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Case Report

Anatomical Variations of the Heart and Chest Wall in a Donor Patient with Apert Syndrome

Andrew E. Cox, OMS-III, Edith L. Sperling, PT, DPT, Julia P. Fischer, OMS-III, Whitley F. Nelson, OMS-III, Fengxin Li, OMS-III, Abraham P. Campbell, OMS-IV

Western University of Health Sciences, College of Osteopathic Medicine of the Pacific Northwest, 200 Mullins Dr. Lebanon

Corresponding author: Edith L. Sperling, Western University of Health Sciences, College of Osteopathic Medicine of the Pacific Northwest, 200 Mullins Dr. Lebanon, OR 97355, Tel: 541-259-0229; Email: esperling@westernu.edu

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Abstract

Apert Syndrome is a rare genetic disorder that often manifests as distinct anatomic variations of the skull, hands and feet. Mutations in the Fibroblast growth factor 2 gene (FGFR2) are known to contribute to the disease progression and anatomic variations seen in those with Apert Syndrome. Of these anatomic variations, the most commonly seen and well documented variations are those of the skull bones, hands and feet, specifically fusion of the fingers and toes. Less is known about the anatomic variations of the heart and chest wall in patients with Apert Syndrome and this is the first known study to investigate this region [1]. This article discusses the anatomic variations of the heart and chest wall discovered during anatomical dissection of one donor patient (cadaver) with Apert Syndrome. Specific attention is paid to the anatomic variations of an elongated ligamentum arteriosum, atypical venous drainage of the heart, double internal thoracic artery bilaterally, and abnormal muscle fibers of the pectoralis minor muscle.

Keywords: Thorax; Anatomy; Abnormal; Atypical; Ligamentum arteriosum: Internal thoracic artery; Cadaver; Dissection; Pectoralis minor; Levator costarum pectoralis

Introduction

Apert Syndrome is a rare genetic disorder which was first described clinically in 1906. Mutations in the Fibroblast growth factor 2 gene (FGFR2) are known to contribute to the disease progression and anatomic variations seen in those with Apert Syndrome, which is one of the most severe forms of the craniosynostosis syndromes [2].

There are eight disorders comprising the FGFR-related craniosynostosis spectrum: Pfeiffer syndrome, Apert syndrome, Crouzon syndrome, Beare-Stevenson syndrome, FGFR2-related isolated coronal synostosis, Jackson-Weiss syndrome, Crouzon syndrome with acanthosis nigricans, and Muenke syndrome [3]. Diagnosis of Apert Syndrome is based on clinical findings. Molecular genetic testing of FGFR1, FGFR2, and FGFR3 can establish the specific diagnosis [4]. The FGFR-related craniosynostosis syndromes are inherited in an autosomal dominant manner, with a paternal age effect, and individuals have a 50% chance of passing the pathogenic variant to each child [5]. However, reproductive fitness is low, and more than 98% of cases are thought to be due to new mutation [6]. Individuals with Apert Syndrome typically require life-long medical management by a multi-disciplinary team [7].

Of the possible anatomic variations, the most common and well documented variations are those of the skull bones, hands and feet, specifically syndactyly. Much less is known about the anatomic variations of the heart and chest wall in patients with Apert Syndrome and this is the first known study to investigate this region. This article discusses the anatomic variations of the heart and chest wall discovered during anatomical dissection of one donor patient (cadaver) with Apert Syndrome. Specific attention is paid to the anatomic variations of an elongated ligamentum arteriosum, atypical venous drainage of the heart, double internal thoracic artery bilaterally, and abnormal muscle fibers of the pectoralis minor muscle[Figure 1].

