & Gestation	AGA	46	19	27
	SGA	20	12	8
	LGA	-	-	-
SES	Lower Middle	42	21	21
	Upper Middle	16	9	7
	Upper Lower	5	1	4
	Lower	3	0	3
Maternal Age	21-25yrs	13		
	26-30yrs	41		
	>30yrs	12		

Note: AGA, Appropriate for Gestational Age; SGA, Small for Gestational Age; LGA, Large for Gestational Age; GA, Gestational Age.

Clinical Presentation

The most frequently recorded clinical signs on examination were pallor (n = 24; 36.4%), cyanosis (n = 23; 34.9%), and hepatomegaly (n = 23; 34.9%), followed by oedema (n = 20; 30.3%), retinopathy and dilated neck veins (both n = 19; 28.8%), facial dysmorphism (n = 14; 21.2%), and skeletal abnormalities (n = 8; 12.1%), as shown in Table 2. The majority of infants (n = 32; 48.5%) presented within the first three months of life. Inborn infants were significantly more likely to present in the 0-3 months age group, while outborn infants predominated in older age categories ($\chi^2 = 56.45$, $p < 0.001$), reflecting the pattern of referral-driven detection in community-based cases (Table 3).

Table 2. Distribution of Infants by Clinical Signs at Presentation (n = 66)

Clinical Sign	N	Percentage
Pallor	24	36.36
Cyanosis	23	34.85
Hepatomegaly	23	34.85
Oedema	20	30.30
Retinopathy	19	28.29
Dilated Veins in Neck	19	28.29
Facial Dysmorphism	14	21.21
Skeletal Abnormalities	8	12.12

Table 3. Distribution of Infants by Age at Presentation and Place of Birth

Age Of Presentation	Number of Infants	Inborn	Outborn
0-3 months	32	30	2
3-6 months	21	1	20
7-10 months	10	0	10
>10 months	3	0	3
Total	66	31	35
Chi-square value	56.45		
P value	< 0.0001		

Clinical Impression at Presentation

The leading clinical impressions at the time of first presentation are summarised in Table 4. The most common reason for echocardiographic referral was an incidental or asymptomatic finding (n = 26; 39.4%), typically following the detection of a cardiac murmur. Respiratory distress and shock were each recorded in 22 infants (33.3%). Failure to thrive was the presenting impression in 15 infants (22.7%), and poor feeding in 10 (15.1%). Congestive cardiac failure was the primary clinical concern in 3 infants (4.5%).

Table 4. Distribution of Infants by Clinical Impression at Presentation (n = 66)

Clinical Impression	Number of Infants (%)	Inborn Number (%)	Outborn Number (%)
Incidental (Asymptomatic)	26(39.3)	12	14
Respiratory Distress	22 (33.3)	9	13
Shock	22 (33.3)	11	11
Failure to Thrive	15 (22.7)	4	11
Poor Feeding	10 (15.1)	6	4
Congestive Cardiac Failure	3 (4.5)	1	2
Total	66	31	35

Note: Percentages do not sum to 100% as infants may have presented with more than one clinical feature.

Echocardiographic Findings:

ASD was the most frequently identified lesion, confirmed in 41 infants (62.1%), followed by PDA in 12 (18.2%), combined ASD and PDA in 5 (7.6%), VSD in 3 (4.5%), and patent foramen ovale (PFO) in 2 (3.0%). The distribution of echocardiographic diagnoses is presented in Figure 2.

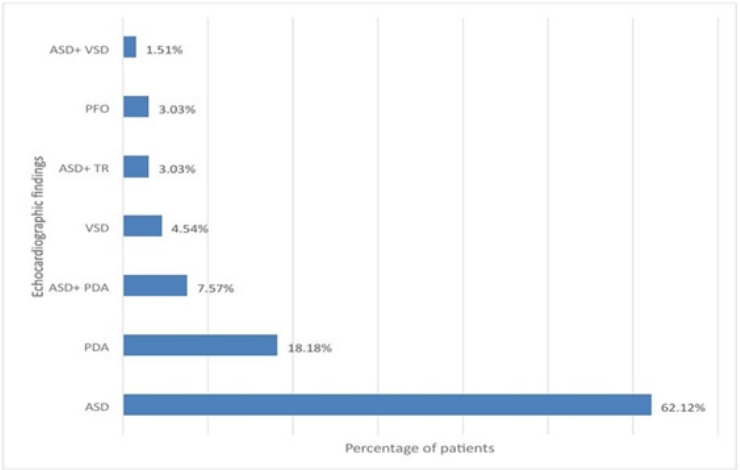

Figure 2. *Distribution of infants by echocardiographic findings*

Association between Demographic Variables and Type of CHD

The association between demographic and perinatal variables including sex, gestational age, mode of delivery, birth weight-for-gestational-age classification, and clinical signs and the echocardiographic type of CHD (ASD, PDA, VSD, PFO) was examined using chi-square analysis. No statistically significant association was found between any of the variables examined and the specific type of CHD (all $p > 0.05$), as detailed in Table 5.

