Needles in haystacks:
Inherited Cardiac Conditions in Primary Care Pediatrics

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Inherited Cardiac Conditions

- 18-year old come to Pediatric Clinic for prescription for seasonal allergies
- Mother present
- Appear to be ‘worse’ this year
- Sudden death of 15-year old sister 6 months prior
  - Prior episode of syncope - ‘hyperventilation’
  - Altitude in Chile - no diagnosis yet available
Inherited Cardiac Conditions

**CARDIOMYOPATHIES**

- Hypertrophic cardiomyopathy
  - 1:500

- Dilated cardiomyopathy
  - 1:3000

- Arrhythmogenic right ventricular cardiomyopathy
  - 1:5000

- Long QT syndrome
  - 1:2000

- Brugada syndrome
  - 1:3500

- Catecholaminergic polymorphic ventricular tachycardia
  - 1:10000

**INHERITED ARRHYTHMIA SYNDROMES**
Inherited Cardiac Conditions

- Variations in the genetic reading frame in specific cardiomyocyte genes

**KCNQ1**

Transmembrane domains
Pore region

**KvLQT1**
Inherited Cardiac Conditions

- Variations in the genetic reading frame in specific cardiomyocyte genes

- Autosomal dominant inheritance

- Highly varied penetrance

- Significant overlap with common physiological and pathophysiological phenomena

- Lifelong asymptomatic to sudden cardiac death

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**Desmosome**
- Desmin

**Arrhythmogenic cardiomyopathy**

**Lamin A/C**
- Sarcomere
- Desmin, Titin
- Dystrophin

**Dilated Cardiomyopathy**

**Sarcomere**
- Hypertrophic Cardiomyopathy

**Cardiac Na⁺/K⁺ channels**
- Long QT syndrome
- Brugada syndrome
- Short QT syndrome

**Cardiac Ca²⁺ channels**
- Catecholaminergic polymorphic ventricular tachycardia
Inherited Cardiac Conditions

- Clinical and genetic features
  - Autosomal dominant inheritance
  - Highly varied penetrance
  - Significant overlap with common physiological and pathophysiological phenomena
  - Lifelong asymptomatic to sudden cardiac death
Inherited Cardiac Conditions

When do these patients *present* to primary care

### The 12-element American Heart Association Recommendations for Preparticipation Cardiovascular Screening of Competitive Athletes

<table>
<thead>
<tr>
<th>Medical History</th>
<th>Physical Examination</th>
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<tbody>
<tr>
<td>• Personal history</td>
<td>• Heart murmur</td>
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<tr>
<td>– Exertional chest pain/discomfort</td>
<td>• Femoral pulses to exclude aortic coarctation</td>
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<tr>
<td>– Unexplained syncope/near-syncope</td>
<td>• Physical stigmata of Marfan syndrome</td>
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<tr>
<td>– Excessive exertional and unexplained dyspnea/fatigue associated with exercise</td>
<td>• Brachial artery blood pressure (sitting position)</td>
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<td>• Family history</td>
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<tr>
<td>– Premature death (sudden and unexpected, or otherwise) before age 50 years due to heart disease in first-degree relative</td>
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<tr>
<td>– Disability from heart disease in a close relative younger than 50 years of age</td>
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<tr>
<td>– Specific knowledge of certain cardiac conditions in family members:</td>
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<tr>
<td>hypertrophic or dilated cardiomyopathy, long QT syndrome and other ion channelopathies, Marfan syndrome, and clinically important arrhythmias</td>
<td></td>
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</tbody>
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Inherited Cardiac Conditions

When do these patients present to primary care

- Personal symptoms
  - Consistent with an inherited cardiac condition
  - Pre-syncope, syncope, palpitations, exertional dyspnoea
- Pre-participation screening or routine medical examination
  - Abnormal history or clinical findings
- Family history of inherited cardiac condition
- Family history of sudden cardiac death
Inherited Cardiac Conditions

When do these patients *present* to primary care

- Initial investigations:
  - Electrocardiogram
  - Echocardiogram

Accurate electrocardiographic assessment of the QT interval: Teach the tangent

Heart Rhythm 2008;5:1015–1018

Pieter G. Postema, MD, Jonas S.S.G. De Jong, MD, Ivo A.C. Van der Bilt, MD, Arthur A.M. Wilde, MD, PhD

**METHODS** Four previously published ECGs (two LQTS, two normal) were assessed by 151 medical students using the following QT measurement method: (1) the end of the T wave is the intersection of a tangent to the steepest slope of the last limb of the T wave and the baseline, in lead II or V5; (2) $QTc = QT / \sqrt{RR}$; (3) $QTc > 450$ ms is prolonged. Four months later, 71 students measured the ECGs again. Student results were compared with previously published results on the same ECGs of 25 LOTS experts, 106 arrhythmia experts, and 771 cardiologists and noncardiologists.

