

# Unraveling Unique Prevalence Patterns: Localized Insights into Associated Cardiac Anomalies in Anorectal Malformation Populations

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## Abstract

**Background:** Anorectal malformation (ARM) is a congenital condition affecting the development of the rectum and anus. Brunei exhibits a notably higher incidence of ARM compared to the global average, prompting a closer examination of associated cardiac anomalies (CAs).

**Methods:** A retrospective observational study (2016-2022) at Brunei's tertiary care centre reviewed ARM cases, emphasizing the identification of associated CAs and their clinical impact.

**Results:** Among 34 cases, 18% demonstrated concurrent CAs and only half of these were significant, necessitating cardiac intervention. These CAs were predominantly associated with low-type ARMs. Patent ductus arteriosus (PDA) and ventricular septal defects (VSD) were the most prevalent associated cardiac anomalies. Attributed cardiac mortality was noted in 1 case.

**Conclusion:** Our study provides unique insights into co-occurring cardiac anomalies in Bruneian ARM cases. Deviations from global averages prompt reevaluation of clinical approaches. The lower incidence of interventions suggests a distinct clinical scenario, highlighting the importance of localized studies in managing ARM cases in this specific population. The study also emphasizes the need for cautious consideration of routine echocardiograms for all ARM cases.

**Keywords:** Cardiac Anomalies, Anorectal Malformation, Associated Anomalies, Neonatal Echocardiogram

## Introduction

Anorectal malformation (ARM) encompasses a diverse range of congenital abnormalities affecting the development of the rectum and anus [1]. Its incidence varies globally, with Brunei notably exhibiting a prevalence higher than the global average, affecting approximately 1 in 1350 births compared to the global average of 1 in 3000 to 5000 births [1,2]. This demographic peculiarity prompts a closer examination of associated anomalies, which extend beyond cardiac conditions.

ARM frequently presents with a constellation of associated anomalies, spanning genitourinary, spinal, gastrointestinal, skeletal, and cardiac systems [2,3]. Among these, cardiac anomalies (CAs) represent a significant subset. Existing literature indicates that at least one-third of individuals with ARM may present with an associated CA. These cardiac anomalies range from minor defects to complex conditions necessitating medical intervention [3,4,5]. Mortality rates within this subgroup range from 10 to 20% [6], underscoring the clinical significance of recognizing and managing these comorbidities.

While the association between ARM and CAs is established, the prevalence and specific types of cardiac anomalies may exhibit considerable variation between populations. Understanding these variations is crucial for tailoring clinical management strategies and providing targeted care for affected individuals. Therefore, this study aims to elucidate the prevalence of associated cardiac anomalies within the Bruneian population, seeking to provide localized insights into the co-occurrence of ARM and CAs in this specific demographic.

This study may offer valuable insights that contribute to a more nuanced understanding of ARM and associated CAs in this population. Moreover, the findings may have broader implications for the management and care of individuals with ARM, emphasizing the importance of tailored clinical approaches that consider demographic variations in congenital anomalies.

## Methods

A retrospective observational study, conducted from January 2016 to December 2022 at Brunei's sole Paediatric Surgery tertiary care centre, involved a meticulous review of medical records for all ARM cases born or transferred to RIPAS Hospital. Only Bruneian citizens were included, and foreign patients were excluded. Emphasis was placed on identifying associated cardiac anomalies, evaluating their clinical significance, and their effect on the course of ARM corrective surgery. A significant CA was defined as a cardiac anomaly that requires medical treatment and/or subsequent surgical intervention. Isolated patent ductus arteriosus (PDA) and persistent foramen ovale (PFO) that have no impact and require no treatment were not considered within the CAs category since both are considered part of normal findings in newborns [7].

## Results

Among the 38 cases of ARM examined, only 34 were included as 4 patients were foreigners, thus excluded. All included patients had their records available and their follow-up ranged between 14 and 66 months. Six cases (18%) demonstrated concurrent CAs, with an equal distribution between genders. Two more patients found to have isolated PDA and PFO and they received no medical treatment for these normal findings, thus both were not considered within the CAs category.

The lower type of ARM (rectum below the level of the pubococcygeal line) appears to be the dominant type associated with CAs (Table-01), with the exception of the 6th case that resulted in neonatal mortality, leaving the type of ARM unknown.

Patient	Gender	Cardiac Anomaly	Type of ARM
*1	Male	ASD, PDA, VSD	Low- No fistula
2	Male	VSD	Low- No Fistula
3	Female	ASD, PDA	Low- Perineal Fistula
*4	Female	VSD, TA	Low- Vestibular Fistula
5	Female	ASD, PDA	Low- Perineal Fistula
*6	Male (D)	VSD, PDA, PA	Unknown
* Requiring intervention (D) Death due to cardiac anomaly			

The majority of these cases exhibited more than one CA, spanning a spectrum including patent ductus arteriosus (PDA), ventricular septal defects (VSD), atrial septal defects (ASD), tricuspid atresia (TA), and pulmonary atresia (PA). Significantly, three of these cases required intervention. The intervention entailed both initial medical therapy and subsequent surgery, excluding the patient who did not survive long enough to undergo cardiac surgery.

None of the patients with CAs experienced issues related to general anesthesia within the neonatal period, including those with significant CAs.

