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SPECTRUM OF CONGENITAL HEART DISEASE AMONG PEDIATRIC PATIENTS WITH CONGENITAL SURGICAL ANOMALIES: A RETROSPECTIVE STUDY

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Abstract

Background

Congenital heart disease (CHD) is the most prevalent congenital disorder and is associated with extracardiac malformations arising during early organogenesis. Children with congenital surgical systemic anomalies (CSSA) are a high-risk group in whom cardiac defects may be clinically silent yet influence perioperative outcomes. Data on the prevalence and spectrum of CHD across CSSA remain limited

Methods

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A retrospective cross-sectional study was conducted from June 2023 to June 2025 in a tertiary care pediatric teaching hospital. Children aged 0–5 years with CSSA who underwent routine echocardiography were included. Surgical anomalies were categorized into gastrointestinal, urogenital, musculoskeletal, craniofacial, and other systemic groups. Demographic data and echocardiographic findings were analyzed to determine CHD prevalence and patterns.

Results

Of 51 children, 24 (47.1%) had structural CHD. Structural CHD was present in all children with urogenital (n=4) and musculoskeletal anomalies (n=6), though subgroup sizes were small. Gastrointestinal anomalies contributed the highest number of CHD cases (n=18, 47.4%), while no structural CHD was observed in isolated craniofacial anomalies.

Atrial septal defect was the most common lesion (n=33, 64.7%), followed by ventricular septal defect (n=12, 23.5%) and patent ductus arteriosus (n=8, 15.7%). Valvular pulmonary stenosis was seen in 3 children (5.9%). Pulmonary arterial hypertension and patent foramen ovale were noted in 25 (49.0%) and 14 (27.5%) children, respectively, and were excluded from CHD prevalence calculations.

Conclusion

Structural CHD was present in nearly half of children with congenital surgical systemic anomalies, with a predominance of septal defects. Given the high prevalence and frequent absence of overt cardiac symptoms, routine preoperative echocardiographic screening is essential to ensure optimal perioperative management in this high-risk population.

Keywords: congenital heart disease, congenital anomalies, pediatric surgery, gastrointestinal malformations, echocardiography,

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INTRODUCTION

Congenital Heart Disease (CHD), defined as a structural or functional cardiac abnormality arising during intrauterine development, affects approximately 8–12 per 1,000 live births globally and remains the leading congenital disorder in pediatric populations.[1,2,3] Although CHD may occur in isolation, its frequent coexistence with extracardiac congenital malformations highlights the concept of "developmental field defects," wherein disruptions during organogenesis affect multiple organ systems simultaneously.[4]

Congenital Surgical Systemic Anomalies (CSSA)—including anorectal malformations (ARM), esophageal atresia/tracheoesophageal fistula (EA/TEF), congenital diaphragmatic hernia (CDH), abdominal wall defects, and select urogenital and musculoskeletal anomalies—represent major neonatal surgical conditions. Many of these anomalies exhibit significantly increased rates of associated CHD, with reported prevalence ranging from 16.5% to 65% depending on the anomaly group.[5,6,7,8]

This association is rooted in embryological overlap between cardiac and extracardiac development, particularly during weeks 4–10 of gestation, when teratogenic, genetic, or environmental insults may simultaneously affect multiple organ systems.[9] Genetic evidence reinforces this link, as several chromosomal or microdeletion syndromes—such as 22q11.2 deletion and trisomy 21—present with both cardiac and extracardiac anomalies.[10,11]

Early recognition of CHD is crucial for preoperative optimization because cardiac lesions substantially increase anesthesia-related risk and perioperative morbidity in surgical neonates.[12,13] However, selective screening may miss clinically silent lesions. This study evaluates the prevalence and spectrum of CHD in CSSA patients and reinforces the importance of universal echocardiographic screening.

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MATERIALS AND METHODS

Study Design and Setting

This study was designed as a retrospective cross-sectional observational study conducted in the Department of Pediatrics at Bapuji Child Health Institute & Research Centre, J.J.M. Medical College, Davangere, Karnataka. The hospital is a tertiary care pediatric teaching institution with dedicated NICU and PICU facilities, enabling comprehensive evaluation and management of neonates and young children with congenital surgical anomalies. Data were extracted for all eligible patients during the period from 1 June 2023 to 30 June 2025. The study population comprised pediatric patients aged 0–5 years who were diagnosed with congenital surgical systemic anomalies (CSSA). Children were eligible for inclusion if they had a documented congenital surgical anomaly that required surgical intervention or evaluation, and had undergone cardiac assessment via echocardiography performed by a pediatric cardiologist. Patients were excluded if they had acquired, non-congenital cardiac diseases or if their medical records were incomplete or missing key diagnostic data. Data were obtained from electronic medical records (EMR), which included demographic details, family history, prenatal history, type of congenital surgical anomaly, and echocardiographic findings. A structured data collection proforma was used to ensure uniform extraction. Surgical anomalies were classified into Gastrointestinal, Urogenital, Musculoskeletal, Craniofacial, and Other systemic anomalies based on established pediatric surgical categorizations.

