PERINATAL JOURNAL

Original Article

Perinatal Journal 2026; 34(2):187-192

https://doi.org/10.57239/prn.26.03420018

Profile of congenital heart disease with pulmonary arterial hypertension

Gina Noor Djalilah[1,6], Bagus Setyoboedi[2*], Mahrus A Rahman[2], Meity Ardiana[3], Ali Rohman[4], I Ketut Alit Utamayasa[2], Taufiq Hidayat[2], Muhammad Perdana Airlangga[6], Sri Widyaningsih[5,6], Adicitra Aurel Bestari6, Clevia Revi[6]

[1]Doctoral Program of Medical Science, Faculty of Medicine, Universitas Airlangga, Surabaya 60131, Indonesia
[2]Department of Paediatrics, Faculty of Medicine, Universitas Airlangga, Surabaya 60286, Indonesia
[3]Department of Cardiology, Faculty of Medicine, Universitas Airlangga, Surabaya 60286, Indonesia
[4]Department of Chemistry, Faculty of Science and Technology, Universitas Airlangga, Surabaya, 60115, Indonesia
[5]Department of Clinical Pathology, Siti Khodijah Hospital, Indonesia, Sidoarjo 61257, Indonesia
[6]Faculty of Medicine, Universitas of Muhammadiyah Surabaya, Surabaya 60113, Indonesia

Abstract

Congenital Heart Disease (CHD) is a significant health issue globally, with Pulmonary Arterial Hypertension (PAH) being a serious and life-limiting complication. Understanding the clinical profile of patients with this comorbidity is crucial for effective management and improved outcomes. This study aims to describe the demographic and clinical characteristics of CHD patients with PAH. This was a retrospective, descriptive study conducted at Siti Khodijah Hospital, Sidoarjo, East Java, Indonesia. Data were collected from the medical records of all patients diagnosed with CHD and PAH from 2023-2024. Patient data included demographics age, gender, type of CHD, clinical symptoms, echocardiography, cardiac catheterization results, and treatment modalities. The data were analyzed to identify the prevalence of specific CHD types and the associated clinical features. A total of 107 pediatrics patients were included in the study. The majority were female (52%). The most common type of CHD identified was ventricular septal defect (31%), atrial septal defect (27%), followed by patent ductus arteriosus (17%), and others (25%). Common clinical symptoms at diagnosis included shortness of breath, cyanosis, fatigue. Diagnostic findings frequently revealed right ventricular hypertrophy, increased pulmonary artery pressure with mild PAH (29%), moderate PAH (38%) and severe PAH (34%). A proportion of patients had received prior surgical intervention, but still developed or had persistent PAH. The findings of this study provide a preliminary profile of CHD patients with PAH at our institution hospital. The data highlight the prevalence of certain CHD types and the associated clinical manifestations. These insights are essential for informing clinical practice, improving diagnostic protocols, and developing targeted treatment strategies for this specific patient population in our hospital.

Keywords: Congenital heart disease, Pulmonary arterial hypertention, Profile characteristic, Echocardiography, Catheterization.

Introduction

Globally, Congenital Heart Disease (CHH) represents a significant public health challenge, with an incidence of approximately 8 cases per 1000 live births. While significant advancements have been made in its diagnosis and management, complications remain a major concern. One of the most serious and life-limiting complications is Pulmonary Arterial Hypertension (PAH). This condition significantly impacts the quality of life and survival rates of CHD patients.[1]

The pathogenesis of pulmonary arterial hypertension in congenital heart disease patients is rooted in long-standing increases in pulmonary blood flow stemming from an unrepaired left-to-right shunt. This hemodynamic stress induces vascular remodeling within the lungs, ultimately leading to elevated pulmonary vascular resistance.[2] This process can ultimately progress to Eisenmenger syndrome, which is irreversible and associated with high morbidity and mortality. Key to the progressive nature of PAH in CHD are the underlying structural and molecular changes in the lung's blood vessels, which have a direct impact on patient prognosis. Accordingly, the updated clinical guidelines classify CHD-related PAH as a highly complex subgroup that necessitates a multidisciplinary management approach.[3]

Although advances in diagnosis and therapy have improved the life expectancy of CHD patients, the presence of PAH significantly alters the clinical spectrum and management strategy. International registry data indicate that demographic characteristics, type of congenital heart defect, timing of surgical correction, and degree of pulmonary

Correspondence: Bagus Setyoboedi, Department of Paediatrics, Faculty of Medicine, Universitas Airlangga, Surabaya 60286, Indonesia, **e-mail:** bagus.setyoboedi@fk.unair.ac.id, **Received:** December 26, 2025 **Accepted:** March 1, 2026

hypertension influence patients' clinical outcomes.[4] Nevertheless, the distribution of clinical and demographic characteristics of patients varies greatly between regions, particularly in developing countries that face limited access to early diagnosis and definitive therapy.[5] This condition indicates the need for mapping of patient profiles specifically based on the local healthcare service context.

