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ORIGINAL RESEARCH

Congenital Cardiac Malformations in Adult and Foetal Cadavers

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Abstract:

Background: Congenital cardiac malformations are a significant subset of developmental abnormalities, affecting approximately 6-8 per 1,000 live births and 2% of stillbirths. These malformations account for nearly one-quarter of all developmental anomalies, yet the etiology remains unknown in over 90% of cases. Hereditary factors and environmental conditions, such as high altitude and seasonal variations, have been implicated. The prevalence and types of congenital cardiac malformations in North India are not well-documented, prompting this study to investigate these aspects in adult and fetal cadavers at Bhagalpur.

Materials and Methods: The study was conducted in the Department of Anatomy at Jawaharlal Nehru Medical College, Bhagalpur, using 25 adult and 45 fetal cadaver hearts, preserved in 10% formaldehyde solution. The hearts were dissected using a standardized method, examining and classifying malformations according to abnormalities in position, developmental progress, and visceral arches. Morphological features were assessed with tools including a dissection kit, magnifying lens, torch, probe, and measuring tape.

Results: Out of 70 cases studied, 14 (20%) displayed congenital cardiac malformations, with a higher incidence in adults (24%) than in fetuses (17.78%). A total of 25 anomalies were identified among the 14 cases, with 10 types of anomalies observed. The most common malformation was atrial septal defect, followed by anomalous positions of the great arteries and cor biloculare. Coexistent external anomalies were found in 3 of the 14 cases (21.43%), affecting the face and multiple systems.

Conclusion: The incidence of congenital cardiac malformations in Bhagalpur is consistent with findings from other regions of India and internationally. Atrial septal defect is the most prevalent malformation in this population, aligning with patterns observed in other Indian studies but differing from international reports.

Keywords: Congenital cardiac malformations, atrial septal defect, cadaveric study, North India, cardiac anomalies, developmental abnormalities.

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Introduction

Congenital cardiac malformations (CCMs) represent a significant portion of developmental abnormalities, comprising approximately one-quarter of such anomalies globally (1). They occur at a rate of 6-8 per 1,000 live births and contribute to 2% of stillbirths (2). Despite their prevalence, the etiology of congenital heart diseases is largely unknown, with less than 10% of cases having a clear cause-effect relationship (3). As a result, preventative measures remain elusive for the majority of these conditions.

Both sexes are equally affected by CCMs, although specific malformations may show a preference for one gender (4). Familial patterns of CCMs, particularly in siblings, suggest a hereditary component, as siblings often present with the same type of congenital heart disease (5). This familial tendency is notably strong in atrial septal defects associated with bony abnormalities, such as in Holt-Oram syndrome (6). However, heredity appears less significant in twins and triplets than previously assumed (7).

Environmental factors are also implicated in the development of CCMs. High altitude and seasonal variations are known to increase the incidence of conditions like patent ductus arteriosus and atrial septal defects (8). For instance, children born at high altitudes exhibit higher rates of these malformations. Seasonal peaks occur from December to February, with the highest incidence reported from April to September (9).

Genetic and chromosomal aberrations also predispose individuals to CCMs. Syndromes such as Marfan's, Ehlers-Danlos, and Hurler's are well-documented genetic disorders associated with these malformations (10). Chromosomal abnormalities, including those seen in Down syndrome (Mongolism), Turner syndrome, and trisomy syndromes, are linked with a high percentage of congenital heart disease cases (11).

Some cardiac anomalies are associated with extracardiac malformations. Acardia, a rare condition where the heart is absent, occurs in 1 in 34,600 births and typically affects monochorionic twins reliant on the co-twin's heart for development (12). Despite the global recognition of these issues, there is a scarcity of data on the prevalence of CCMs in North India. This study aims to fill that gap by examining the frequency and types of congenital cardiac malformations in adult and fetal cadavers at Bhagalpur.

Materials and Methods

Collection of Samples: The present study was conducted in adult and fetal cadavers at the Department of Anatomy, Jawaharlal Nehru Medical College, Bhagalpur. A total of 70 specimens, comprising 25 adult hearts and 45 fetal hearts, were examined. All specimens were preserved in 10% formaldehyde solution prior to dissection and analysis.

Method of Collection of Specimens: The dissection process involved precise anatomical techniques to ensure comprehensive examination of the heart structures. The procedure was as follows:

1. **Preparation of Sternum:**
    - The manubrium of the sternum was transversely cut with a saw immediately inferior to its junction with the first costal cartilage, extending as far posteriorly as possible.
    - Subsequent ribs and intercostal spaces up to the level of the xiphisternal joint were also cut.

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- The inferior part of the sternum, along with costal cartilages, was turned downward onto the superior part of the anterior abdominal wall.
- Fat, fibrous tissue and the thymus were carefully separated from the pericardium.

2. **Examination of Fetal Cadavers:**
   - Fetal cadavers were inspected for external malformations.
   - After retracting the sternum, the position of the heart and the pericardium was assessed.
   - A vertical cut was made, and lower ends of these incisions were joined by a horizontal cut at the diaphragm level.
   - The fibroserous pericardium was turned upwards to examine the pericardial cavity.
   - The great vessels were then cut, and the heart was removed en masse for detailed examination.

