Background

• SCD accounts for ~400 deaths per year in UK population (1-34 years old).

• Undiagnosed *inherited cardiac conditions* cause ~50% of SCD cases.

• Aetiology of paediatric SCA specifically is unknown.
  - Lack of studies
  - Differences in research design, inclusion criterias, etc.

Aim: What is the aetiology of out-of-hospital cardiac arrest in paediatric survivors?
Methods

• Single-centre, retrospective study

• 37 patients (0-16 years) – survived out-of-hospital VF arrest between Aug 1996 and Oct 2017

• Significant clinical notes and investigation findings reviewed and recorded

• 21 out of 37 (57%) were males
• Median age at arrest was 11.0 years (IQR = 6-13 years)

• Exclusion criteria:
  - SCA secondary to a non-cardiac cause
  - Previously known relevant cardiac condition
Diagnoses

- 33/37 patients (89.2%) diagnosed with inherited cardiac condition.

- LQTS and HCM most commonly diagnosed condition.

Investigations used to make diagnosis

- ECG and ECHO most frequently used investigation to confirm diagnosis.
# Genetic Testing

- 25/37 genetic results available → 17 causative mutations found

<table>
<thead>
<tr>
<th>Diagnosis</th>
<th>Number of patients identified with a gene mutation/total number of patients tested (% yield)</th>
<th>Gene Mutations</th>
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</table>
| LQTS      | 4/9 (44%)                                                                                 | a) SCN5A (LQT3)  
|           |                                                                                         | b) KCNH2 (LQT2)  
|           |                                                                                         | c) CACNA1C (LQT8) |
| HCM       | 5/7 (71.4%)                                                                              | a) MYBPC3       
|           |                                                                                         | b) TNNI3        |
| CPVT      | 6/7 (85.7%)                                                                              | a) RYR2         |
| SQTS      | 1/1 (100%)                                                                               | a) SLC22A5      |
| BrS       | 1/1 (100%)                                                                               | a) SCN5A        |

- Genetic testing important as a diagnostic tool
Conclusions

• Inherited cardiac conditions the most common cause of out-of-hospital cardiac arrest

• LQTS and HCM most commonly diagnoses

• ECG and ECHO most useful investigations

• Highlights importance of genetic testing.
Clinical Implications

- Full clinical and genetic workup of proband is needed
- Important to screen first-degree relatives