Coarctation of the aorta (CoA) is a congenital heart defect (CHD) characterized by the narrowing of the aorta that can be of varying degree and at any point from the transverse arch to the iliac bifurcation. CoA occurs in about 3 cases per 10,000 births and presents across all age ranges, with varying clinical symptoms, in isolation, or association with other CHDs. Discrepancies between clinical and autopsy diagnoses persist despite progress in medical skills and technology. Hence, we conducted the present study to analyze the level of discrepancy between clinical and autopsy diagnosis of CoA in children.

After taking permission from our institutional ethics committee, we extracted the autopsy records for patients aged 1 day to 12 years diagnosed with coarctation of aorta (by gross and histopathology examination) over 10 years (January 2006 to December 2015). The medical records of these patients were then retrieved from the medical record department and reviewed to check whether a clinical diagnosis of CoA was considered antemortem by the treating physician(s). After a thorough review of each patient’s clinical data, autopsy diagnosis, and the cause of death in the autopsy report, the discrepancy was evaluated as per the Goldmann classification. Data are presented as absolute numbers and percentages for discrete variables and as medians for continuous variables. The study protocol was registered with the clinical trials registry of India (CTRI/2017/10/010293) retrospectively.

**ABSTRACT**
This retrospective study analyzed the level of concordance between clinical and autopsy diagnosis of coarctation of aorta over 10 years. Utilizing the Goldmann classification, the concordance rate was found to be 16%. Major discrepancies (Class I and II) were found in 56% cases and minor discrepancies (Class III and IV) in 28% cases.

**KEY WORDS:** Congenital heart defects, diagnostic errors, heart failure, medical audit, transthoracic echocardiography

How often is coarctation of aorta correctly diagnosed antemortem in children with fatal illnesses? A retrospective review of medical and autopsy records

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The total number of pediatric autopsies performed during the study period was 1907, and in 50/1907 (2.6%) cases CoA was confirmed. The median age of the 50 CoA cases was 2 months (IQR 8.8); 33 males vs. 17 females (male:female ratio 1.9:1). A clinical diagnosis of CoA was considered in 19/50 (38%) cases and the recommended first-line of investigation, namely, a transthoracic echocardiogram (TTE)\(^6\) was done. However, CoA had been detected antemortem in only 8 of these 19 cases.

Our analyses revealed that in only 8/50 (16%) cases the clinical diagnosis of CoA was also confirmed on autopsy to be directly related to death (Class V; no discrepancy).\(^5\) In all these eight cases, a clinical diagnosis of CoA was considered due to the presence of congestive heart failure (CHF) with tachypnea with bilateral weak femoral pulses while a TTE had confirmed the diagnosis. Of these eight cases; four had post-ductal CoA (isolated), one had post-ductal CoA with atrial septal defect (ASD), one had post-ductal CoA with ASD and ventricular septal defect (VSD), and one had pre-ductal CoA with ASD, and were being treated medically awaiting surgery. One case of post-ductal CoA with total anomalous pulmonary venous connection (TAPVC) had died postoperatively.

In 16/50 (32%) cases, the discrepancy was major (Class I) wherein if the correct diagnosis had been done clinically, it would have changed patient management and might have resulted in cure or prolonged survival [Table 1].\(^5\) In only three of these 16 Class I discrepancy cases, a clinical diagnosis of CoA was considered but CoA was not detected on TTE [Table 1].

In 12/50 (24%) cases, the discrepancy was major (Class II) as a missed clinical diagnosis of CoA would have not changed therapy nor impacted survival [Table 1].\(^5\) All 12 cases received appropriate resuscitative management but had succumbed before a TTE could be done.

### Table 1: Coarctation of aorta cases with major Class I and II discrepancies\(^5\) identified in the present study (n=16 and 12, respectively)