Understanding the unique challenges posed by the coexistence of CHD and PAH is vital for improving patient care. Managing these patients requires a comprehensive treatment plan, as their clinical presentation is often complex and highly variable. Therefore, to tailor effective management strategies, healthcare providers require a detailed understanding of patients' demographic and clinical characteristics.[1,6]

The primary objective is to provide a comprehensive description of the demographic and clinical features of patients suffering from both congenital heart disease and pulmonary arterial hypertension. The anticipated detailed profile will advance the current understanding of this comorbidity and serve as a foundation for strategies to enhance clinical outcomes.

Materials and Methods

The clinical profiles of patients with Congenital Heart Disease (CHD) complicated by Pulmonary Arterial Hypertension (PAH) were analyzed using a retrospective, descriptive study design. The study was conducted at Siti Khodijah Hospital using data from January 2023 to December 2024. A total sampling approach was employed, including all patients who had received a diagnosis of both CHD and PAH during the study period. Data were systematically extracted from the medical records of each patient. Data collection encompassed demographics (age at diagnosis, gender), clinical characteristics (CHD type, symptoms like dyspnea, fatigue, and cyanosis), diagnostic results (echocardiography, cardiac catheterization), and treatment details. This information was then analyzed using descriptive statistics to identify the frequency of various CHD types and delineate the associated clinical features.

Results

This investigation enrolled 107 pediatric cases involving the comorbidity of Congenital Heart Disease (CHD) and Pulmonary Arterial Hypertension (PAH). The cohort was characterized by a slight female predominance, with females accounting for 52% (n=56) and males 48% (n=51) of the sample.

Distribution of Congenital Heart Defects Analysis of the patient records revealed a varied distribution of CHD types. The most common defect was Ventricular Septal Defect (VSD), making up nearly a third of cases. Atrial Septal Defect (ASD) and Patent Ductus Arteriosus (PDA) followed in frequency. The remaining patients had other, less common or more complex CHD types, categorized as "Others." A complete breakdown of these findings is available in Table 1.

Table 1. Distribution of congenital heart disease types (n=107)

CHD Type	Number of Patients (n)	Percentage (%)
Ventricular Septal Defect (VSD)	33	31
Atrial Septal Defect (ASD)	29	27
Patent Ductus Arteriosus (PDA)	18	17
Others	27	25
Total	107	100

Clinical and diagnostic assessments frequently highlighted significant cardiac and pulmonary changes. Common findings included right ventricular hypertrophy, indicating the heart's adaptation to increased pressure, and elevated pulmonary artery pressure.

The severity of pulmonary arterial hypertension was categorized based on established criteria, with the distribution as shown in Table 2.

Table 2. Severity of Pulmonary Arterial Hypertension (PAH) (n=107)

PAH Severity	Number of Patients (n)	Percentage (%)
Mild PAH	31	29
Moderate PAH	40	38
Severe PAH	36	33
Total	107	100

Prior interventions and persistent PAH

A notable finding from this study was that a substantial proportion of the pediatric patients included in the cohort had a history of prior surgical intervention for their underlying congenital heart disease. These interventions typically aimed to correct the primary cardiac defect and, ideally, prevent or mitigate the development of pulmonary hypertension. However, despite these efforts, these patients either developed new-onset PAH subsequent to their surgery or experienced persistent PAH that remained significant even after surgical repair.

This observation underscores the complex and often progressive nature of pulmonary arterial hypertension in the context of CHD, particularly in pediatric populations. It suggests that while surgical correction of the structural defect is crucial, it may not always be sufficient to fully reverse or halt the progression of pulmonary vascular disease. Tis highlights the need for continued vigilance, long-term follow-up, and potentially advanced medical therapies even in patients who have undergone successful surgical repair. The presence of persistent or de novo PAH post-surgery presents a significant challenge in patient management and contributes to the morbidity associated with this dual diagnosis.

