

# Sudden arrhythmic death syndrome (SADS): Diagnostic yield of comprehensive clinical evaluation of paediatric first-degree relatives

V.Giudici<sup>1</sup>, A.Spanaki<sup>1</sup>, J. Hendry<sup>1</sup>, S.Mead-Regan<sup>1</sup>, E.Field<sup>1</sup>, D.Abrams<sup>3</sup>, M.Lowe<sup>1</sup>, J.P. Kaski<sup>1,2</sup>

<sup>1</sup>Inherited Cardiovascular Diseases Unit, Great Ormond Street Hospital, London, UK

<sup>2</sup>Institute of Cardiovascular Science, University College London, UK

<sup>3</sup>Boston Children's Hospital, Boston MA, USA

## Background

- Sudden arrhythmic death syndrome (SADS) is most often caused by heritable cardiac diseases
- Studies in adults have identified evidence of inherited cardiovascular diseases in up to 53% of families
- Data on the prevalence of familial disease in children are scarce

The aim of this study was to evaluate the yield of clinical screening in paediatric first-degree relatives of victims of SADS or aborted cardiac arrest (ACA) using a systematic and comprehensive protocol.

## Methods

### Patients

- 110 consecutive patients (63 families)
- Referred for screening between 2003 and 2013 after a SCD or ACA of a 1<sup>st</sup>-degree relative
- Median age 7.3 years (IQR 3.8-10.8)
- 42% female

### Systematic evaluation

- Family history
- Resting, ambulatory and signal-averaged ECG
- Echocardiogram
- Exercise testing
- Cardiac MRI
- Ajmaline provocation testing

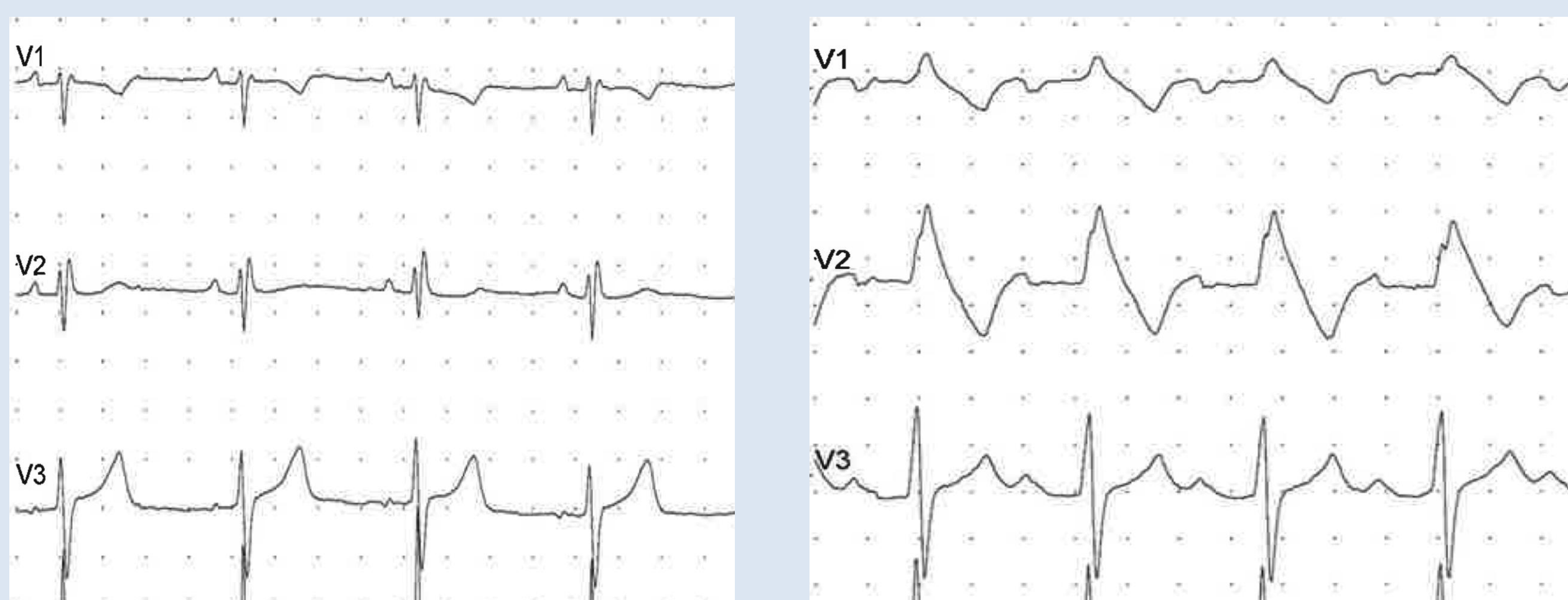


Figure 1: Positive ajmaline provocation test in a 15 year-old girl referred following SADS during sleep in her father

## Results

An inherited cardiac disease was identified in 10 children from 9 families (15%)

Family	Dx	Test	Event	Child's age at Dx	Relation to proband
1	BrS	Ajmaline test	SADS	15	Offspring
2	BrS	Ajmaline test	ACA	10	Offspring
		Ajmaline test		9	Offspring
3	BrS	Ajmaline test	SADS	8	Offspring
4	BrS	Ajmaline test	SADS	13	Offspring
5	BrS	Ajmaline test	SADS	15	Offspring
6	BrS	Ajmaline test	SADS	13	Offspring
7	CPVT	Event recorder	SADS	15	Offspring
8	LQTS	ECG, exercise test, genetics	SADS	13	Offspring
9	LQTS	ECG, exercise test	ACA	13	Sibling

- 2 additional patients had late potentials on SAEKG

## Conclusion

- This study shows a high prevalence of inherited heart disease detected with the systematic and comprehensive clinical screening of paediatric first-degree relatives of SADS victims.
- The results highlight the importance of early referral and evaluation of all paediatric members of SADS families, with important implications for the prevention of SCD in other at risk family members