All patients with significant CAs underwent straightforward colostomy formation on the second day of life. Two patients underwent corrective cardiac surgery within the first year of life. Regrettably, the remaining case with significant CAs underwent uneventful colostomy on day two of life, yet passed away at the age of nine weeks due to cardiac arrest attributed to the CAs. Consequently, mortality attributable to CA was noted in one case (3% of all ARM cases and 17% of the CA subgroup).

An echocardiogram (EC) was performed for 26 patients (76%), all within the first week of life. EC was prompted by an abnormal cardiac shadow on chest x-ray in five cases, with two of them confirming significant CAs (Cases 4 and 6 in Table-01). The presence of a murmur led to EC in four cases, with two of them presenting significant CAs, and one with insignificant ones (Cases 1, 2, and 6 in Table-01). The remaining ECs were conducted as part of routine screening for associated anomalies in patients with other anomalies besides ARM (spinal, skeletal, or urinary), two of which had insignificant CAs (Cases 3 and 5 in Table-01).

None of the patients who underwent neonatal surgery had an echocardiogram prior to the procedure, and none experienced haemodynamic instability during or within the first week after the surgery. Discovery of significant CAs in all patients occurred only after colostomy formation, except for female patients with perineal fistula. In our centre, it is customary to initiate with dilatation and perform delayed repair after the age of three months for those with perineal fistula. The anorectoplasty for both surviving patients with significant CAs was postponed until after the cardiac surgery was performed.

## Discussion

Our study presents a distinctive perspective on the co-occurrence of CAs in individuals with ARM within Brunei, revealing findings that deviate from established global trends.

The prevalence of associated CA in our cohort (18%) is notably lower than the reported global average of 30-40% [3-6,8]. Moreover, the rate of intervention for these anomalies in our population (9%) is substantially less than the reported global range of nearly 20% [2,3].

The prevalence of specific cardiac anomalies in our cohort differs from previous studies [9,10]. As depicted in Table-01, patent ductus arteriosus (PDA) and ventricular septal defects (VSD) were the most commonly observed anomalies, followed by atrial septal defects (ASD). Notably, tricuspid atresia and pulmonary atresia were present in our cohort, although they are less commonly reported in ARM cases globally. Surprisingly, Tetralogy of Fallot, a commonly reported association with ARM, was not observed in our series during the study period [10,11,12]. This discrepancy may suggest that Bruneian patients with ARM possess a different CA profile. Despite these differences, the observed mortality rate attributed to CA (17% of cases with ARM associated with CA) aligns with the available data (between 10-20%) [5,12,13].

The majority of patients with ARM and associated CAs in our study exhibit the low-type ARM. It's crucial to highlight that this inverse relationship between the severity of ARM and the occurrence of CAs doesn't apply to associated genitourinary or spinal anomalies. These anomalies, more commonly associated with the high-type ARM, were not the primary focus of our study [3,4].

Our results indicate that performing routine echocardiograms (EC) may not be necessary, as only 11% revealed CAs, and these were insignificant ones. EC should be reserved for cases where radiological and/or clinical suspicion suggests a likelihood of associated CA. This approach is consistent with recommendations for many other congenital anomalies [14].

In our series, the presence of CAs did not hinder the progression of colostomy formation during the neonatal period, which was well-tolerated. Therefore, the authors suggest that short neonatal surgeries (colostomy or simple anoplasty) should not be delayed if EC can not be done in time even if the situation specifically calls for preoperative EC due to radiological and/or clinical suspicion as long as haemodynamic stability is ensured.

It is worth noting that despite the heightened prevalence of ARM in Brunei, the incidence of associated significant cardiac anomalies remains lower. This did not impact neonatal surgical planning. These findings underscore the importance of tailored clinical strategies in managing patients with anorectal malformations within this specific population.

In addition, This anomaly warrants further investigation into potential genetic or environmental factors influencing this deviation from global trends. Genetic studies may hold the key to uncovering underlying genetic variations contributing to this unique prevalence pattern in Brunei [15].

## Conclusions

This study provides valuable insights into the co-occurrence of CAs in individuals with ARM within the Bruneian population. The prevalence of associated CAs in our cohort demonstrates a notable deviation from the global average.

The lower incidence of significant CAs requiring intervention in our cohort raises intriguing questions about the unique cardiac profile of Bruneian patients with ARM.

Our study also highlights the potential reconsideration of routine echocardiograms for all ARM cases.

In summary, our study underscores the importance of localized studies in uncovering demographic variations in congenital anomalies. The distinctive prevalence pattern observed in Brunei prompts a reevaluation of clinical approaches in managing individuals with anorectal malformations in this specific population.

## Limitations

This study sheds light on the prevalence of cardiac anomalies in individuals with anorectal malformations in Brunei. However, it's important to note limitations. The absence of a control group and varying definitions of "significant" cardiac anomalies in the literature may lead to data variability among studies. The small sample size and single-center focus may limit generalizability. Additionally, external factors and unique influences in the Bruneian population are not considered. Importantly, variations in data published in the literature can also impact the actual rates and incidence of cardiac anomalies associated with anorectal malformations. Future research should address these limitations for a more comprehensive understanding.

## Conflict of Interest

The authors declare no conflict of interest.

## Acknowledgement

Not applicable

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