Cardiac evaluation was performed using a standardized echocardiographic protocol by a single pediatric cardiologist to minimize inter-observer variability. Echocardiography was conducted using a GE Healthcare echocardiography system (GE Healthcare Vivid S6, made in France,2021) and included assessment for septal defects, duct-dependent lesions, outflow tract

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abnormalities, valvular lesions, and complex congenital heart diseases. This ensured consistent diagnostic methodology across the study period

A formal sample size calculation was not required, as the study was retrospective in nature. Instead, a total enumeration approach was adopted, including all children who met the inclusion criteria during the two-year study period. This yielded a sample size of 55 patients (Figure 1), and 4 were excluded in view of missing data, representing the complete cohort available for analysis.

Selection bias was minimized by including all consecutive eligible cases within the study period (a sample of convenience from the EMR, without omission). Measurement bias was limited as all echocardiograms were performed by a single pediatric cardiologist using the same equipment and protocol. Information bias was minimized through the use of a structured proforma and standardized classification of anomalies

Quantitative variables such as age were recorded as continuous data, while categorical variables (e.g., anomaly category, CHD type) were expressed as proportions. No transformations or re-coding of variables were required.

Descriptive statistics were used to summarize the data. Categorical variables were expressed as frequencies and percentages. Statistical analysis adhered to standard epidemiological reporting guidelines as recommended by STROBE.

Ethical approval was obtained from the Institutional Ethics Committee before initiation of the study. As this investigation relied solely on retrospective chart review, individual patient consent was waived in accordance with institutional and ICMR ethical guidelines. Patient confidentiality was strictly preserved by anonymizing data and restricting access to authorized investigators only

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RESULTS

A total of 55 children were screened initially and later 4 were excluded in view of missing data and total 51 children with congenital surgical systemic anomalies (CSSA) who underwent complete echocardiographic evaluation were included in the study. The data were entered into Microsoft Excel and analyzed using SPSS version 29.0, and variables were expressed as frequencies and percentages.

The age and sex distribution of the study population is shown in Table 1. Most children presented during the neonatal period, with n = 30 (58.82%) aged less than one month, followed by n = 6 (11.76%) aged 1–6 months, n = 8 (15.68%) aged 6–12 months, and n = 7 (13.72%) aged more than one year. A mild male predominance was observed, with 29 males (56.86%) and 22 females (43.13%).

Family history, prenatal history, associated extracardiac anomalies, system-wise involvement, and the spectrum of congenital surgical anomalies are summarized in Table 2. None of the children had a positive family history of congenital anomalies (n = 0, 0%). Antenatal or prenatal risk factors were present in n = 2 children (3.92%).

After regrouping overlapping terminologies, anorectal malformations were the most common congenital surgical anomaly, observed in n = 21 children (41.2%). This was followed by esophageal atresia with or without tracheoesophageal fistula in n = 8 (15.7%) and congenital diaphragmatic hernia in n = 6 (11.8%). Other anomalies included thyroglossal cyst in n = 5 (9.8%), biliary atresia (cystic) in n = 2 (3.9%), ileal atresia in n = 2 (3.9%), and cleft lip and cleft palate in n = 2 (3.9%). Cleft uvula, developmental dysplasia of the hip, meningomyelocele, rectal prolapse, and recurrent umbilical hernia were each observed in n = 1 child (2.0%). Each child was classified under a single primary surgical diagnosis to avoid duplication.

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Details regarding surgical intervention are shown in Table 3. Surgical procedures for surgical anomaly were performed in n = 49 (96.08%), while n = 2 (3.92%) did not undergo surgery during the study period. cardiac surgical intervention was done in n=1(1.8%), and no surgical cardiac intervention in n=50(98.2%)

Structural congenital heart disease was identified in 24 of 51 children (47.1%), as summarized in Table 4. The detailed echocardiographic findings are presented in Tables 5A and 5B. Multiple cardiac lesions were observed in several children, resulting in lesion-wise frequencies exceeding patient-level prevalence.

DISCUSSION

The present study demonstrates a high prevalence of structural congenital heart disease (CHD) among children with congenital surgical systemic anomalies (CSSA), with nearly half of the cohort (47.1%) showing associated cardiac defects. This finding confirms that cardiac involvement is common in children presenting with major congenital surgical anomalies and is not limited to a specific organ system. Gastrointestinal anomalies constituted the largest subgroup in this cohort, followed by musculoskeletal and urogenital anomalies. Among structural cardiac lesions, septal defects were the most common. Pulmonary arterial hypertension was a frequent associated echocardiographic finding but was analyzed separately and excluded from CHD prevalence calculations. Despite the high prevalence of CHD, only a small proportion of children required cardiac intervention, suggesting that many lesions were clinically silent but potentially significant in the perioperative period.

