Radiology Case Reports 20 (2025) 3179–3185

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Case Report

Right aortic arch with aberrant left subclavian artery and anomalous Circle of Willis supply ☆,☆☆

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ARTICLE INFO

Article history:
Received 18 January 2025
Revised 12 March 2025
Accepted 13 March 2025

Keywords:
Aberrant left subclavian artery
Cerebrovascular disorders
Circle of Willis variants
Kommerell diverticulum
Right aortic arch

ABSTRACT

Right aortic arch (RAA) anomalies are rare congenital vascular malformations with a prevalence of 0.01% to 0.1% in the general population. These anomalies are often associated with congenital cardiac abnormalities, though many individuals remain asymptomatic and are diagnosed incidentally. However, cerebrovascular anomalies associated with RAA have not been previously reported. We present the case of a 23-year-old male with an incidental finding of a Type II RAA variant characterized by an aberrant left subclavian artery and an unusual, complex configuration of the Circle of Willis (CoW). This CoW anomaly includes an absent right internal carotid artery, an absent left vertebral artery, and unique collateralization pathways. Despite the variant anatomy, the patient has remained neurologically asymptomatic. No underlying genetic abnormalities were detected during further investigation with a chromosomal analysis. This case highlights the novel association between RAA and anomalies of the CoW, emphasizing the importance of detailed cerebrovascular imaging in patients with RAA. Since an anomalous CoW is inherently associated with an increased risk of cerebrovascular accidents and poorer outcomes, information on cerebrovascular anatomy can facilitate proactive risk factor monitoring, and is valuable for surgical/neurointerventional planning when indicated.

Introduction

Right aortic arches (RAAs) are rare anatomical anomalies occurring in 0.01%-0.1% of the population; they result from abnormal organogenesis of the primitive aortic arches during fetal development [1]. RAAs can be associated with a range of congenital heart defects, such as truncus arteriosus, tetralogy of Fallot, and tricuspid atresia, as well as chromosomal abnormalities, including DiGeorge syndrome [2]. Despite these

☆ Competing Interests: The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

☆☆ Acknowledgements: The authors would like to express their gratitude to the patient and his family for his time and commitments involved in this project. We also would like to thank Blacktown Hospital Medical Imaging Department for their contributions in this interesting case.

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https://doi.org/10.1016/j.radcr.2025.03.049

associations, most RAAs are diagnosed incidentally, as most patients remain asymptomatic [1]. In rare instances, certain RAA variants can lead to tracheal and/or esophageal compression by forming vascular rings encircling these mediastinal structures [2]. However, to our knowledge, cerebrovascular anomalies associated with RAAs have not been previously reported. In this report, we present the case of a patient with an RAA variant associated with an anomalous Circle of Willis (CoW) anatomy. Such an association should prompt consideration of detailed cerebrovascular imaging in such cases to evaluate potential risks and guide clinical management.

Case presentation

We present the case of a 23-year-old male of Peruvian descent with several anatomical anomalies diagnosed during childhood. These included polydactyly (an extra first digit on the left hand), agenesis of the left kidney, and a large ventricular septal defect - which led to the development of Eisenmenger syndrome with pulmonary hypertension. Over his childhood, he was evaluated for a potential heart transplant; however, his condition stabilized during adolescence, and transplant surgery was indefinitely deferred in favor of medical therapy to manage his pulmonary hypertension. His only other significant medical history included isolated thrombocytopenia. In terms of medications, the patient was on Macitentan (an endothelin receptor antagonist) and tadalafil (a PDE5 inhibitor) to manage pulmonary hypertension, with nil other regular medications. The remainder of his developmental history was unremarkable, with all developmental milestones achieved appropriately. He completed his education up to Year 11 in the Australian public school system.