| Age/Sex | DHS | Clinical Diagnosis/Condition | Autopsy Diagnosis |
|---------|-----|------------------------------|------------------|
| 1d/M    | 1d  | RDS                          | Post-ductal CoA with ASD |
| 2d/F    | 19h | ACHD with CHF                | Pre-ductal CoA (isolated) |
| 2d/M    | 2d  | ACHD with CHF                | Post-ductal CoA with ASD |
| 2d/M    | 3d  | ACHD with CHF                | Post-ductal CoA with ASD |
| 3d/M    | 3d  | ACHD with CHF*               | Post-ductal CoA (isolated) |
| 21d/M   | 17d | ASD with CHF*                | Post-ductal CoA with ASD |
| 40d/M   | 16d | VSD with CHF*                | Post-ductal CoA with VSD |
| 1.5 months/M | 8h  | DCM with CHF                | Post-ductal CoA with TAA |
| 2 months/F | 1d  | ACHD with CHF with pneumonia | Post-ductal CoA with ASD with pneumonia |
| 2 months/F | 6d  | VSD with CHF                | Post-ductal CoA (isolated) |
| 2.5 months/F | 1d  | Acute myocarditis            | Pre-ductal CoA with ASD |
| 2.5 months/F | 2d  | ACHD with CHF                | Pre-ductal CoA (isolated) |
| 3.5 months/M | 11d | VSD with CHF with pneumonia | Pre-ductal CoA with VSD with pneumonia |
| 5 months/F | 3h  | ACHD with CHF                | Pre-ductal CoA (isolated) |
| 5 months/F | 6h  | ACHD with CHF                | Post-ductal CoA with ASD |
| 9 months/F | 12h | ACHD with CHF                | Post-ductal CoA with VSD |

**Class II discrepancies**

| Age/Sex | DHS | Clinical Diagnosis/Condition | Autopsy Diagnosis |
|---------|-----|------------------------------|------------------|
| NB/M    | 30 min | Gassing soon after birth       | Post-ductal CoA (isolated) |
| NB/F    | 30 min | Gassing soon after birth       | Post-ductal CoA with ASD |
| 1d/F    | 2h   | Gassing soon after birth       | Post-ductal CoA with ASD |
| 1d/M    | 3h   | Admitted in critical condition | Post-ductal CoA with CAVC |
| 3d/M    | 1h   | Admitted in critical condition | Post-ductal CoA with ASD/VSD |
| 4d/M    | 3h   | Admitted in critical condition | Post-ductal CoA with PAPVVC |
| 4d/M    | 3h   | Admitted in critical condition | Pre-ductal CoA with VSD |
| 1.5 months/M | 4.5 h| Admitted in critical condition | Pre-ductal CoA with ASD/VSD |
| 2 months/M | 30 min | Admitted in critical condition | Post-ductal CoA (isolated) |
| 15 months/M | 1h  | Admitted in critical condition | Ductal CoA with VSD |
| 3 years/M | 30 min | Admitted in critical condition | Post-ductal CoA (isolated) |
| 3 years/M | 30 min | Admitted in critical condition | Post-ductal CoA with TAA |

*Transthoracic Echocardiogram was done but CoA not detected. min: minutes; h: hour(s); d: day(s); M: male; F: female; DHS: duration of hospital stay; RDS: respiratory distress syndrome; CoA: coarctation of aorta; ASD: atrial septal defect; ACHD: acyanotic congenital heart defect; CHF: congestive heart failure; VSD: ventricular septal defect; DCM: dilated cardiomyopathy; TAA: tubular hypoplasia of transverse aortic arch; NB: newborn; CAVC: complete atrioventricular canal defect; PAPVVC: partial anomalous pulmonary venous connection.
In 8/50 (16%) cases, the discrepancy was minor (Class III) and the missed clinical diagnosis of CoA was not directly related to death but related to the terminal disease process, namely, pulmonary hypertension (PH) leading to CHF. All these eight cases had already developed pulmonary hypertension before admission due to an associated dominant CHD (three had post-ductal CoA with VSD; two had post-ductal CoA with ASD and VSD, one had post-ductal CoA with double outlet right ventricle, one had post-ductal CoA with transposition of great arteries and one had pre-ductal CoA with VSD) and were being treated medically. One case of post-ductal CoA with VSD with PH and CHF had died postoperatively due to renal failure. Besides, TTE did not detect the CoA in any of these eight cases.

In 6/50 (12%) cases, the discrepancy was minor (Class IV) and the missed clinical diagnosis of CoA was not directly related to death nor the terminal disease process. Of these six cases, four had sepsis due to intestinal perforation, one had bilateral choanal atresia with imperforate anus, and one had hypoxic-ischemic encephalopathy. TTE was not done in these six cases.

To our knowledge, no study has analyzed the level of discrepancy between clinical and autopsy diagnosis of CoA in children. Our results reiterate that CoA is a commonly missed diagnosis, especially in neonates and young infants. Routine evaluation of lower limb pulses and the use of pulse oximeter to measure SpO2 and thorough TTE screening of the aortic arch are urgently needed to avoid oversight of CoA.

There are limitations to our study. It is a single-center study wherein documentation had been completed by different attending physicians and pathologists. Moreover, an autopsy is usually requested when the cause of death is uncertain. This might have led to a false high incidence of major diagnostic errors detected in our study.

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