Discussion

Analysis of cases at Siti Khodijah Hospital showed that Ventricular Septal Defect (VSD) was the predominant congenital heart lesion associated with PAH, occurring in 33 patients. Atrial Septal Defect (ASD) was also common, with 29 cases, and Patent Ductus Arteriosus (PDA) was observed in 18 patients. Similarly, the *Pediatrics Textbook* from the Faculty of Medicine, University of Indonesia (2017) also states that VSD is the most frequent congenital defect linked with PAH.[7] Data from the Centers for Disease Control and Prevention (CDC) confirms that VSD is the most prevalent form of CHD globally. The present findings align with international research, which consistently identifies VSD and ASD as the congenital cardiac defects most frequently linked to the development of PAH in pediatric populations.[8–11] This is attributed to the prolonged left-to-right shunt, which increases pulmonary blood flow and consequently leads to pulmonary vascular remodeling and elevated pulmonary artery pressure.[12]

In terms of gender distribution, the results showed a relatively balanced ratio with a slight predominance of females (52%). This aligns with prior studies from pediatric cardiology centers, which have reported a higher proportion of female patients.[13] Certain congenital cardiac defects, particularly ASD and atrioventricular septal defect (AVSD), occur more frequently in females and are strongly associated with the risk of secondary PAH.[14] Several hypotheses suggest that estrogen signaling may interact with BMPR2 gene expression, contributing to this trend. Furthermore, left-to-right shunt lesions (ASD, VSD, PDA) tend to remain asymptomatic for longer in females, thereby increasing their risk of developing PAH.[15,16]

The main clinical manifestations observed were dyspnea, cyanosis, and easy fatigability, which are consistent with the typical presentation of CHD patients with PAH. Echocardiographic and cardiac catheterization assessments revealed right ventricular hypertrophy and elevated pulmonary artery pressure.[17,18] Interestingly, some patients continued to experience PAH despite undergoing corrective surgery.[19–21] This highlights that while early surgical intervention is crucial, it does not always prevent the persistence of PAH, especially when intervention is delayed or irreversible pulmonary vascular changes have already occurred.[22–24]

The severity distribution of PAH in this study (29% mild, 38% moderate, 33% severe) illustrates that a substantial proportion of patients presented with already advanced disease. This underscores the importance of early detection and timely referral to prevent disease progression.[25–28]

In conclusion, the results of this study align with existing literature, which underscores that the prognosis for patients with CHD is largely determined by the timeliness of diagnosis, the specific nature of the cardiac defect, and the implementation of comprehensive management strategies, inclusive of long-term postoperative surveillance.

Conclusion

Among those with PAH, ventricular septal defect was the predominant congenital heart disease, found in 31% of the study population. This was followed by atrial septal defect, which accounted for 27%, and patent ductus arteriosus, present in 17% of patients. The majority of patients present with chief complaints of shortness of breath, cyanosis, and easy fatigability, reflecting hemodynamic disturbances due to increased pressure in the pulmonary circulation. Diagnostic findings frequently encountered include right ventricular hypertrophy and elevated pulmonary arterial pressure. The severity of PAH in this study varied, with the highest proportion being moderate-grade PAH at 38%, followed by severe PAH at 33% and mild PAH at 29%. This study's findings reinforce the critical role of early detection, timely intervention, and holistic management for congenital heart disease patients vulnerable to PAH, strategies which are essential for enhancing quality of life and minimizing complication rates.

