The Importance of Incorporating Health Literacy in Translating Cardiogenetic Knowledge to Clinical Practice

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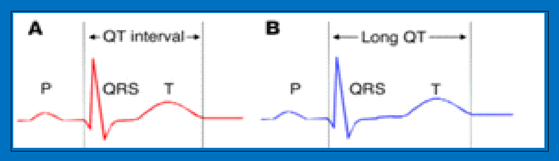
Introduction

In the growing era of personalized genomic medicine, the ability to offer disease risk assessments and predictive testing to individuals presents ethical, legal and social concerns as well as significant communication challenges to clinicians. These challenges are amplified in the area of cardiogenetics, where molecular genetic technology exists to detect mutations responsible for cardiac channelopathies including Long QT Syndrome (LQTS) and Brugada Syndrome, which can explain a previously sudden unexplained death in an adult (SUDS) or infant (SIDS). Results from this genetic testing may have implications for surviving family members, who may benefit from lifesaving personalized medical and lifestyle recommendations. The health literacy area is vital in this respect, as the genetics field continues to grapple with the challenges of conveying information accurately to patients and ensuring comprehension.

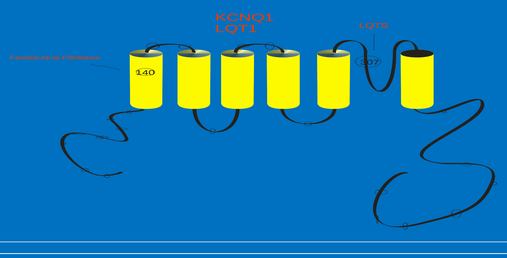
Background

SIDS: Sudden Infant Death Syndrome
SUDS: Sudden Unexplained Death Syndrome

	SIDS/Crib Death	SUDS
Timing	1st year of life	Ages 1-35 years
Prevalence	54 per 100,000 births	Unknown
Genetics	Approx. 10-20% of cases associated with a cardiac ion channel mutation	~50% identifiable genetic etiology, 75% of which is an ion channel gene mutation

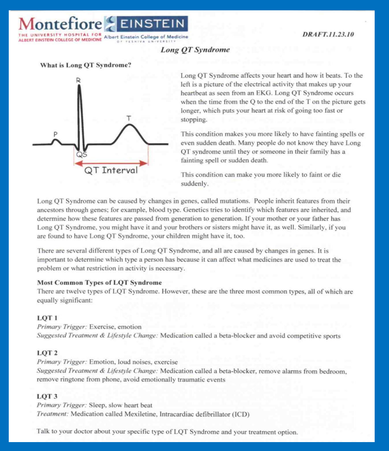


QT interval can be prolonged by mutations in K^+ (potassium) or Na^+ (sodium) channel



Methods regarding Patient Health Literacy

I. Genetic testing for proband and at-risk relatives in the clinic
II. New gene discovery
III. Conducting interviews on subjective patients learning about Long QT syndrome
IV. Focus Groups discussing implications of fact sheets and their strengths and weaknesses
V. Implications surrounding usage and education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Long QT Syndrome

What is Long QT Syndrome?

Long QT Syndrome affects your heart and how it beats. To the left is a picture of the electrical activity that makes up your heartbeat as seen from an EKG. Long QT Syndrome occurs when the time from the Q to the end of the T on the picture gets longer, which puts your heart at risk of going too fast or stopping.

This condition makes you more likely to have fainting spells or even sudden death. Many people do not know they have Long QT syndrome until they or someone in their family has a fainting spell or sudden death.

This condition can make you more likely to faint or die suddenly.

Long QT Syndrome can be caused by changes in genes, called mutations. People inherit features from their ancestors through genes; for example, blood type. Genetics tries to identify which features are inherited, and determine how these features are passed from generation to generation. If your mother or your father has Long QT Syndrome, you might have it and your brothers or sisters might have it, as well. Similarly, if you are found to have Long QT Syndrome, your children might have it, too.

There are several different types of Long QT Syndrome, and all are caused by changes in genes. It is important to determine which type a person has because it can affect what medicines are used to treat the problem or what restriction in activity is necessary.

