

*It is extremely important that all family members be tested for these conditions once one family member is identified as having the condition.*

*Any young person who has experienced unexplained fainting should be tested.*

## **SADS' Mission**

To save the lives and support the families of children & young adults who are genetically predisposed to sudden death due to heart rhythm abnormalities.

## Who Should be Screened?

All children should be carefully screened, using the Risk Assessment Form ([www.StopSADS.org](http://www.StopSADS.org)), every few years at these times: preschool, before/during middle school, before/during high school, and before participating in organized sports.

Long QT Syndrome (LQTS), Hypertrophic Cardiomyopathy (HCM), Arrhythmogenic Right Ventricular Displasia (ARVD) should be considered in any otherwise healthy child or young adult who:

- experiences loss of consciousness (syncopal or near syncopal spells) during or immediately following exercise or emotional distress - particularly in cases where the events are repetitive.
- has a family history of unexplained sudden death in otherwise healthy young persons (under age 40).
- experiences consistent or unusual chest pain and/or shortness of breath during exercise.

## The Screening Process

**Family History:** Using the Risk Assessment Form from SADS ([www.StopSADS.org](http://www.StopSADS.org)), a parent and the child's physician can work together to determine the family risk of these conditions. This is extremely important and should include unexplained death during swimming, death during seizures, a family history of "seizure" disorders and other sudden deaths or death before age 40 in the extended family. Remember, this history should be taken for both the mother's family & the father's family!

**Resting Electrocardiogram:** The ECG will detect some people who have the Long QT Syndrome. However, sometimes the resting ECG is not able to diagnose LQTS. In this case, doing serial ECGs (more than 1 ECG in a row on different days) or ECGs on other family members (parents, siblings) will help to diagnose LQTS. Since this is a genetic condition, most likely a parent and siblings will have the syndrome as well.

**Exercise (stress) Electrocardiogram:** An exercise test, which allows the person to exercise for 10 to 15 minutes without achieving a heart rate more than 150-160, beats per minute works best for this process. The standard treadmill test, which is used for detecting coronary artery disease, is *not as good* for LQTS as is the slower, longer test.

**Echocardiogram:** This is a non-invasive sound wave imaging test that allows the physician to observe the heart as it is beating. It allows for the measurement of muscle thickness, degree of obstruction, chamber size, valve movement and blood flow. It is also possible to determine pressures inside the heart chambers and major vessels.

**Genetic Testing:** Two commercial CLIA-certified companies (GeneDx and Transgenomic/FAMILION) offer a clinical diagnostic test that searches for genetic misspellings in the five major genes that cause approximately 70-80% of LQTS. One family member will be tested first. Once/if a family member has a gene mutation identified, testing of other family members for that mutation is available, and can assist in clarifying those family members with non-definitive ECG findings. In fact, if a genetic diagnosis of LQTS is established for the index case, the **ONLY** definitive test to rule in or rule out LQTS for family members and relatives is the LQTS genetic test.



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FOUNDATION

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