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Persistent Fifth Aortic Arch With Multisystem Congenital Anomalies: A Two-Case Report

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Case Report

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Abstract

Persistent fifth aortic arch (PFAA) is a rare congenital vascular anomaly characterized by a double-lumen transverse aortic arch resulting from persistence of an embryologic vascular channel that normally regresses during fetal development. It is typically identified during evaluation for congenital heart disease and may be associated with additional intracardiac or extracardiac abnormalities. Although PFAA has been described in association with congenital heart defects, concurrent multisystem congenital anomalies have been rarely reported.

We report two male infants with systemic-to-systemic persistent fifth aortic arch confirmed by cross-sectional imaging. In the first case, PFAA was identified during evaluation of a cardiac murmur in an infant with sagittal craniosynostosis and a small ventricular septal defect. In the second case, PFAA was identified in an infant with microcephaly, cerebellar vermis hypoplasia, posterior corpus callosum hypoplasia, right hydronephrosis, and cryptorchidism, who also had a fenestrated atrial septal defect requiring surgical closure. In both patients, the arch was widely patent without obstruction and did not require intervention.

These cases highlight the clinical and imaging features of PFAA and underscore the importance of thorough evaluation in infants presenting with multisystem congenital anomalies. Accurate diagnosis through echocardiography and cross-sectional imaging is essential to distinguish PFAA from other arch abnormalities that may require intervention.

Introduction

Persistent fifth aortic arch (PFAA) is a rare congenital anomaly of the aortic arch system resulting from persistence of an embryologic arch segment that normally regresses during development. During early embryogenesis, the aortic arch system arises from a series of paired pharyngeal arch arteries that remodel to form the mature aortic arch and great vessels [4]. The vascular structure described as persistent fifth aortic arch represents an additional channel connecting the ascending and descending aorta, producing a characteristic double-barreled appearance of the transverse aortic arch [1, 2].

Several anatomical variants of PFAA have been described, including systemic-to-systemic and systemic-to-pulmonary configurations [2]. The systemic-to-systemic form may be hemodynamically insignificant when unobstructed but must be distinguished from other arch abnormalities such as coarctation or interrupted aortic arch [1, 2].

The true incidence of PFAA is unknown. Most descriptions arise from isolated case reports and small case series, and some authors have suggested that the anomaly may be underrecognized because of its resemblance to other arch variants [3]. PFAA may coexist with intracardiac defects such as septal defects or more complex congenital heart disease [2, 5]. Although intracardiac associations have been described, reports detailing concurrent extracardiac congenital anomalies remain limited. We present

two infants with systemic-to-systemic persistent fifth aortic arch in the setting of additional congenital abnormalities.

Case Reports

Case 1

A 5-week-old male born at term via cesarean section was referred for cardiology evaluation after a cardiac murmur was detected at a routine well-child visit. Prenatal history was notable for craniosynostosis. Postnatal evaluation confirmed sagittal craniosynostosis with fusion of the anterior two-thirds of the sagittal suture, resulting in scaphocephaly. He was otherwise clinically stable and feeding well without respiratory distress or cyanosis.

Electrocardiogram demonstrated normal sinus rhythm. Transthoracic echocardiography revealed normal segmental cardiac anatomy and preserved biventricular systolic function. A small anterior mid-muscular ventricular septal defect with left-to-right shunting and a patent foramen ovale were identified. There was no chamber enlargement, and right ventricular systolic pressure was normal.

Evaluation of the aortic arch demonstrated a double-lumen configuration suggestive of persistent fifth aortic arch. Doppler interrogation showed laminar, unobstructed flow through both channels. Computed tomography angiography confirmed a systemic-to-systemic double-barreled aortic arch with unobstructed flow into the descending aorta (Fig. 1).

The ventricular septal defect was hemodynamically insignificant and no cardiac intervention was required. The patient continues routine cardiology follow-up, and neurosurgical management of the craniosynostosis is planned.

Contrast-enhanced computed tomography angiography (CTA), sagittal reconstruction, demonstrating a systemic-to-systemic persistent fifth aortic arch. A parallel accessory channel courses inferior to the native aortic arch, creating a double-barreled configuration. Both lumens are widely patent without evidence of coarctation.

Case 2

A male infant with multiple congenital anomalies was evaluated for congenital heart disease. His medical history was notable for microcephaly with neuroimaging demonstrating hypoplasia of the cerebellar vermis and posterior corpus callosum. Additional congenital anomalies included right hydronephrosis and cryptorchidism, suggesting multisystem developmental involvement.

Electrocardiogram demonstrated sinus rhythm with right atrial enlargement and right ventricular hypertrophy. Transthoracic echocardiography revealed normal segmental cardiac anatomy with preserved biventricular systolic function (Fig. 2). A fenestrated atrial septal defect produced a moderate left-to-right shunt with mild right-sided chamber dilation and septal flattening.