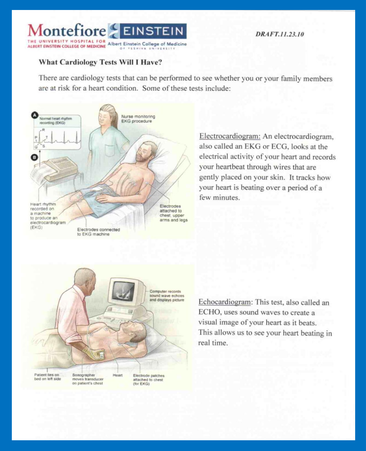
Most Common Types of LQT Syndrome
There are twelve types of LQT Syndrome. However, these are the three most common types, all of which are equally significant:

LQT 1
Primary Trigger: Exercise, emotion
Suggested Treatment & Lifestyle Change: Medication called a beta-blocker and avoid competitive sports

LQT 2
Primary Trigger: Emotion, loud noises, exercise
Suggested Treatment & Lifestyle Change: Medication called a beta-blocker, remove alarms from bedroom, remove ringtone from phone, avoid emotionally traumatic events

LQT 3
Primary Trigger: Sleep, slow heart beat
Treatment: Medication called Mexiletine, Intracardiac defibrillator (ICD)

Talk to your doctor about your specific type of LQT Syndrome and your treatment option.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What Cardiology Tests Will I Have?

There are cardiology tests that can be performed to see whether you or your family members are at risk for a heart condition. Some of these tests include:

Electrocardiogram: An electrocardiogram, also called an EKG or ECG, looks at the electrical activity of your heart and records your heartbeat through wires that are gently placed on your skin. It tracks how your heart is beating over a period of a few minutes.

Echocardiogram: This test, also called an ECHO, uses sound waves to create a visual image of your heart as it beats. This allows us to see your heart beating in real time.

Preliminary Results: Related to Health Literacy

Qualitative research is particularly well suited to studying the subjective experiences of clinic patients with the goal of improving health literacy, informed decision-making, and compliance with medicine and lifestyle recommendations.

Theme: Family Interaction

Finding:
Differences between and within families in their attitudes toward sharing genetic information with other family members.
a) "You can tell people…I got this (LQT) you should get tested, but it's up to them"
b) "Your heart takes a little bit longer to re-start than most people's. So they want to give you this (ICD) so if something happens, you'll be OK"

Finding Issues:
1. Do clinicians have a duty to warn at-risk family members if the proband refuses to disclose testing results?
2. What are the obligations of the clinician to an adolescent family member regarding informing and genetic testing?
3. Should parents be required to seek testing for an inform their minor children of results if the family history indicates increased risk? Are there special considerations for adolescents considering their age and/or psychological maturity?

Theme: Comprehension of Health Information

Finding:
Difficulty understanding both the health condition (LQTS) and the meaning of genetic testing
a) "I don't know what LQT means-you gotta explain it to me-talk to me in plain English."
b) "Every time I go into the hospital, I'm so nervous- they might be telling me exactly what it is, but it's not going in there."

Finding Issue:
1. How must we, as clinicians, change the way we explain things based on education level? Language? Cultural or religious beliefs?

Theme: Standardization

Finding:
Desire for standardization of emergency room, hospital, and medical examiner procedures (in families that experienced a sudden death of a family member)
a) "I would have liked someone to walk with you; explain the tests; tell you who to call."
b) "You have so many people asking you the same questions over and over again. It's understandable, but it's a lot."
c) "I think all doctors should have you walk out of their offices with something in your hand – a piece of paper."

Finding Issues:
1. How might the timing of desiring genetic information vary for different families who have had experiences with SIDS? Near death experience? Death of a family member?
2. What procedures should be developed to assess a family's readiness to receive genetic information?
3. What cultural or religious beliefs might influence an individual's desire for explanations or amount of discussion?

Summary

Our preliminary thematic findings have already influenced our practices in the MECC; for example, we have developed informational handouts for patients. These fact sheets are unique in that they provide basic definitions of the condition, use pictures to illustrate meaning and are written at an easy-to-understand level. These fact sheets can also be translated to usage with patients who have other cardiac channelopathy conditions and aid in communicating and defining these conditions to the public in general. The results from this interdisciplinary model provide a framework for using clinical, genetic, and health literacy information to personalize preventive health strategies and improve research, policy and programs in cardiogenetics.

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