**RESULTS** Correct QT interval interpretations were achieved by 71% and 77% of students during the first and second test, respectively, as compared with 62% by the arrhythmia experts and <25% by the cardiologists and noncardiologists.
Inherited Cardiac Conditions

When do these patients present to primary care

- Personal history
- Syncope - exertional (swimming), emotion

QTc 533ms  QTc 640ms

CONGENITAL DEAF-MUTISM, FUNCTIONAL HEART DISEASE WITH PROLONGATION OF THE Q-T INTERVAL, AND SUDDEN DEATH

ANTON JERVELL, M.D., AND FRED LANGE-NIELSEN, M.D.
TÖNSBERG, NORWAY
Inherited Cardiac Conditions

When do these patients *present* to primary care

- Personal history
- Syncope - exertional (swimming), emotion
Inherited Cardiac Conditions

When do these patients *present* to primary care

- Personal history
- Syncope - auditory stimulation
Inherited Cardiac Conditions

When do these patients present to primary care

- Personal history
- Atypical epilepsy

An 8-year old boy investigated for recurrent syncope associated with parental separation, a dog-attack, school fire drills and with ‘psychic’ stimuli. Witnessed episodes associated with generalized convulsive movements, frothing at the mouth and incontinence. He was given a presumptive diagnosis of epilepsy … but episodes were considered hysterical as examination and EEG normal.

Another woman had been diagnosed with and treated for epilepsy for many years before reading an article …… in a magazine. The description was so consistent with her own experience that she was prompted to seek a further medical opinion at which time a diagnosis ……was established.
Inherited Cardiac Conditions

When do these patients present to primary care

- Personal history
- Febrile seizures
Inherited Cardiac Conditions

When do these patients *present* to primary care

- Family history
- Sudden cardiac death <40 years of age
Inherited Cardiac Conditions

When do these patients *present* to primary care

- Family history
  - Sudden cardiac death <40 years of age
  - Drowning in competent swimmers
  - Unexplained motor vehicle accidents
  - Refractory epilepsy never responsive to medication
  - Sudden infant death/late fetal demise
Inherited Cardiac Conditions

When do these patients *present* to primary care

- Abnormal clinical findings
  - Murmur
Inherited Cardiac Conditions

Boston model
Boston Children’s & Brigham & Women’s

- ‘Family’ is seen as the unit of care
  - Rapid access service
  - Comprehensive, coordinated program irrespective of age
  - Detailed familial evaluation in one day - ‘one-stop shop’
  - Evaluation begins at the moment of referral
Inherited Cardiac Conditions

Boston model
Boston Children’s & Brigham & Women’s

- Detailed clinical and genetic evaluation in the context of the extended family
Inherited Cardiac Conditions

Boston model
Boston Children’s & Brigham & Women’s

- Education for the family

Boston Children’s Hospital Heart Center
Connect, Share, Learn:
Long QT Syndrome Family Day

Saturday, November 8, 2014
The Joseph B. Martin Conference Center at Harvard Medical School
77 Avenue Louis Pasteur, Boston, MA

The Heart Center at Boston Children’s Hospital invites you to its first-ever Long QT Syndrome Family Day, a day-long free conference bringing together families and clinical experts to discuss the latest information surrounding this inherited heart arrhythmia condition.

We look forward to seeing you in November!

REGISTER FOR THIS FREE EVENT*
bostonchildrens.org/longqtconference

*Space is limited. Register by October 15 to reserve a spot for you and up to four family members.
Inherited Cardiac Conditions

Boston model
Boston Children’s & Brigham & Women’s

- Concept of ‘shared care’ between primary care & the Inherited Arrhythmia Program
  - Physician education
    - Medications to avoid - long QT syndrome, Brugada syndrome
  - Sports participation
  - Point of easy contact
Inherited Cardiac Conditions

Boston model
Boston Children’s & Brigham & Women’s

• Who to refer

  • Individual with clinical suspicion
  • Following diagnosis in a family member
  • Following sudden cardiac death age <40yrs
  • Exclusion of inherited cardiac disease identified in athletes as part of (pre)participation screening
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Boston model
Boston Children’s & Brigham & Women’s

- ECG recorded after episode of ‘hyperventilation’
- Faxed to BCH
- Family seen within 48 hrs
- Diagnosis of long QT syndrome made in both sisters and mother
Inherited Cardiac Conditions

Boston model
Boston Children’s & Brigham & Women’s

• How to refer

Patient/family referral
Elizabeth Hobin - Program Coordinator
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1-617-355-6432

Clinical questions/opinions
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Clinical questions/opinions
Dominic Abrams - Program Director
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1-617-355-6432
Inherited Cardiac Conditions

• Conclusions

  • Inherited cardiac conditions are rare

  • Symptoms include palpitations, syncope and sudden cardiac death

  • Significant overlap with the normal spectrum of cardiovascular signs and symptoms

  • Early discussion and referral may be life saving
Inherited Cardiac Conditions
Care Coordination Information

For more information on Care Coordination or to review slides, please visit:

http://www.childrenshospital.org/care-coordination-curriculum/care-coordination-measurement

For additional questions, please email:
Richard.antonelli@childrens.harvard.edu