The patient came to clinical attention when he presented to the emergency department with left-sided facial numbness, left ear pain, and a sensation of ear fullness. A comprehensive stroke workup was attempted, starting with a routine CT brain with Arch-CoW angiography and perfusion studies. Images were captured using a Phillips 64-Slice CT Brilliance scanner. His CT brain and angiography were completed successfully; however, the perfusion studies were abandoned due to significant patient motion artefacts. Despite this, his CT angiography demonstrated patent intracranial vasculature, preserved distal flow and no evidence of asymmetry. Further neuro-imaging was performed with an MRI-brain diffusion weighted imaging (DWI) sequence using a 3T MAGNETOM PRIMSA FIT MRI scanner. This similarly did not display evidence of acute stroke or ischemia. However, an incidental finding of a RAA variant was identified on CT imaging, as shown in Figs. 1 and 2.

Imaging revealed the following aortic arch branching pattern (from anterior to posterior):

1. Left common carotid artery (CCA) with early bifurcation.
2. Right external carotid artery (ECA), with absent right common and right internal carotid arteries.
3. Right subclavian artery (giving rise to a dominant right vertebral artery).
4. Left subclavian artery (arising from the descending aorta via a Kommerell diverticulum, and taking a retro-esophageal/retro-tracheal course, with absent left vertebral artery).

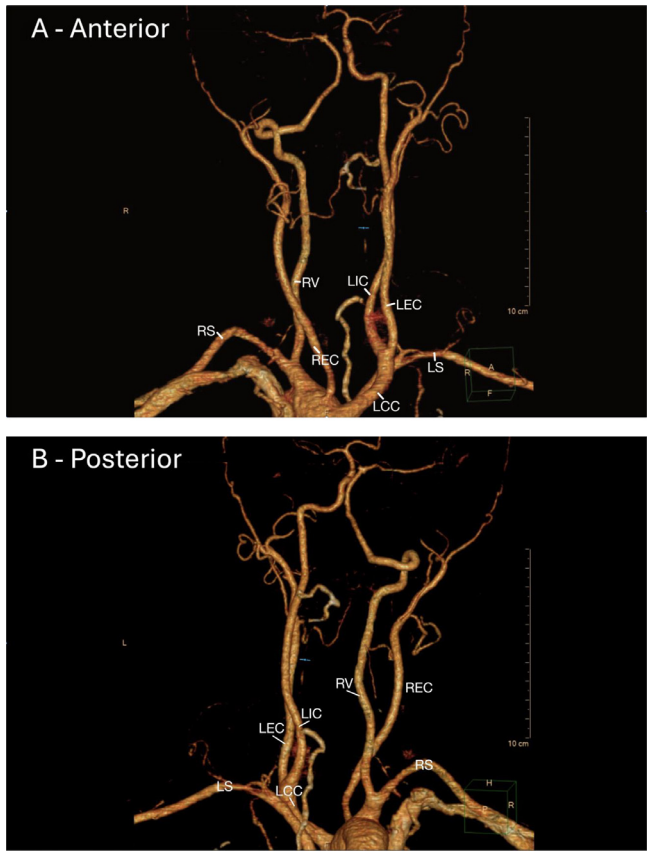

Fig. 1 – 3D anterior and posterior reconstructed views generated from CT angiography Arch-CoW. (A) Anterior view. (B) Posterior view. Notably, the patient's right internal carotid and left vertebral arteries are absent. LS = left subclavian artery, LCC = left common carotid artery, LEC = left external carotid artery, LIC = left internal carotid artery, RV = right vertebral artery, REC = right external carotid artery, RS = right subclavian artery.

Further assessment demonstrated additional unique anatomical variants in the CoW, as show in in Fig. 3:

- The right vertebral artery is dominant and the left vertebral artery is absent.
- The right ICA and A1 segment of the right anterior cerebral artery (ACA) is absent.
- The right middle cerebral artery (MCA) is supplied via a large right posterior communicating artery (PCOMA). The left PCOMA is absent.
- Both ACAs are supplied via the left ICA and anterior communicating artery (ACOMA).

