MEDPED
A humanitarian project to find and help persons with familial hypercholesterolemia (FH)

FH is under-recognized and under-treated!

Web site:  www.medped.org
A striking family history

Key
- Red square: Affected
- Heart: MI age 40
- Number: Total Chol

Hopkins PN. Clin Lipidol 2010 in press
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What are some of the challenges of FH screening?

♥ Identifying FH patients
  - DNA (gold standard) versus clinical?
  - Who pays?

♥ There is urgency to finding FH
  - Previously, the Dx usually came after MI
  - Earlier, aggressive Rx → better outcomes
    • Universal screening of teenagers cost-effective
Additional challenges in US

♥ No current data on:

- How many FH are diagnosed
  - <33%? (Williams RR, Am J Cardiol 1993; 72:18D)
- What percent are treated (any Rx)
- How well FH are treated (current LDLs)
- CURRENT CAD RISKS OR OUTCOMES
Entry levels of total cholesterol in the MEDPED Treatment Support Program

Total cholesterol (mg/dL) vs. Year of entry (n)

- <1993 (14)
- 1993 (21)
- 1994 (43)
- 1995 (75)
- 1996 (176)
- 1997 (56)

Hopkins PN. Clin Lipidol 2010 in press
Challenges for Dx

♥ Physical findings
  • Tendon xanthomas
  • Arcus

♥ Serum LDL levels

♥ DNA Dx
Darryl Kile

Pitcher for the St. Louis Cardinals
Died June 22, 2002, age 33
Survived by widow and 3 young children.
Autopsy: 80-90% coronary artery stenoses.
FHx: father died of MI age 44.
Prevalence (%) in 346 Utah FH Heterozygotes

Age (years)

- <30
- 30-39
- 40-49
- 50-59
- 60+

Xanthoma

Full Arcus

Hopkins PN. Current Treatment Options in Cardiovascular Medicine 2002; 4:121
LDL cholesterol levels

<table>
<thead>
<tr>
<th></th>
<th>mean</th>
<th>SD</th>
</tr>
</thead>
<tbody>
<tr>
<td>FH</td>
<td>289</td>
<td>63.0</td>
</tr>
<tr>
<td>Non-FH</td>
<td>130</td>
<td>31.4</td>
</tr>
</tbody>
</table>

80% probability of FH

False negatives

False positives

Hopkins PN. Clin Lipidol 2010 in press
## Practical LDL Criteria for Diagnosing Heterozygous FH in U.S. Population

Predicted total-C and (LDL-C) in mg/dl cut points achieving at least 80% probability for an individual to be affected.

<table>
<thead>
<tr>
<th>Age</th>
<th>First</th>
<th>Second</th>
<th>Third</th>
<th>General</th>
<th>“100%”</th>
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</thead>
<tbody>
<tr>
<td>&lt;20</td>
<td>220 (155)</td>
<td>230 (165)</td>
<td>240 (170)</td>
<td>270 (200)</td>
<td>(240)</td>
</tr>
<tr>
<td>20-29</td>
<td>240 (170)</td>
<td>250 (180)</td>
<td>260 (185)</td>
<td>290 (220)</td>
<td>(260)</td>
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<tr>
<td>30-39</td>
<td>270 (190)</td>
<td>280 (200)</td>
<td>290 (210)</td>
<td>340 (240)</td>
<td>(280)</td>
</tr>
<tr>
<td>40+</td>
<td>290 (205)</td>
<td>300 (215)</td>
<td>310 (225)</td>
<td>360 (260)</td>
<td>(300)</td>
</tr>
</tbody>
</table>

Cumulative Probability of Non-Fatal CAD in Utah FH vs a Random U.S. Population

6-fold greater risk in men

Hopkins PN. Clin Lipidol 2010 in press
Why focus on FH?

♥ FH provides the only example of a genetic cause of premature CAD for which a systematic, population-based approach to proband identification and family screening is clearly warranted at this time.
Thank You!