Assessment of the aortic arch demonstrated a double-barreled configuration consistent with systemic-to-systemic persistent fifth aortic arch. Doppler interrogation demonstrated unobstructed flow across both arches. Cross-sectional imaging confirmed patency of the accessory arch without evidence of coarctation or arch hypoplasia.

The atrial septal defect was subsequently closed surgically. The patient remains under cardiology follow-up as part of ongoing care for his congenital cardiac and extracardiac anomalies.

Transthoracic echocardiogram, suprasternal notch view, demonstrating a persistent fifth aortic arch. A double-lumen transverse aortic arch is visualized, with an accessory channel coursing parallel and inferior to the native arch. Color Doppler confirms laminar, unobstructed flow through both channels.

Discussion

Persistent fifth aortic arch (PFAA) is a rare vascular anomaly characterized by persistence of an additional embryologic arch segment, resulting in a double-lumen transverse aorta. Early reports described the presence of a parallel vascular channel beneath the true aortic arch and established its distinction from other arch abnormalities [1]. More recent literature reviews confirm the rarity of the condition, describe classifications, and note possible associations [2].

PFAA is most commonly classified into systemic-to-systemic and systemic-to-pulmonary variants [2]. The systemic-to-systemic form, as observed in both patients described here, presents as a parallel channel connecting the ascending and descending aorta and may remain hemodynamically insignificant when unobstructed [2]. Recognition of this anatomy is important to avoid misclassification because other conditions may require surgical intervention or more thorough surveillance.

Modern imaging studies emphasize the role of echocardiography and cross-sectional imaging in confirming the diagnosis and defining arch anatomy [6]. In both infants described, cross-sectional imaging confirmed a widely patent systemic-to-systemic PFAA without obstruction. The anomaly did not require intervention but was identified during evaluation of intracardiac defects.

In both patients, persistent fifth aortic arch was identified during evaluation of congenital abnormalities involving multiple organ systems. While PFAA has been reported in association with congenital heart defects [2, 5], most published reports focus primarily on intracardiac anatomy or surgical management. The presence of additional congenital anomalies in these patients highlights the importance of comprehensive evaluation in infants with complex developmental findings.

Both patients demonstrated extracardiac congenital anomalies, including craniosynostosis in one and structural central nervous system and genitourinary findings in the other. Although PFAA has been reported alongside congenital heart defects [2, 5], documentation of concurrent multisystem congenital anomalies remains limited. These cases contribute additional clinical descriptions of systemic-to-systemic PFAA identified during early cardiac evaluation in infants with additional congenital

abnormalities. These cases also help to guide clinicians to consider PFAA when dealing with infants with multiple developmental findings. Although in these cases neither patient required surgical intervention for PFAA and remain asymptomatic, this report provides insight into the rare condition and how the presentation may differ on a patient-to-patient basis.

Conclusion

Persistent fifth aortic arch is an uncommon vascular variant that may be detected during assessment of congenital heart disease. In these two infants, the anomaly was non-obstructive and required no intervention but occurred in the setting of additional congenital abnormalities. Careful imaging and anatomic characterization support accurate diagnosis and appropriate longitudinal follow-up.

Declarations

Author Contributions: R.S. contributed to conception, data acquisition, and drafting of the manuscript. R.E. provided clinical care, acquired imaging data, and critically reviewed and revised the manuscript for important intellectual content. Both authors approved the final version for submission.

Ethics approval: This case report was conducted in accordance with the ethical standards of the institutional research committee and with the 1964 Declaration of Helsinki and its later amendments. Formal IRB approval was not required as this is a case report. All patient data were fully anonymized and no identifying information is included.

Consent to participate: Verbal informed consent was obtained from the legal guardians of both patients.

Consent to publish: Verbal informed consent was obtained from the legal guardians of both patients prior to publication. All data have been fully anonymized.

Competing interests: The authors have no relevant financial or non-financial interests to disclose.

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Data availability: The data that support the findings of this study are available from the corresponding author upon reasonable request, subject to privacy/ethical restrictions.

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Figures

Figure 1

CTA of Systemic-to-Systemic Persistent Fifth Aortic Arch

Contrast-enhanced computed tomography angiography (CTA), sagittal reconstruction, demonstrating a systemic-to-systemic persistent fifth aortic arch. A parallel accessory channel courses inferior to the

native aortic arch, creating a double-barreled configuration. Both lumens are widely patent without evidence of coarctation.

Figure 2

Echocardiographic Features of Persistent Fifth Aortic Arch

Transthoracic echocardiogram, suprasternal notch view, demonstrating a persistent fifth aortic arch. A double-lumen transverse aortic arch is visualized, with an accessory channel coursing parallel and inferior to the native arch. Color Doppler confirms laminar, unobstructed flow through both channels.