Materials and Methods

This study consists of a detailed dissection of one donor patient (cadaver) with Apert Syndrome. This research was deemed Not Human Subject Research by the IRB of Western University of Health Sciences, IRB reference # X18/IRB/092. The donor patient was accepted by the Willed Body Program at Western University of Health Sciences, College of Osteopathic Medicine of the Pacific Northwest, upon donation by his family. Radiologic imaging was captured prior to gross dissection by a licensed x-ray technician at Samaritan Lebanon Community

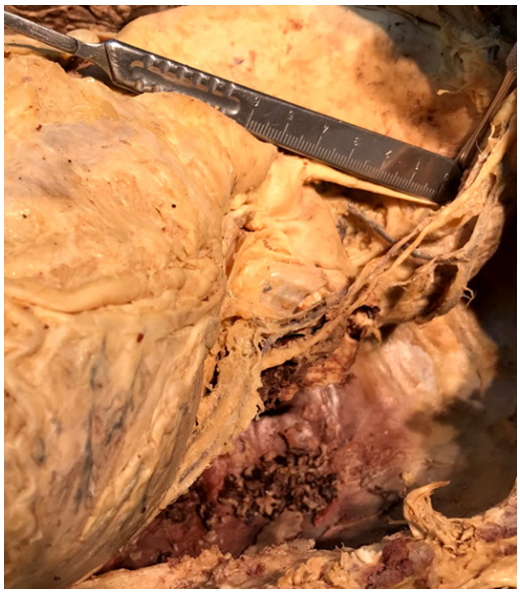

Figure 1. Ligamentum arteriosum

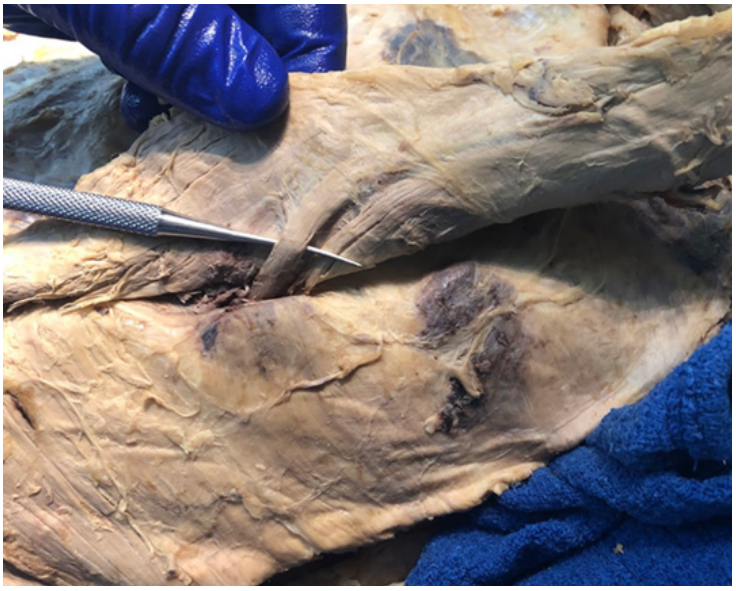

Figure 2. Levator Costarum Pectoralis

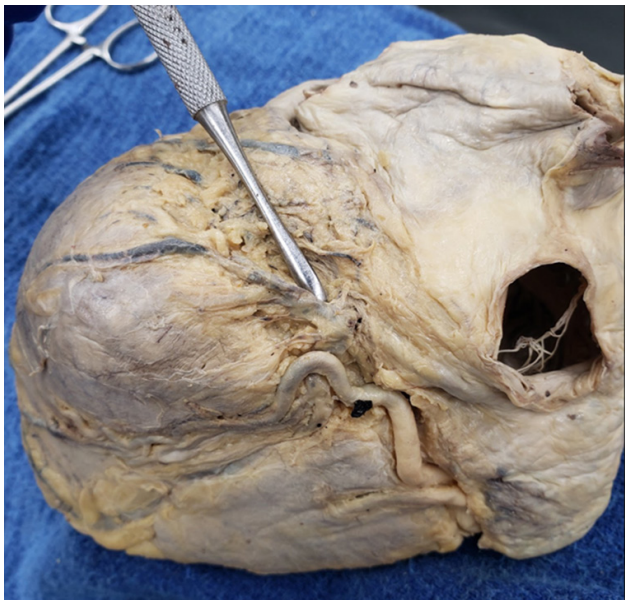

Figure 3. Coronary sinus, diminished

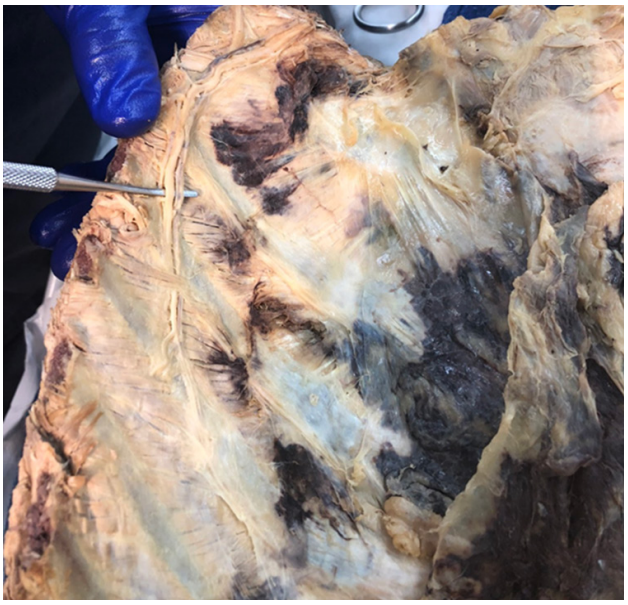

Figure 4. Duplicated internal thoracic arteries, descending roughly 4 inches lateral to internal thoracic artery

Hospital. The views captured in this region were AP and lateral thorax. After documenting abnormalities of the external chest wall, a thorough, detailed dissection of the donor patient was completed by all authors. The specific dissection described in this article was done with an emphasis on the anterior chest wall, heart, and mediastinum. Abnormalities of nerves, muscles, arteries, veins, and several other structures were elucidated and documented with photography during dissection.

Results:

1. General observation: Significant left ventricular hypertrophy is appreciated. Bilateral rib fractures and evidence of substernal hemorrhage is noted.

2. Ligamentum Arteriosum: The ligamentum arteriosum is an anatomical remnant of the ductus arteriosus, a fetal structure that maintains proper oxygenation of the blood in a developing fetus. In most patients, the ligamentum arteriosum spans a length of 15-16mm [7]. In this donor patient, the ligamentum arteriosum measured 41mm.

3. "Levator Costarum Pectoralis": Branching off of bilateral pectoralis minor muscles and inserting on the internal surface of the 4th rib was a separate group of muscle fibers, which we have termed the Levator Costarum Pectoralis [Figure 3].