When compared with population-based data, the prevalence of structural CHD observed in the present study far exceeds the reported birth prevalence of approximately 0.8–1.2% in the

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general pediatric population.[1,2,3] This marked difference supports earlier observations that extracardiac congenital anomalies and CHD frequently coexist due to shared embryological pathways. The developmental field defect theory suggests that disturbances occurring during early organogenesis may simultaneously affect multiple organ systems, including the heart and gastrointestinal tract.[10,11]

The predominance of gastrointestinal anomalies in the present cohort and their frequent association with CHD is consistent with several previous studies. Gokhroo et al. reported CHD in approximately 42–44% of children with major gastrointestinal malformations[4], while Cho et al. and Balasubramaniyam et al. documented even higher prevalence rates, approaching 54–65% in selected groups[5,7]. Similar findings have been reported in neonatal cohorts by Oncel et al.[6]. These observations are biologically plausible, as the development of the primitive foregut and the heart occurs during overlapping periods of gestation, making both systems vulnerable to common embryological insults .[4,5]

Anorectal malformations and esophageal atresia with or without tracheoesophageal fistula were among the most frequently observed surgical diagnoses in the present study. These anomalies are well-recognized components of associations such as VACTERL, in which cardiac defects form a core diagnostic criterion.[8,9] The strong association between anorectal malformations, EA/TEF, and CHD observed in this study is therefore consistent with established syndromic and non-syndromic patterns described in the literature.

Although urogenital and musculoskeletal anomalies accounted for a smaller proportion of cases in this cohort, the high frequency of associated structural CHD within these groups is noteworthy. Similar associations have been described in chromosomal and genetic syndromes, including 22q11.2 deletion syndrome and trisomy 21, where cardiac, renal, and skeletal anomalies frequently coexist.[13,14,15] While subgroup sizes in the present study were small,

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these findings support the need for careful cardiac evaluation in children with multisystem involvement.

With respect to the spectrum of cardiac lesions, atrial septal defects (excluding isolated patent foramen ovale) were the most common structural abnormalities identified in the present study. Comparable patterns have been reported in Indian and international cohorts evaluating CHD in children with extracardiac anomalies .[4,7,16] Ventricular septal defects constituted the second most common lesion, consistent with epidemiological data from population-based and hospital-based studies .[20,21,22] The relatively lower proportion of duct-dependent and complex cardiac lesions observed in this study aligns with reports suggesting that such defects often present very early and are identified during initial neonatal evaluation.[23]

The coexistence of CHD in children undergoing surgery for extracardiac malformations has important clinical implications. Several studies have shown that undiagnosed cardiac defects may increase perioperative morbidity, prolong ventilation, and adversely affect postoperative outcomes, particularly in neonates and young infants.[12,17,19] The findings of the present study support these concerns, as many cardiac lesions identified were not clinically apparent at presentation, underscoring the importance of routine preoperative echocardiographic screening.

Genetic factors likely contribute significantly to the coexistence of CHD and extracardiac anomalies. Advances in molecular genetics have demonstrated that up to one-third of CHD cases may be associated with identifiable genetic or chromosomal abnormalities.[13,14] Recognition of multisystem involvement, particularly in children with urogenital or musculoskeletal anomalies, should therefore prompt consideration of genetic evaluation and counseling, as emphasized in previous studies.[15,24,25]

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This study has certain limitations. Its retrospective design relies on the completeness of available medical records, and subtle anomalies may have been underreported. The single-centre nature of the study may limit generalizability, although the tertiary-care setting ensured inclusion of a wide spectrum of complex congenital anomalies. Small numbers in certain anomaly subgroups also limit the strength of subgroup comparisons. Despite these limitations, the study provides valuable real-world data highlighting the substantial burden of structural congenital heart disease among children with congenital surgical systemic anomalies.

CONCLUSION

The present study demonstrates a substantial coexistence of congenital heart disease among children with congenital surgical systemic anomalies, with a prevalence far exceeding that of the general pediatric population. This association was consistently observed across multiple anomaly groups, particularly gastrointestinal anomalies. Septal defects were the most frequently detected cardiac lesions, and a significant proportion of children had clinically silent CHD. These findings strongly support the incorporation of routine echocardiographic screening into the diagnostic evaluation of all children with congenital surgical anomalies to optimize perioperative planning, reduce morbidity, and ensure appropriate long-term cardiology follow-up.