Dissection of Heart Chambers:

- **Right Atrium:**
  - The right atrium was opened by inserting a knife at the apex of the auricle and extending it backward close to the anterior aspect of the superior vena cava and downward to the inferior vena cava, anterior to the sulcus terminalis.
  - The resulting flap was turned forward, and the cavity was cleaned of blood clots.
  - The following morphological features were examined: openings of the superior and inferior vena cavae, opening of the coronary sinus, interatrial septum for fossa ovalis and patent foramen ovale, and the right atrioventricular orifice.
- **Right Ventricle:**
  - The cavity was opened via three incisions:
    1. A transverse incision across the infundibulum below the pulmonary trunk.
    2. An oblique incision from the end of the first incision downward along the left margin of the coronary groove.
    3. An incision following the line of the anterior interventricular groove.
  - The triangular flap was turned to the right, and the cavity was examined for the supraventricular crest, hypoplasia, papillary muscles, chordae tendineae, moderator band, and inflow/outflow orifices and valves of the right ventricle, as well as the interventricular septum.
- **Left Ventricle:**
  - A vertical incision was made along the left border towards the apex.
  - The cavity was cleaned and examined for the aortic vestibule, papillary muscles, chordae tendineae, hypoplasia, orifices and valves of the left ventricle, and both membranous and muscular parts of the interventricular septum.
- **Left Atrium:**
  - The left atrium was opened with three incisions: one horizontal and two vertical, passing medial to the terminations of the pulmonary veins.
  - The posterior wall was turned upwards, and the cavity was examined for the orifices of the left atrium, fossa lunata, and interatrial septum.

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Results

The cadaveric study on congenital cardiac malformations included 70 specimens, comprising 25 adult and 45 fetal hearts. Among these, 14 cases (20%) exhibited congenital cardiac malformations. The incidence of congenital cardiac malformations was higher in adults (24%) than in fetuses (17.78%).

A total of 25 anomalies were identified in the 14 cases with congenital malformations, encompassing 10 distinct types of anomalies. The most common congenital cardiac malformation was atrial septal defect (ASD), followed by anomalous positions of the great arteries and cor biloculare, listed in decreasing order of frequency. Three of the 14 cases (21.43%) with congenital cardiac malformations also had coexistent external anomalies, affecting facial and multiple systems.

Summary of Congenital Cardiac Malformations

Parameter	Adults (n=25)	Fetuses (n=45)	Total (n=70)
Total cases with malformations	6 (24%)	8 (17.78%)	14 (20%)
Total anomalies identified	15	10	25
Types of anomalies	6	4	10
Most common malformation	ASD	ASD	ASD
Coexistent external anomalies	1 (16.67%)	2 (25%)	3 (21.43%)

Distribution of Anomalies

Anomaly Type	Number of Cases	Percentage (%) of Total Malformations
Atrial Septal Defect (ASD)	6	42.86
Anomalous Position of Great Arteries	4	28.57
Cor Biloculare	2	14.29
Other Anomalies	3	21.43

Coexistent External Anomalies

Anomaly Type	Number of Cases	Percentage (%) of Total Cases with Malformations
Facial Anomalies	2	14.29
Multiple System Anomalies	1	7.14

Discussion

The prevalence of congenital cardiac malformations (CCMs) in this study, observed in 20% of the examined cadavers, aligns with previous reports indicating a high incidence of developmental cardiac abnormalities [1]. This finding underscores the significance of

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congenital heart defects, which account for nearly a quarter of all developmental anomalies [1,3].

The most common anomaly identified was atrial septal defect (ASD), consistent with other studies that highlight its prevalence among congenital heart defects [3,5]. The incidence of ASD in this study is similar to findings in other parts of India, indicating a possible regional trend [4]. However, variations in ASD prevalence between India and other countries suggest the influence of genetic and environmental factors [2].

The presence of multiple anomalies in several cases suggests that congenital cardiac malformations often occur in complex patterns, requiring detailed examination to understand their interactions and impact on cardiac function. This complexity may reflect the multifactorial etiology of CCMs, involving both genetic predispositions and environmental triggers [5].

Heredity plays a notable role in the development of congenital heart disease, as evidenced by the higher incidence of CCMs in siblings and certain syndromes such as Holt-Oram syndrome, which is associated with atrial septal defects and skeletal anomalies [1]. Genetic syndromes like Marfan's, Ehlers-Danlos, and Hurler's, along with chromosomal abnormalities such as Down syndrome and Turner's syndrome, are known to predispose individuals to CCMs [5,4].

Environmental factors also contribute to the development of congenital heart defects. High altitude and seasonal variations have been associated with increased incidence rates, particularly for patent ductus arteriosus and atrial septal defects [1]. This study supports the notion that individuals with a genetic predisposition may develop CCMs in response to adverse environmental conditions [2].

The presence of coexistent external anomalies in 21.43% of cases with CCMs indicates the potential for widespread developmental disruptions during embryogenesis [4]. Facial and multiple system anomalies observed in this study highlight the need for comprehensive examinations in cases of suspected congenital heart disease to identify and manage associated anomalies effectively [1].

The findings of this study emphasize the need for continued research into the genetic and environmental factors contributing to congenital cardiac malformations, particularly in regions like North India, where data is limited [5]. Improved understanding of these factors can enhance early detection and intervention strategies, ultimately reducing the burden of CCMs on affected individuals and healthcare systems [2].

Conclusion

In conclusion, the study provides valuable insights into the prevalence and complexity of congenital cardiac malformations in North India, reinforcing the need for integrated clinical and genetic research to address these significant developmental disorders effectively.

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