Other findings included a right cervical rib partially fused with the right first rib, and a dilated pulmonary trunk (consistent with the patient's known history of pulmonary hypertension). The patient was discharged from the emergency department with a diagnosis of otitis media. However, due to the unique anatomical findings, his case was discussed at our

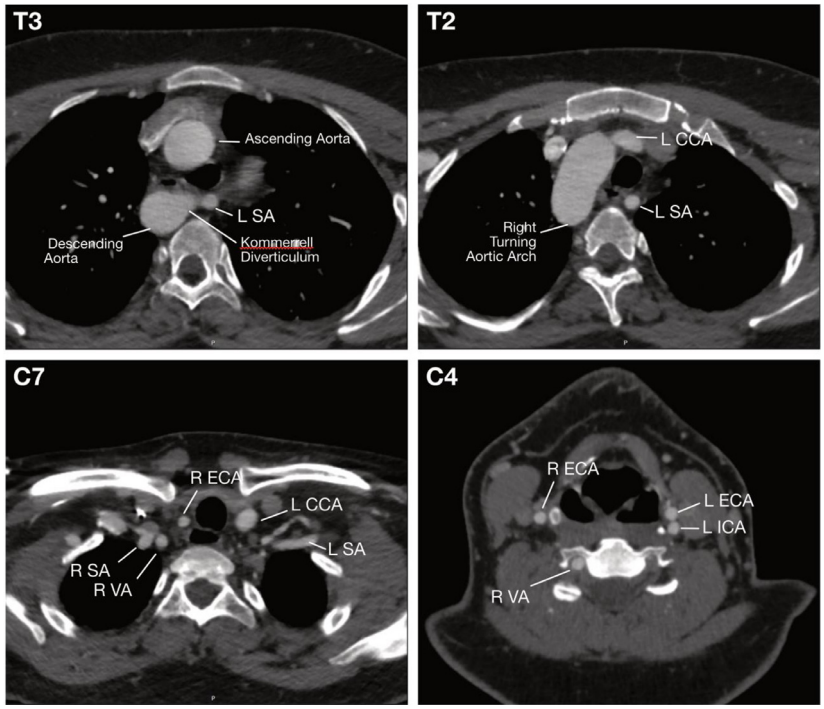

Fig. 2 – Serial axial CT angiography Arch-CoW slices labelled at corresponding vertebral levels (vascular window settings). L SA = left subclavian artery, L CCA = left common carotid artery, R SA = right subclavian artery, R VA = right vertebral artery, L ECA = left external carotid artery, L ICA = left internal carotid artery.

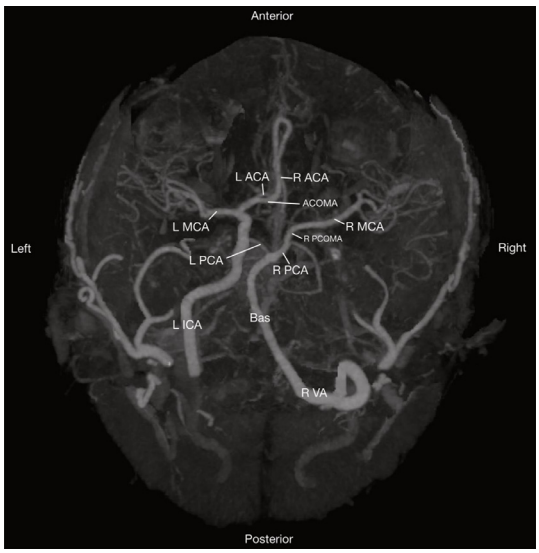

Fig. 3 – CT angiography 3D maximum intensity projection of Circle of Willis vessels. A dorsal/cranial view of the brain is shown (anterior at the top of the figure, posterior at the bottom of the figure). L ICA = left internal carotid artery, R VA = right vertebral artery, Bas = basilar artery, R PCA = right posterior cerebral artery, L PCA = left posterior cerebral artery, L MCA = left middle cerebral artery, R MCA = right middle cerebral artery, R PCOMA = right posterior communicating artery, ACOMA = anterior communicating artery, L ACA = left anterior cerebral artery, R ACA = right anterior cerebral artery.

weekly departmental neuroradiology meeting (where interesting and complex cases are reviewed between available neurology and radiology consultants for consensus opinion and educational purposes).