Table 5. Association between Demographic and Perinatal Variables and Type of CHD (n = 66)

Variable		ASD (41)	PDA (12)	VSD (3)	PFO (2)	X2	Pvalue
Gender	Male	24	6	1	0	3.29	0.35
	Female	17	6	2	2		
Gestation	Term	8	1	2	1	6.08	0.11
	Preterm	33	11	1	1		
Mode of Delivery	LSCS	14	0	1	1	6.96	0.07
	NVD	27	12	2	1		
Gestation &Birth weight	AGA	31	6	1	2	5.00	0.17
	SGA	10	6	2	0		
Clinical Sign							
Pallor		14	4	2	0	2.44	0.49
Cyanosis		16	5	1	1		
Edema		14	5	0	0		
Hepatomegaly		17	3	1	1		
Dilated veins		12	4	1	1		
Dysmorphism		8	3	1	0		
Retinopathy		14	4	0	0		
Skeletal Abnormality		6	1	0	0		
Birth Place	Inborn	18	6	2	1		
	Out-born	23	6	1	1		

Note: ASD, Atrial Septal Defect; PDA, Patent Ductus Arteriosus; VSD, Ventricular Septal Defect; PFO, Patent Foramen Ovale; LSCS, Lower Segment Caesarean Section; NVD, Normal Vaginal Delivery; AGA, Appropriate for Gestational Age; SGA, Small for Gestational Age. χ^2, Pearson chi-square statistic. All p-values > 0.05 (not statistically significant).

DISCUSSION

The present study documented a CHD prevalence of 4.6 per 1,000 live births among infants screened at a tertiary care hospital in Patiala, Punjab the first study to report population-specific echocardiographic CHD prevalence data from this region of northern India. This figure is consistent with rates reported in comparable hospital-based studies in South Asia. Mazhani et al. reported a prevalence of 2.8–4.95 per 1,000 live births in a pediatric echocardiography series from Botswana [6], and Rizvi et al. documented a prevalence of 3.4 per 1,000 in rural communities in Pakistan [7]. The present estimate is notably lower than the 27.7 per 1,000 reported by Jain et al. from Central India [8] and the 19.14 per 1,000 reported by Bhardwaj et al., [9] differences that are largely attributable to the broader age range of study populations in those reports (0–15 years and 0–5 years, respectively), the larger cumulative sample sizes, and the varying thresholds for echocardiographic referral. Conversely, Bhosgi et al. reported a substantially lower prevalence of 1.12 per 1,000 [10] likely reflecting differences in healthcare access and the proportion of infants with clinically detectable CHD in the catchment population.

The predominance of preterm infants (78.8%) in this CHD cohort is consistent with the established association between prematurity and structural cardiac anomalies particularly PDA, which is both a physiological correlate of preterm birth and

a lesion independently associated with haemodynamic compromise in this population [11,12]. Prematurity is associated with delayed closure of the ductus arteriosus and, in some instances, with broader disruptions of cardiac morphogenesis. These mechanisms likely explain the high representation of preterm infants in studies of CHD conducted in neonatal and infant populations.

A mild male preponderance was observed in this cohort (54.5% male), a pattern documented in several published series on CHD in South Asian populations [12-14]. While some structural cardiac lesions such as TOF and transposition of the great arteries are genuinely more prevalent in males, it is important to acknowledge that healthcare-seeking behaviour in the Indian sociocultural context may introduce a systematic ascertainment bias, with male infants more likely to be brought for medical evaluation by parents.

The mean birth weight in this cohort (2,014 ± 362 g) was lower than reported by Dey et al. (mean 2,376 ± 783.85 g) [15], who studied a largely term NICU population, and was markedly lower than the ≥ 2,500 g mean reported by Khasawneh et al. [16]. The lower birth weight in the present series reflects the high proportion of preterm infants enrolled. When birth weight was combined with gestational age, AGA infants predominated (69.7%), as in the study by Dey et al. (80.7%), while Khasawneh et al. found a higher proportion of SGA infants (43%) [15, 16]. The concentration of SGA infants in the inborn subgroup in the present study likely reflects the more complex perinatal presentations managed at this tertiary centre.

The socioeconomic distribution of CHD cases, with 63.6% of infants from lower-middle-income households, mirrors findings from Meshram et al., [17] who similarly observed the majority of CHD patients from middle and lower socioeconomic groups. This pattern may reflect both the higher birth rates and greater exposure to maternal health risk factors including suboptimal antenatal care, nutritional deficiencies, and uncontrolled gestational conditions in lower-income populations, as well as the referral patterns of a government tertiary centre that predominantly serves lower-income communities.