References

1. Fernandes, C. J., Calderaro, D., Assad, A. P. L., Salibe-Filho, W., Kato-Morinaga, L. T., Hoette, S., Piloto, B., Castro, M. A., Lisboa, R. P., & Silva, T. A. F. da. (2021). Update on the treatment of pulmonary arterial hypertension. *Arquivos Brasileiros de Cardiologia*, *117*(4), 750–764. https://doi.org/10.36660/abc.20200702
2. Widiastari, E. F., Graha, W. A., & Pardede, M. (2022). Lung Bullae Due to Septic Pulmonary Embolism in a 4-Year-Old Child: A Case Report. *Pharmacology, Medical Reports, Orthopedic, and Illness Details (COMORBID)*, *1*(3). https://doi.org/10.55047/comorbid.v1i3.355
3. Brida, M., Nashat, H., & Gatzoulis, M. A. (2020). Pulmonary arterial hypertension: closing the gap in congenital heart disease. *Current Opinion in Pulmonary Medicine*, *26*(5), 422–428. https://doi.org/10.1097/MCP.0000000000000695
4. Fathallah, M., & Krasuski, R. A. (2018). A multifaceted approach to pulmonary hypertension in adults with congenital heart disease. *Progress in Cardiovascular Diseases*, *61*(3–4), 320–327. https://doi.org/10.1016/j.pcad.2018.07.017
5. Zimmerman, M., & Sable, C. (2020). Congenital heart disease in low-and-middle-income countries: focus on sub-Saharan Africa. *American Journal of Medical Genetics Part C: Seminars in Medical Genetics*, *184*(1), 36–46. https://doi.org/10.1002/ajmg.c.31769
6. Goldstein, S. A., & Krasuski, R. A. (2022). Pulmonary hypertension in adults with congenital heart disease. *Cardiology Clinics*, *40*(1), 55–67.
7. Pascall, E., & Tulloh, R. M. R. (2018). Pulmonary hypertension in congenital heart disease. *Future Cardiology*, *14*(4), 343–353. https://doi.org/10.2217/fca-2017-0065
8. Kaemmerer, H., Gorenflo, M., Huscher, D., Pittrow, D., Apitz, C., Baumgartner, H., Berger, F., Bruch, L., Brunnemer, E., & Budts, W. (2020). Pulmonary hypertension in adults with congenital heart disease: real-world data from the international COMPERA-CHD registry. *Journal of Clinical Medicine*, *9*(5), 1456. https://doi.org/10.3390/jcm9051456
9. Ahmed, A., Ahmed, S., & Rådegran, G. (2023). Risk assessment in pulmonary arterial hypertension: a step towards clinical implementation based on the 2022 ESC/ERS pulmonary hypertension guidelines. *Pulmonary Circulation*, *13*(2), e12253. https://doi.org/10.1002/pul2.12253
10. Roth, T. S., & Aboulhosn, J. A. (2016). Pulmonary hypertension and congenital heart disease. *Cardiology Clinics*, *34*(3), 391–400. https://doi.org/10.1016/j.ccl.2016.04.002
11. Kaemmerer, H., Apitz, C., Brockmeier, K., Eicken, A., Gorenflo, M., Hager, A., de Haan, F., Huntgeburth, M., Kozlik-Feldmann, R. G., & Miera, O. (2018). Pulmonary hypertension in adults with congenital heart disease: Updated

recommendations from the Cologne Consensus Conference 2018. *International Journal of Cardiology*, *272*, 79–88. https://doi.org/10.1016/j.ijcard.2018.08.078

