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Rare case of adult truncus arteriosus from the Vernon-Roberts Museum of The University of Adelaide (Australia)

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Abstract. Persistent truncus arteriosus (TA) is a congenital heart disorder that is characterized by various anomalies that can be fatal without corrective medical intervention. Surgical techniques were initially developed to correct this condition in infants, consequently, this has led to a paucity of scientific research on adults with TA. In this case study, the authors examine TA in the heart of an adult. The specimen derives from the pathology museum from the Vernon-Roberts Museum of The University of Adelaide. Given the rarity of individuals surviving with TA into adulthood, more scientific attention needs to be invested in unique people with TA in order to provide a comprehensive understanding of this condition as well as adequate medical treatment and care.

Keywords: truncus arteriosus, congenital disorder, Australia, clinical cardiology, pathology museum.

ANATOMICAL AND CLINICAL INTRODUCTION

Truncus arteriosus (TA), also known as persistent truncus arteriosus is a rare congenital heart abnormality. Truncus arteriosus is a single arterial trunk exiting the heart (by way of one semilunar valve) to supply systemic, pulmonary, and coronary arteries (Abbott 1936, Anderson et al. 1957, Sharma et al. 1985, Abid et al. 2015, Martin et al. 2016). Other anomalies are occasionally associated with TA such as pulmonary arterial hypoplasia, interrupted aortic arch (IAA), ventricular septal defect (VSD) and quadricuspid aortic valves (Martin et al. 2016, Alamri et al. 2020). Although, preva-

lence of TA varies between studies, it can safely be noted that it accounts for <4% of congenital heart disorders (Hoffman 2011, Martin et al. 2016, Nabati 2017, Safi 2018, Alamri et al. 2020).

TA is typically detected during the neonatal period or early childhood, where symptoms of pulmonary hyper-circulation induced congestive heart failure and cyanosis are evident (Martin de Miguel et al. 2022). It has been estimated that 34% of patients with TA have DiGeorge Syndrome (22q11 deletion) (Lupski et al. 1991, Laohaprasitiporn et al. 2008). Furthermore, diabetic mothers have a 12 to 13-fold risk of having an infant developing TA compared to their non-diabetic equivalents (Ferencz et al. 1997).

In 1798, Wilson produced the first account of TA (Wilson 1798, Mavroudis et al. 2015). Later Buchanan (1864) presented an examination of undivided TA in a six-month-old infant, while Shapiro (1930), and Lev and Saphir (1942) made important contributions to defining and classifying TA. Although the surgical technique of pulmonary artery banding (PAB) was conceived by Muller and Dammann in 1952, pulmonary artery banding was only introduced in the 1960s with substandard outcomes in infants (Muller and Dammann 1952, Craig and Sirak 1963, Williams et al. 1963, Goldblatt et al. 1965, Mavroudis et al. 2015). However, it was not until 1968, when correction of truncus arteriosus abnormality was achieved by using a homograft of the ascending aorta, that also included a heart valve. This technique established a new outflow from the right ventricle to the pulmonary trunk (Dwight et al. 1968).