The mean maternal age of 27.8 ± 2.73 years is consistent with national demographic trends for primiparous mothers in urban northern India, and is lower than the mean of 30.5 ± 7.7 years reported by Zaidi et al [18] in a Pakistani NICU cohort a difference attributable to regional demographic variation. The family history of CHD was present in 25.8% of infants in the present study, a figure intermediate between the 46% reported by Zaidi et al [18], and the 9.2% reported by Al-Fahham et al.,[12] reflecting the known regional and genetic variability in CHD inheritance and the degree of consanguinity within different populations.

Pallor, cyanosis, and hepatomegaly were the predominant clinical signs at presentation, each recorded in approximately one-third of infants. This pattern differs from Chimoriya et al., [13] who reported cyanosis in only 3.6% and hepatomegaly in 2.4% of cases in a broader pediatric echocardiography series difference attributable to the broader sample composition in that study. Meshram et al. [17] reported cyanosis in 40% and hepatomegaly in 71.9% of cases, a higher prevalence reflecting their inclusion of older children with more advanced disease. Notably, cyanosis in the present cohort resolved with conservative management in the majority of affected infants, suggesting that many cases represented functional or transitional cyanosis rather than fixed structural desaturation.

The predominance of presentations in the 0–3-months age group (48.5%) is consistent with other published series in which CHD typically comes to clinical attention within the first trimester of life [8,17], either through neonatal clinical screening, echocardiographic surveillance in preterm infants, or parental presentation following the recognition of feeding difficulties, failure to gain weight, or respiratory symptoms. The statistically significant difference in age at presentation between inborn and outborn infants ($\chi^2 = 56.45$, $p < 0.001$) reflects the earlier detection facilitated by in-hospital monitoring of newborns in the tertiary care setting, compared with the delayed referral patterns typical of community-detected cases.

The high proportion of incidental echocardiographic diagnoses (39.4%) most commonly following the detection of a cardiac murmur on routine examination is consistent with data from Khasawneh et al., [16] who found that the majority of echocardiographic referrals in their infant cohort were triggered by the incidental finding of a murmur, and with Al-

Fahham et al.,[12] who reported an incidental echocardiographic finding in 35% of CHD cases. This highlights the continuing diagnostic value of careful clinical auscultation in the newborn examination and the importance of systematic follow-up echocardiography in infants with murmurs, even in the absence of overt symptoms. Failure to thrive, documented in 22.7% of infants in the present study, is consistent with the 27% reported by Gupta et al [14].

ASD was the predominant echocardiographic diagnosis in this cohort (62.1%), followed by PDA (18.2%) and combined ASD and PDA (7.6%). This pattern differs from studies that include a broader age spectrum, where VSD consistently emerges as the most common CHD lesion [10,14,19,20]. The dominance of ASD in infant-specific cohorts has been documented in comparable series: Rizvi et al. found ASD as the most frequent lesion (40%) in their population-based study [7], and Sun et al. identified ASD as the single most common defect in a large infant screening programme in China [21]. This age-specific pattern is explained by the natural history of these lesions: many small VSDs close spontaneously within the first year of life and may therefore be underrepresented in infant cohorts, while haemodynamically significant ASDs are more likely to be clinically detected and to persist. The higher PDA prevalence in studies including predominantly older children with diverse diagnoses [18] likely reflects the higher representation of ex-preterm infants with persistent ductal patency in those settings.

The absence of statistically significant associations between demographic and perinatal variables (sex, gestational age, mode of delivery, birth weight classification) and the specific type of CHD ($p > 0.05$ for all comparisons) is consistent with findings from comparable studies [13,16] and reflects the multifactorial aetiology of CHD, in which no single perinatal variable reliably predicts the type of structural lesion.

CONCLUSION

This study documents a CHD prevalence of 4.6 per 1,000 live births among infants evaluated at a tertiary care hospital in Patiala, Punjab representing the first echocardiographically confirmed CHD prevalence data from this region of northern India. CHD was more frequent in preterm and male infants, and the majority of cases were diagnosed within the first three months of life. ASD was the dominant lesion in this infant-specific cohort, consistent with the age-dependent lesion distribution documented in comparable literature. Nearly 40% of cases were identified incidentally, underscoring the diagnostic importance of systematic newborn cardiac auscultation. These findings highlight the need for accessible echocardiographic screening, including bedside echocardiography for critically ill neonates, and advocate for strengthening of pediatric cardiac care services in Punjab to reduce the diagnostic and therapeutic gap that currently limits outcomes for infants with CHD in this region.

Limitations

Several methodological limitations should be acknowledged. As a single-centre, hospital-based study, the findings may not be representative of the broader Punjab population, and referral bias may have resulted in selective inclusion of clinically detectable or more severe cases. Bedside echocardiography was not available, leading to the exclusion of two critically ill infants and potentially other infants whose clinical condition precluded transport. The study did not include a systematic genetic or teratogenic risk factor assessment, and longitudinal outcome data were not collected.

Contributors: MS conceptualised the study design and wrote the manuscript. AK collected the data and prepared the initial draft. BK supervised data collection and performed critical analysis. All authors reviewed and approved the final version of the manuscript.

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