Given an RAA and multiple other anatomical variants, a chromosomal microarray was requested to investigate potential underlying chromosomal abnormalities, such as DiGeorge syndrome. DiGeorge syndrome was considered because of its association with RAAs, congenital cardiac anomalies, and an increased risk of neurological complications, including cognitive impairment and structural brain anomalies [2]. The chromosomal microarray returned normal, and there was no family history of consanguinity.

On follow-up with these tests, the patient continued to exhibit normal neurological and cognitive function. His neurologic symptoms did not recur since his initial presentation. He was informed of his genetic and neurovascular findings, and was recommended for monitoring by his cardiology and respiratory physicians to ensure ongoing surveillance and risk factor optimization. Overall, it was concluded that his previous symptoms were likely unrelated to these incidental anatomical variants of his vasculature.

Discussion

Aortic arch formation variations can be explained using Edward's hypothetical double aortic arch model, described in his original 1948 paper [3]. This model suggests that during normal fetal development, a single aortic stem arises from the left

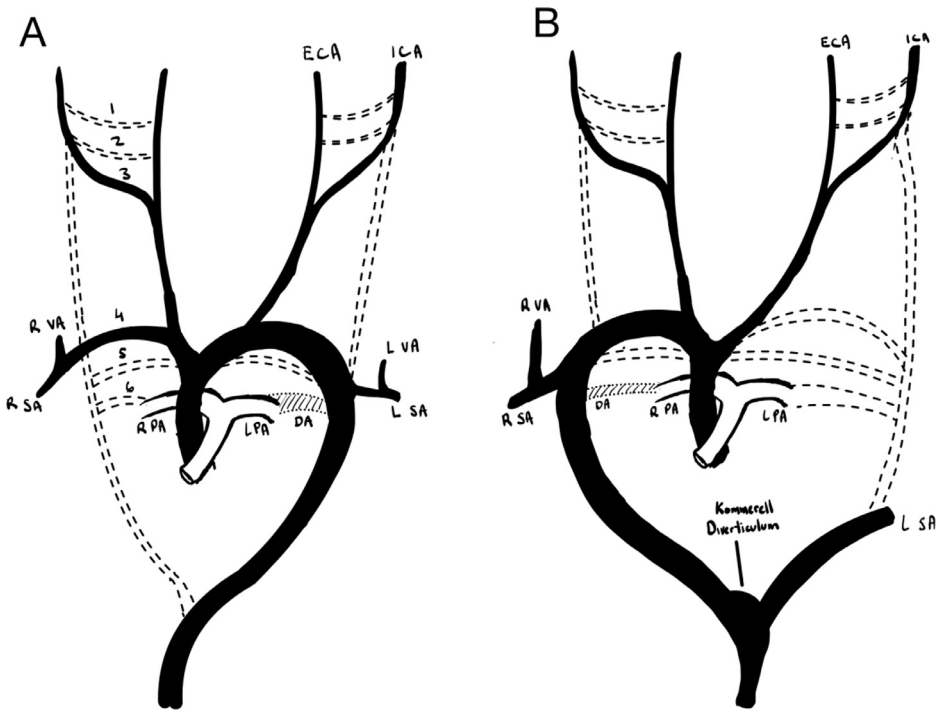

Fig. 4 – (A) Conventional left sided aortic arch. Dashed lines show regressed arches/derivatives. Hatched line shows ductus arteriosus. (B) Right sided aortic arch with aberrant left subclavian artery arising from a left aortic arch remnant (i.e. a Kommerell diverticulum). R SA = right subclavian artery, L SA = left subclavian artery, ECA = external carotid artery, ICA = internal carotid artery, DA = ductus arteriosus, R VA= right vertebral artery, L VA = left vertebral artery, R PA = right pulmonary artery, L PA = L pulmonary artery. Adapted from Fekadu et al. (2024) [5].

