Sudden cardiac death: improving our pathological diagnosis—are we there yet?

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This editorial refers to ‘The importance of specialist cardiac histopathological examination in the investigation of young sudden cardiac deaths’ by S.V. de Noronha et al., on page 899.

Any truth is better than indefinite doubt. Sherlock Holmes

Sudden cardiac death (SCD) is by nature unexpected and not infrequently present in a person without pre-existing heart disease. The abrupt and unexpected presentation carries a significant burden on family members leading to great anxiety and the need to demand answers regarding the cause of the unexpected loss. Sudden cardiac death is defined as a death occurring within 1 h of an acute change in clinical status, or an unexpected death that occurred within the previous 24 h.1–3 The specificity of this definition is questionable, and the final diagnosis derived from autopsy reports also varies significantly.1

The incidence of SCD is widely variable and in the USA ranges between 180 000 and 450 000 cases annually.4 Recent studies in different geographical regions including the USA,5,6 the Netherlands,7 England, and Ireland,8–10 and China11 indicate that SCD rates range from 50 to 100 per 100 000 in the general population.1 Coronary artery disease continues to be the primary cause of SCD in the population studies1 However, in younger populations (<40 years) hereditary channelopathies are the leading cause of SCD.1

Determination of the cause of SCD has significant implications for screening and potential for prophylactic treatment of first-line family members of the deceased. Unfortunately, resources are inadequate and forensic expertise in this area remains limited. Whether the need for specialized cardiac histopathological examination improves the accuracy of post-mortem examination particularly in young subjects presenting with SCD remains to be determined.

In this issue of the Journal, de Noronha et al.12 address the role of expert cardiac histopathological examination in a series of SCD cases in the young. The authors report their experience from a prospective observational study conducted at a tertiary centre in the UK. Data on hearts from 720 consecutive cases of SCD referred by coroners and pathologists from 2007 to 2009 were included in this report. The median age of this sample was 32 years, and the majority of the deaths occurred at home (57%) and most were males (66%). Sudden cardiac death was mainly associated with a morphologically normal heart in 45%, followed by cardiomyopathy in 29%, and coronary artery disease in only 10%.

The investigators also had the opportunity to review and compare the diagnostic accuracy of the referring pathologist in 158 cases. Interestingly, referring pathologists tended to overestimate the presence of cardiomyopathy instead of identifying a morphologically normal heart with 37% of the hearts being incorrectly labelled as cardiomyopathy. Of note, the authors identified ‘idiopathic’ left ventricular hypertrophy as the most frequent structural abnormality in this large cohort. This finding did not appear to have any correlation with the circumstances of SCD, i.e. exercise or rest and did not discriminate between athletic and non-athletic individuals. The role of ‘idiopathic’ left ventricular hypertrophy and its causality with SCD remains puzzling. The same investigators have made further observations on this pathological finding and in half the cases identified a trend towards documenting more evidence of cardiomyopathy that this ‘idiopathic’ left ventricular hypertrophy is in fact not a variant of hypertrophic cardiomyopathy. The present report confirms previous findings related to the causes of SCD and the circumstances of death. In this report, SCD at rest was associated two times more often with no evidence of structural heart disease, whereas a trend towards documenting more evidence of cardiomyopathy was observed in those dying during exercise.

The present study has several limitations that are recognized by the investigators and that merit further comment. In the midst of the 21st century, ‘molecular autopsy’ should be practically mandatory, particularly for SCD in subjects under the age of 40.15 Clearly, we have to stimulate the availability of this resource worldwide and more cardiac pathologists with expertise in this area should be trained. Overdiagnosis of ‘structural’ alterations including arrhythmogenic right ventricular cardiomyopathy may in part be related to

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the pressure of providing some type of diagnosis offering closure to the devastated families that have to deal with a sudden unexpected death in a young member. Finally, further investigation into the puzzling role of ‘idiopathic’ left ventricular hypertrophy should be stimulated in an attempt to determine whether this finding is noise or actually can aid in the early identification of subjects at risk of SCD. Newer cardiac imaging technologies may be able to aid in this context.

The authors should be commended for presenting a systematic pathological approach in subjects who demised due to SCD. These findings highlight the need for an expert histopathological review in cases of SCD in general and particularly due to the social impact that an unexpected death has when the subject is young and has a life ahead to complete. In the end as Sherlock Holmes would wisely remark: ‘Any truth is better than indefinite doubt’.16

**Conflict of interest:** none declared.

**References**