4. Venous Drainage of the Heart: Venous drainage of the heart is normally accomplished primarily by the coronary sinus, a venous structure located in the posterior atrioventricular groove

and drains into the right atrium. In this donor patient, venous drainage of the heart was dominated by a left antero-lateral venous structure that split the base of the great vessels and the left atrium, draining directly into the superior vena cava [Figure 3].

5. Internal Thoracic Arteries: Typically found descending along the internal surface of the anterior thoracic wall bilaterally, adjacent to the sternum, internal thoracic arteries in this donor patient were accompanied by duplicate descending arteries approximately 4 inches lateral to the internal thoracic arteries [Figure 4].

Discussion: While the cause of death in this patient is unknown, evidence of chest compressions and life-saving efforts are apparent due multiple fractured ribs and evidence of internal bleeding in substernal areas adjacent to the heart. Significant left ventricular hypertrophy may indicate increased work of the heart brought on by an increase in afterload. In the presence of ischemic damage to the heart, the internal thoracic arteries are commonly harvested when placing a coronary artery bypass graft. A better understanding of both the venous drainage of the heart and the presence of a set of descending arteries lateral to the internal thoracic arteries may have presented an opportunity to prevent further ischemic damage to the heart through surgical intervention.

Orthopedically, the presence of bilateral levator costarum pectoralis may have influenced upper limb biomechanics, in particular the available range of motion of shoulder elevation and scapular retraction, which would be limited with a shortened pectoralis minor. It is worth noting that this donor patient had significant shoulder abnormalities outside of just these muscle fibers.

While the anatomy in patients with Apert Syndrome can be highly variable, variations discovered in this study may offer clinical insight into cardiguac function and preventative measures available for patients with Apert Syndrome. These findings are also of interest to anatomists for their contribution to anatomical knowledge for further analysis of Apert Syndrome.

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