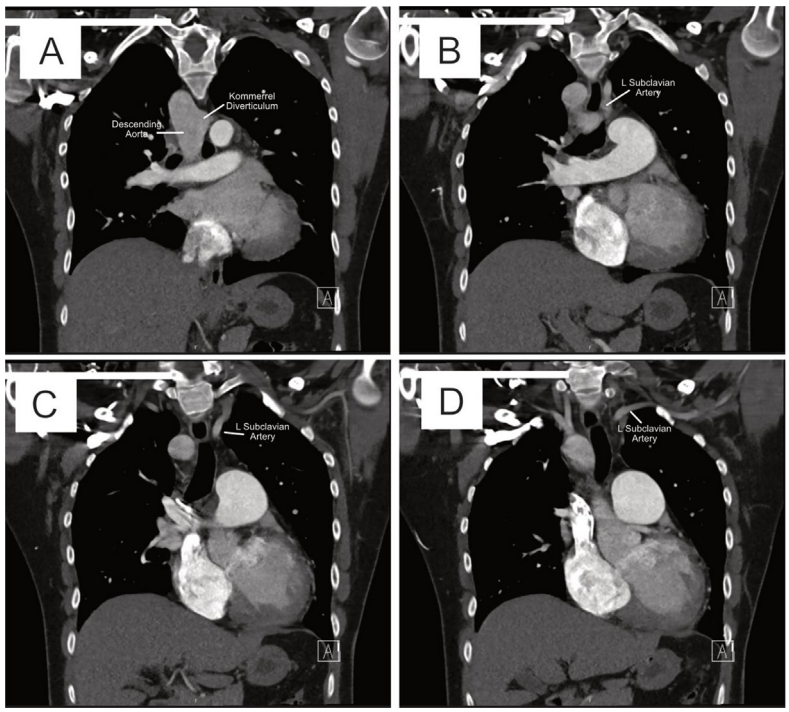

Fig. 5 – Coronal slices of CT pulmonary angiography, outlining the Kommerell diverticulum and aberrant course of the left subclavian artery. Slices are displayed from posterior (A) to anterior (D). Note the left subclavian takes a retro-tracheal course, as seen in (B/C).