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Figure 1. Flow diagram of study participant selection and echocardiographic evaluation

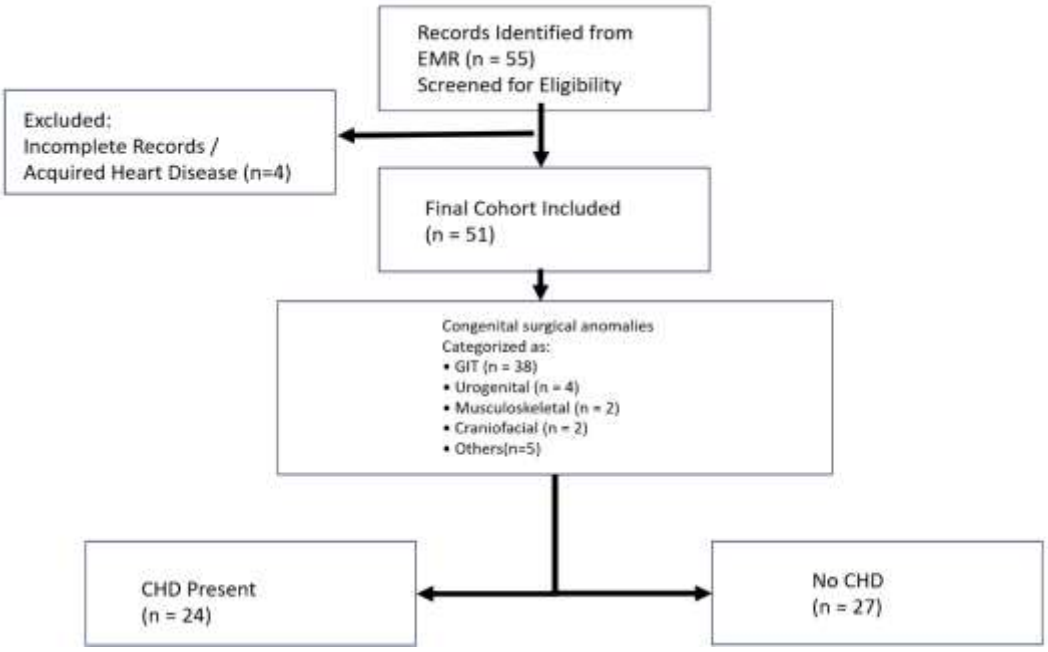

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Table 1. Demographic profile of children with congenital surgical systemic anomalies (n = 51)

Variable	n	%
Age distribution		
< 1 month	30	58.82
1–6 months	6	11.76
6–12 months	8	15.68
> 1 year	7	13.72
Sex distribution		
Male	29	56.86
Female	22	43.13

Table 2. Family history, prenatal history, associated extracardiac anomalies, and surgical intervention details

Variable	n	%
Family history of congenital anomalies		
Present	0	0
Absent	51	100
Prenatal / antenatal risk factors		
Present	2	3.92
Absent	49	96.08

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Variable	n	%
System-wise involvement		
Gastrointestinal	38	74.50
Urogenital	4	7.83
Musculoskeletal	6	11.76
Craniofacial	2	3.92
Other systems	5	9.80
Spectrum of congenital surgical anomalies (deduplicated)		
Anorectal malformations	21	41.2
Esophageal atresia ± tracheoesophageal fistula	8	15.7
Congenital diaphragmatic hernia	6	11.8
Biliary atresia (cystic)	2	3.9
Ileal atresia	2	3.9
Cleft lip and cleft palate	2	3.9
Thyroglossal cyst	5	9.8
Cleft uvula	1	2.0
Developmental dysplasia of hip	1	2.0

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Variable	n	%
Meningomyelocele	1	2.0
Rectal prolapse	1	2.0
Recurrent umbilical hernia	1	2.0
Total	51	100

Table 3. Surgical and Cardiac Interventions Performed (n = 51)

Intervention	n	%
Surgical intervention for CSSA		
Performed	49	96.08
Not performed	2	3.92
Cardiac intervention		
Surgical intervention (PDA closure)	1	1.8%
No surgical cardiac intervention required	50	98.2%

Table 4. Prevalence of Structural Congenital Heart Disease

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Variable	n	%
Children with ≥1 structural CHD	24	47.06
Children without structural CHD	27	52.94

Table 5A. Structural Cardiac Lesions Identified on Echocardiography (Lesion-wise Frequency)

Structural cardiac lesion	n	%
Atrial septal defect*	33	64.71
Ventricular septal defect	12	23.53
Patent ductus arteriosus	8	15.67
Valvular pulmonary stenosis	3	5.88
Complex congenital heart disease	3	5.88

Table 5B. Associated Echocardiographic Findings (Not Classified as Structural CHD)

Finding	n	%
Pulmonary arterial hypertension	25	49.02
Patent foramen ovale	14	27.45
Pericardial effusion	6	11.76