

CASE REPORT

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Post-Mortem Echocardiography as a Guide to Cardiac Autopsy—A Worthwhile Concept?

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Abstract: Sudden and unexpected death in the young is a common and worldwide problem. Sudden, unexpected death in infancy (SUDI), clinically unexpected death in an infant between one week and one year of age, affects around 1 in 1000 infants. Autopsy will reveal a specific cause of death in only one third of cases. This has led to various ancillary examinations in an effort to increase the diagnostic yield of the autopsy.

In this case report it is suggested that another diagnostic modality, that of the post-mortem echocardiogram might be a worthwhile concept to explore.

Keywords: autopsy, echocardiogram, SUDI

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Introduction

Every year thousands of infants, children, adolescents and young adults die suddenly and unexpectedly.¹ Of these a significant proportion are autopsy negative and are classified as autopsy negative sudden, unexplained death.¹ Sudden unexpected death in infancy (SUDI) is defined as clinically unexpected death in infants from one week to one year of age.² SUDI affects around 1 in 1000 infants, with a peak prevalence at three months of age.²

Detailed autopsy examinations, including various ancillary examinations reveal a specific cause of death in only about one third of cases.^{2,3} These ancillary examinations include radiological skeletal surveys to identify rib fractures,⁴ bacteriological examination,^{5,6} post-mortem genetic testing to identify the subset of channelopathic SUDI, known as the cardiac channel molecular autopsy,¹ as well as DNA and RNA extraction from a variety of tissues to detect serotonin transporter gene polymorphisms.⁷

Various cardiovascular abnormalities, isolated or associated with central nervous system alterations have been observed in cases of SUDI.⁵ These include: accessory atrioventricular pathways (mostly Mahaim fibres), hypoplasia of the cardiac conduction system or the central fibrous body, splitting of the atrioventricular node or the His bundle and a Zahn node.⁵

In this proof of concept study we aimed to explore the feasibility of post-mortem echocardiography in order to alert the pathologist to any possible underlying cardiovascular anomalies before the actual dissection of the heart, in order to avoid any possible damage to underlying delicate structures.

Materials and Methods

This proof of concept study was undertaken with approval of the ethical committee of the Faculty of Health Sciences, University of Pretoria.

The heart of a three month old male infant, whose death fulfilled the criteria for SUDI was used. At autopsy, the heart was removed, irrigated and filled with buffered formalin. The major vessels were tied off with string, in order to maintain the filling of the left ventricle with formalin to facilitate the subsequent echocardiogram.

Echocardiography was performed with a Philips Envisor C echocardiography system. The heart was dissected afterwards.

Results

Echocardiography revealed the presence of a sub-aortic muscular band (see Figure 1). The muscular tendon is marked with +. The left ventricle was then dissected by cutting it open from the lateral aspect in order to prevent any damage to the observed muscular band. The presence of a thick muscular band was confirmed macroscopically (see Figure 2).

Discussion

Autopsy has a very low yield for a specific diagnosis in cases of SUDI.^{2,3} Weber et al performed the largest single-institution autopsy study of SUDI.³ They analyzed 1516 paediatric post-mortem examinations and found 546 SUDI cases. Death could be explained in 37% of these cases by the autopsy findings implying that 63% of SUDI cases remained unexplained. The authors suggest that alternative and/or additional diagnostic techniques are needed to improve the detection rate of an identifiable cause of death at autopsy in an attempt to lower the high number of unexplained SUDI cases.

An intriguing new concept is the so-called “molecular autopsy” where post-mortem genetic analysis in cases of SUDI explores the prevalence of channelopathies as the pathogenic basis for sudden unexplained death in infants.⁸

We propose that post-mortem echocardiography is an additional concept worth exploring. This single case report merits a larger study to determine whether post-mortem echocardiography can ultimately guide the cardiac dissection method in order to preserve delicate underlying cardiac anomalous structures

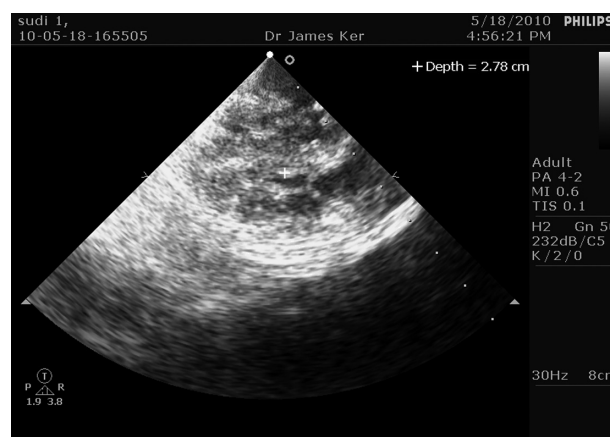


Figure 1. Post-mortem echocardiogram. Note the subaortic muscular band marked with +.

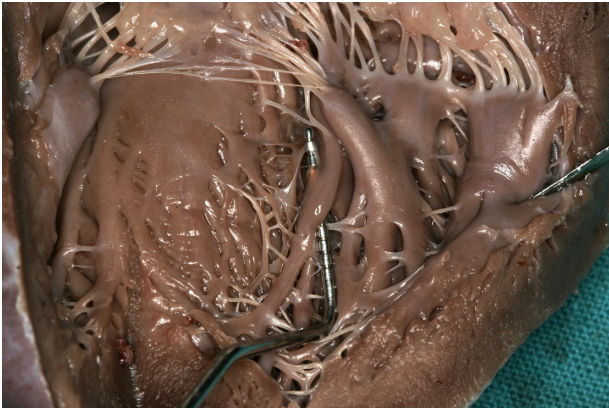


Figure 2. The dissected left ventricle. Note the thick muscular band.

which may ultimately turn out to play a causal role in cases of SUDI.

Disclosures

This manuscript has been read and approved by all authors. This paper is unique and is not under consideration by any other publication and has not been published elsewhere. The authors and peer reviewers of this paper report no conflicts of interest. The authors confirm that they have permission to reproduce any copyrighted material. Written consent was obtained from the patients parents for publication of this study.

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