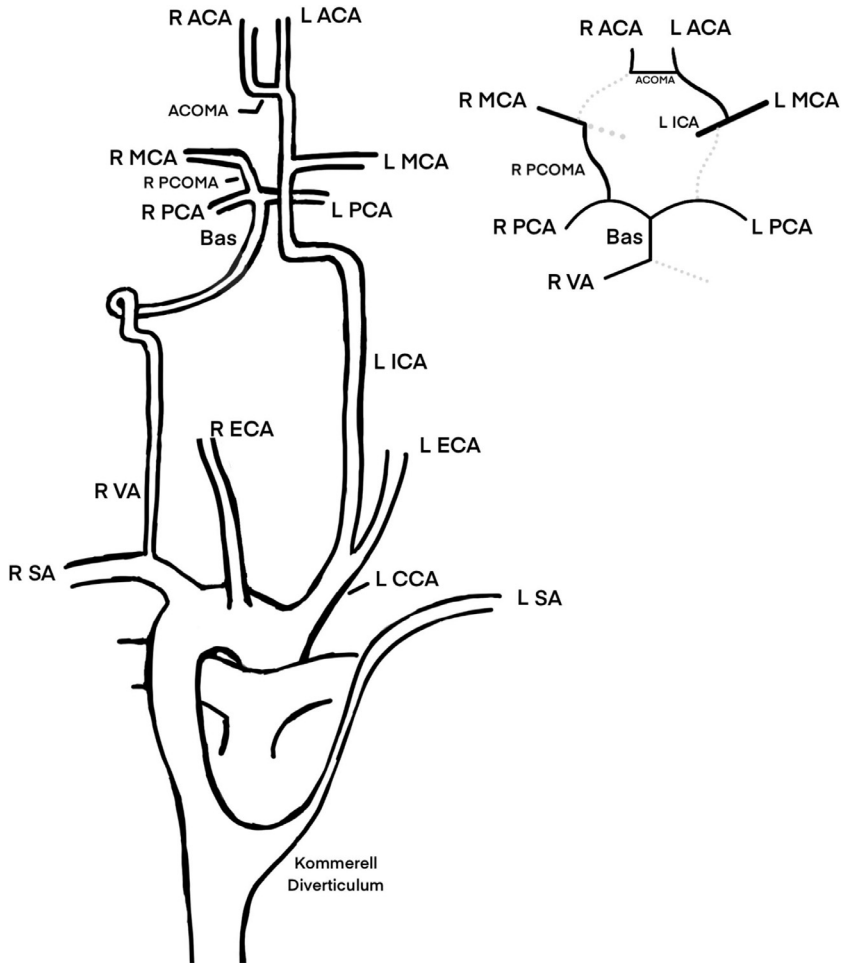

Fig. 6 – Diagrammatic sketch of the patient's vascular abnormalities from the arch of the aorta to CoW. A CoW schematic (top right) is also included to demonstrate missing vessels from conventional CoW anatomy (represented by grey dashed lines). L SA = left subclavian artery, R SA = right subclavian artery, L CCA = left common carotid artery, L ECA = left external carotid artery, L ICA = left internal carotid artery, R ECA = right external carotid artery, R VA = right vertebral artery, Bas = basillary artery, L PCA = left posterior cerebral artery, R PCA = right posterior cerebral artery, R PCOMA = right posterior communicating artery, R MCA = right middle cerebral artery, L MCA = left middle cerebral artery, L ACA = left anterior cerebral artery, R ACA = right anterior cerebral artery, ACOMA = anterior communicating artery.

ventricle and divides into right and left aortic arches, which curve and descend around their respective primary bronchi to join and form a single descending aorta at the vertebral column [3]. Each aortic arch gives rise to its respective carotid and subclavian arteries, as well as ductus arteriosus connecting to its corresponding pulmonary artery. A conventional left-sided aortic arch anatomy forms when the right aortic arch and right ductus arteriosus regress, leaving the right innominate artery as the first branch of the aorta. Different regression patterns can lead to anatomical variations of the aorta. In such instances, the upper portion of the descending aorta will typically be on the same side as the ductus arteriosus (i.e., a right-turning aorta will usually have a ductus arteriosus connecting to the right pulmonary artery, instead of the left) [3].

Right aortic arches (RAAs) are classified into 3 primary subtypes based on their branching patterns [2,4]:

Type I: Mirror image branching of the head and neck vessels. The first branch is the left innominate artery, followed by the right common carotid artery and the right subclavian artery. Type I RAA is the most common subtype (50% prevalence) and is strongly associated with congenital heart defects, such as truncus arteriosus, tetralogy of Fallot, tricuspid atresia, and DiGeorge syndrome. If isolated and asymptomatic, it typically does not require treatment [2,4].

Type II: RAA with an aberrant left subclavian artery arising from a diverticulum of Kommerell (a remnant of the left dorsal aortic root) that takes a retro-esophageal course. The first branch is the left CCA, followed by the right CCA, right subclavian artery, and left subclavian artery (Fig. 4). This variant is the second most common, accounting for 40% of cases. Together with the ascending aorta, the Kommerell diverticulum, and the ligamentum arteriosum, this configuration can form a complete vascular ring around the trachea and esophagus, potentially causing respiratory and gastrointestinal symptoms. It is rarely associated with cardiac malformations [2,4].