12. Ruopp, N. F., & Cockrill, B. A. (2022). Diagnosis and treatment of pulmonary arterial hypertension: a review. *Jama*, *327*(14), 1379–1391. https://doi.org/10.1001/jama.2022.4402
13. Jone, P.-N., Ivy, D. D., Hauck, A., Karamlou, T., Truong, U., Coleman, R. D., Sandoval, J. P., del Cerro Marin, M. J., Eghtesady, P., & Tillman, K. (2023). Pulmonary hypertension in congenital heart disease: a scientific statement from the American Heart Association. *Circulation: Heart Failure*, *16*(7), e00080. https://doi.org/10.1161/HHF.0000000000000080
14. Hayvaci Canbeyli, F., Secgen, K., Ezgu, F. S., Tacoy, G., Unlu, S., Arabacı, H. O., Pektas, A., Inci, A., Kaya, E. B., & Sinan, U. Y. (2025). The Role of Genetics in Congenital Heart Disease-Associated Pulmonary Arterial Hypertension. *Pediatric Cardiology*, *47*, 1–9. https://doi.org/10.1007/s00246-025-03847-z
15. Mares, A., Mukherjee, D., Lange, R. A., & Nickel, N. P. (2022). Targeted therapies in patients with pulmonary arterial hypertension due to congenital heart disease. *Current Vascular Pharmacology*, *20*(4), 341–360. https://doi.org/10.2174/1570161120666220811150853
16. Brida, M., & Gatzoulis, M. A. (2018). Pulmonary arterial hypertension in adult congenital heart disease. *Heart*, *104*(19), 1568–1574. https://doi.org/10.1136/heartjnl-2017-312106
17. Wrobel, J. P., Thompson, B. R., & Williams, T. J. (2012). Mechanisms of pulmonary hypertension in chronic obstructive pulmonary disease: a pathophysiologic review. *The Journal of Heart and Lung Transplantation*, *31*(6), 557–564. https://doi.org/10.1016/j.healun.2012.02.029
18. Konopka, M., & Braksator, W. (2013). Pulmonary hypertension-advances in diagnosis and treatment. *FAMILY MEDICINE AND PRIMARY CARE REVIEW*, *15*(4), 561–566.
19. Sommer, N., Ghofrani, H. A., Pak, O., Bonnet, S., Provencher, S., Sitbon, O., Rosenkranz, S., Hoeper, M. M., & Kiely, D. G. (2021). Current and future treatments of pulmonary arterial hypertension. *British Journal of Pharmacology*, *178*(1), 6–30. https://doi.org/10.1111/bph.15016
20. Dignam, J. P., Sharma, S., Stasinopoulos, I., & MacLean, M. R. (2024). Pulmonary arterial hypertension: sex matters. *British Journal of Pharmacology*, *181*(7), 938–966. https://doi.org/10.1111/bph.16277
21. Tang, H., Desai, A. A., & Yuan, J. X.-J. (2016). Genetic insights into pulmonary arterial hypertension. Application of whole-exome sequencing to the study of pathogenic mechanisms. In *American Journal of Respiratory and Critical Care Medicine* (Vol. 194, Issue 4, pp. 393–397). American Thoracic Society. https://doi.org/10.1164/rccm.201603-0577ED
22. Simonneau, G., Gatzoulis, M. A., Adatia, I., Celermajer, D., Denton, C., Ghofrani, A., Gomez Sanchez, M. A., Kumar, R. K., Landzberg, M., & Machado, R. F. (2014). Updated clinical classification of pulmonary hypertension. *Turk Kardiyoloji Dernegi Arsivi: Turk Kardiyoloji Derneginin Yayin Organidir*, *42*, 45–54. https://doi.org/10.3109/9781420094763-4
23. Li, X., Chen, N., Zhou, X., Yang, Y., Chen, S., Song, Y., Sun, K., & Du, Q. (2019). Exercise training in adults with congenital heart disease: a systematic review and meta-analysis. *Journal of Cardiopulmonary Rehabilitation and Prevention*, *39*(5), 299–307. https://doi.org/10.1097/HCR.0000000000000420
24. Low, A., George, S., Howard, L., Bell, N., Millar, A., & Tulloh, R. M. R. (2018). Lung Function, Inflammation, and Endothelin-1 in Congenital Heart Disease–Associated Pulmonary Arterial Hypertension. *Journal of the American Heart Association*, *7*(4), e007249. https://doi.org/10.1161/JAHA.117.007249
25. Hermawan, B. J., Hariyanto, D., & Aprilia, D. (2018). Profil Penyakit Penyakit Jantung

Bawaan di Instalasi Rawat Inap Anak RSUP Dr. M. Djamil Padang Periode Januari 2013 – Desember 2015. *Jurnal Kesehatan Andalas*, *7*(1), 142–148. https://doi.org/10.25077/jka.v7i1.793

26. Ain, N., Hariyanto, D., & Rusdan, S. (2015). Karakteristik Penderita Penyakit Jantung Bawaan pada Anak di RSUP Dr. M. Djamil Padang Periode Januari 2010-Mei 2012. *Jurnal Kesehatan Andalas*, *4*(3). https://doi.org/10.25077/jka.v4i3.388
27. Baksh, A. K., Waworuntu, D. S., & Umboh, A. (2024). Profil Penyakit Jantung Bawaan di Bagian Ilmu Kesehatan Anak RSUP Prof. Dr. RD Kandou Manado Periode September 2022–Agustus 2023. *E-CliniC*, *12*(3), 330–334. https://doi.org/10.35790/ecl.v12i3.55352
28. Diogenes, T. C. P., Mourato, F. A., de Lima Filho, J. L., & Mattos, S. da S. (2017). Gender differences in the prevalence of congenital heart disease in Down's syndrome: a brief meta-analysis. *BMC Medical Genetics*, *18*(1), 111. https://doi.org/10.1186/s12881-017-0475-7