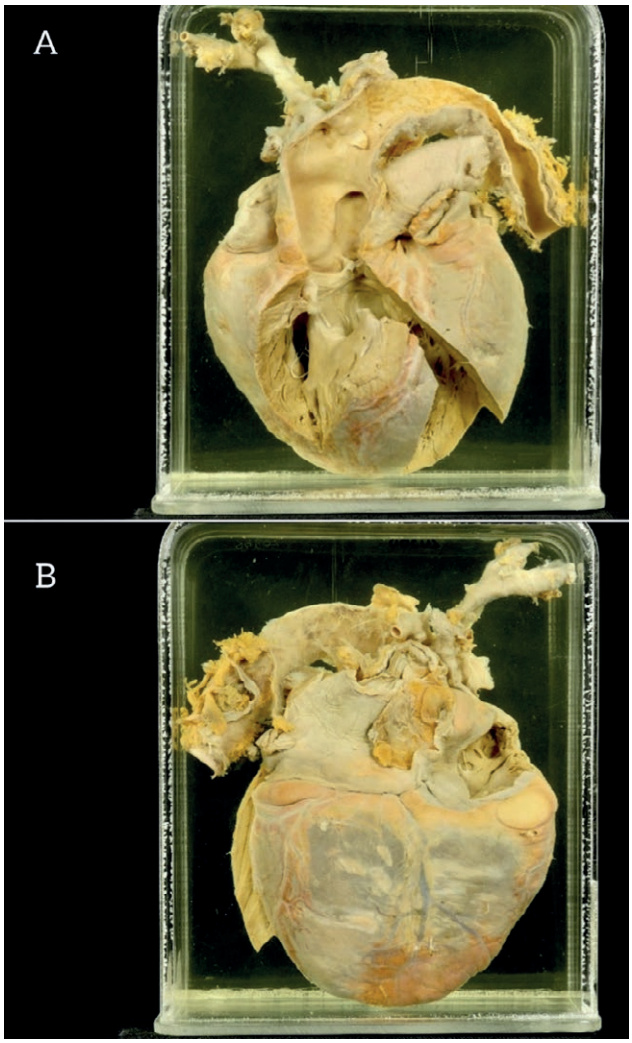

Figure 1. Frontal (a) and posterior (b) view of the heart with evidence of the persistent truncus arteriosus.

MATERIALS AND METHODS

In this case study, the authors examine TA in the heart of an adult individual. The specimen (Fig. 1) derives from the Vernon-Roberts Museum, The University of Adelaide. Given the rarity of individuals surviving with TA into early and middle adulthood, it is useful to describe this case from a morphological perspective to provide a comprehensive understanding of this condition and adequate medical treatment and care.

CASE DESCRIPTION

According to the information supplied by the Vernon-Roberts Museum, The University of Adelaide, the heart specimen is derived from a 45-year-old female. Patient anonymity was maintained. No mention was provided of the decedent's ethnicity, any other morbidities or other personal details. At the age of 35 years the decedent had been diagnosed with truncus arteriosus. The decedent had died from a cardiac arrest on the 6th day after admission to a hospital. Her clinical diagnosis had revealed tachycardia, polycythaemia (erythrocytosis), atrial fibrillation and right/left ventricular failure. The right ventricular wall was hypertrophic. There was a 25 cm atrioventricular septal defect, as well as an overriding single great vessel and a posteriorly arising pulmonary artery. At death the decedent's heart was grossly enlarged and weighed 680 grams.

Although TA prevalence is very low, its signature hemodynamic pathology requires immediate medical intervention. Without corrective surgical treatment, mortality occurs in ≈ 80-90% of neonates in the first year of life (Ebert et al. 1984, Thompson et al. 2001, Kharwar

et al. 2014, Nabati 2017, Alamri et al. 2020). However, even after successful surgical intervention, there is an increased need for long-term cardiac management due to possible continuing complications (Chen et al. 2016, Puri et al. 2017, Bhansali and Phoon 2023). High mortality associated with TA is often due to it causing hypertensive pulmonary arterial pressure with subsequent onset of pulmonary disease (Sharma et al. 1985). Even with such a high mortality rate, there is a small percentage of individuals who can live with TA up until early middle age (i.e. 45-50 or 60-65 years). Unfortunately, there has been comparatively little scientific research on adults with TA. One reason for this could be persistent medical focus on pediatric cases. Second, there have been few individuals who have been able to live with TA beyond early middle age (Porter and Vacek 2008). Therefore, it is unfeasible to systematically monitor and examine this sparse group. Additionally, since it is unclear how individuals managed to survive with TA into adulthood, both outward reach and treatments are difficult.

CONCLUSION

This case shows the importance of anatomical and pathological collections both for educational and research purposes in the fields of biomedicine and anthropology, as previously stressed both in the context of anatomopathological museum studies and the assessment of cardiovascular diseases in the past (Nesi et al. 2007, Henriques de Gouveia et al. 2021). Future research on such specimens could be aimed at combining morphological and clinical data with radiological and genetic investigations capable of offering a comprehensive phenotypic and genotypic background on a pathology still of great relevance for clinical medicine.

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