Type III: RAA with an isolated left subclavian artery arising from the left vertebral artery or ductus arteriosus. This rare variant (1.5% prevalence) may present with subclavian steal syndrome [4].

Comparison to a previous CT pulmonary angiography study (done the previous year for work-up of pulmonary hypertension) clearly demonstrated an aberrant left subclavian artery originating from a Kommerell diverticulum (Fig. 5). Taken together with his aortic branching pattern, his case best matched a Type II RAA. However, this presentation was atypical due to the presence of a ventricular septal defect, as Type II RAAs are generally not associated with congenital cardiac abnormalities [4]. Chromosomal microarray analysis was performed due to the association between RAAs and rare genetic syndromes, such as DiGeorge syndrome, which carry risks of cognitive and neurological complications. In our patient, no chromosomal abnormalities were identified. This aligns with data from D'Antonio et al. (2016), who reported chromosomal abnormalities in only 14.6% of patients with RAAs, and DiGeorge syndrome occurring in 6.4% of cases [1].

Furthermore, this case was associated with anatomical variants of the Circle of Willis (CoW), which is summarized in a diagrammatic sketch in Fig. 6. To our knowledge, this is the first reported case of an RAA pattern coexisting with CoW anomalies. Using Lie and Hage's classification for CoW collateralization in ICA agenesis [6], his case is best described by a Type A variant of ICA agenesis, where circulation to the ipsilateral ACA is compensated by a patent ACOMA, and flow to the ipsilateral MCA is supplied via a hypertrophied a PCOMA. His CoW arrangement can also be summarized by a recent coding system proposed by Ayre et al. (2021) [7] for CoW variants, with the code $A1^{A}$-$^{C}PCOMA^{A}$ (an absent A1 segment of the ACA, along with absence of the contralateral PCOMA). Uniquely, our case coexists with a lack of a left vertebral artery, which is not described in either classification system.

Variations in CoW anatomy have been associated with significant clinical implications. Incomplete CoW configurations are associated with an increased burden of CNS white matter disease [8–11], a greater risk of intraoperative ischemic events during endarterectomy [8], a higher incidence of ischemic stroke [12], and poorer stroke outcomes [13]. Vertebral artery hypoplasia or agenesis further elevates the risk of posterior circulation stroke, especially when combined with atherosclerosis or prothrombotic factors [14]. Similarly, agenesis or severe hypoplasia of the internal carotid artery has an increased incidence of intracranial aneurysms (25%-67%, compared to the baseline population risk of 2%-4%) [15], likely due to altered hemodynamic flow or congenital defects in the cerebral arterial wall [15–19]. A recent 2021 literature review by Takiyama et al. found that the majority (54%) of aneurysms in Type A ICA agenesis were found in the ACOMA [19], however none were specifically identified in our patient.

Conclusion

RAAs are rare embryologic anomalies with various configurations, most of which are identified incidentally [1]. This case report highlights a novel association between RAA and abnormalities of the CoW, which are identified to have significant neurovascular risks, such as ischemic stroke, intracranial aneurysms, and poorer neurological outcomes [8–13]. Our findings suggest that patients with major aortic arch anomalies, such as RAA, should undergo a thorough evaluation of their primary aortic branches and cerebrovascular structures, including the CoW, to facilitate early identification of abnormalities, enable tailored cardiovascular risk stratification and guide long-term optimization of stroke prevention strategies. Furthermore, such imaging also provides critical information for neurovascular surgical planning to improve outcomes in this unique patient population.

Patient consent

Our patient provided written and informed consent for the publication of their case and relevant imaging findings included in the manuscript.

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