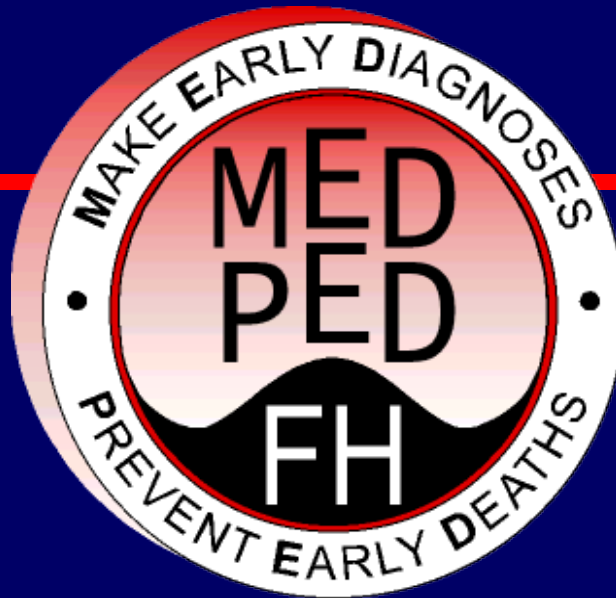


MEDPED

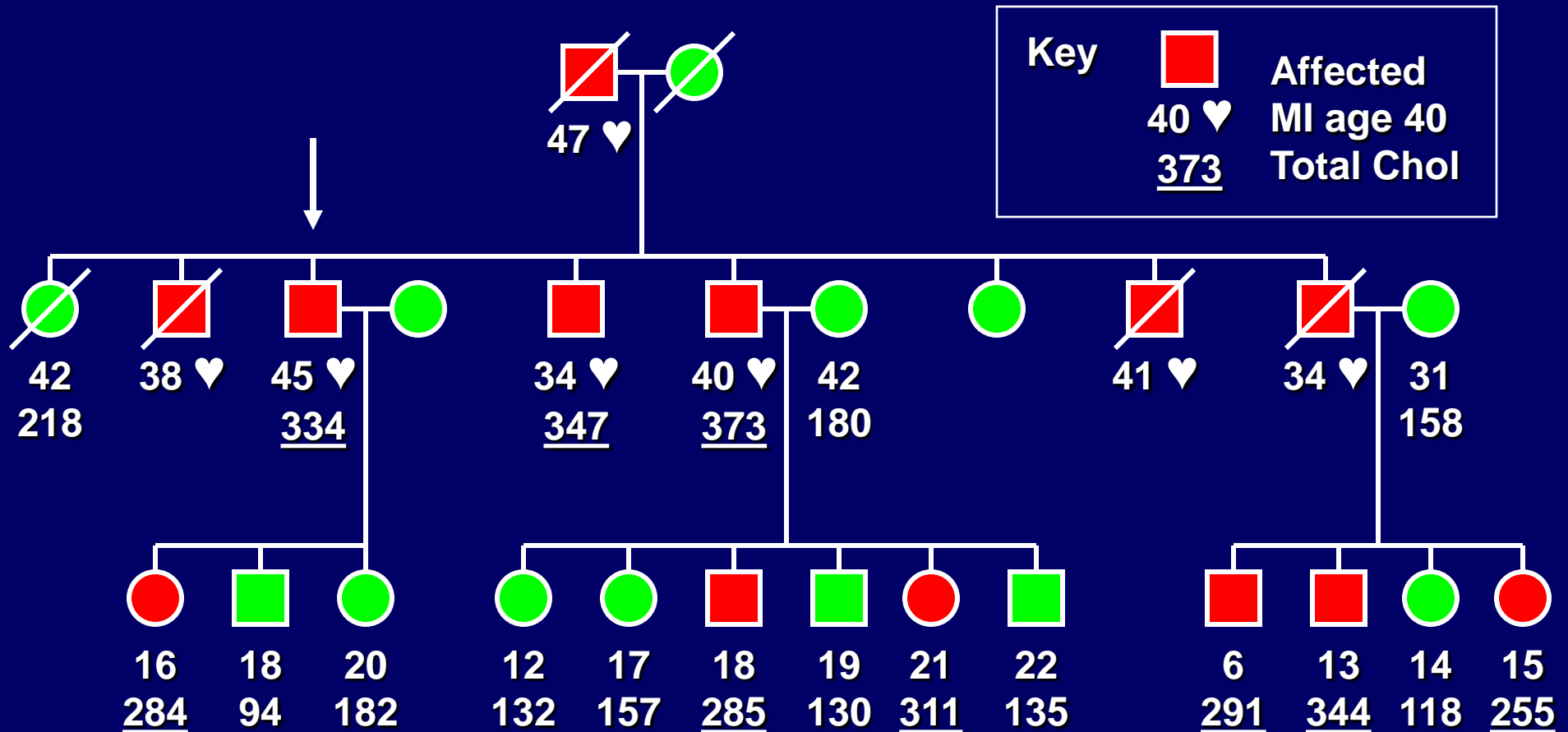
A humanitarian project to find and help persons
with familial hypercholesterolemia (FH)



FH is under-recognized and under-treated!

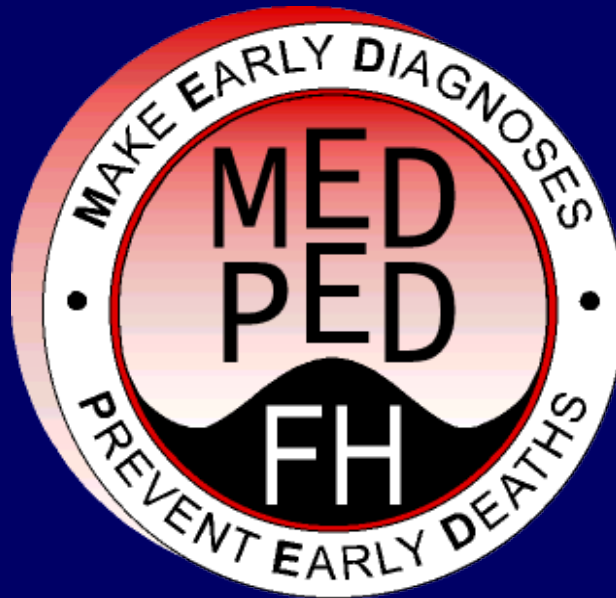
Web site: www.medped.org

A striking family history



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

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

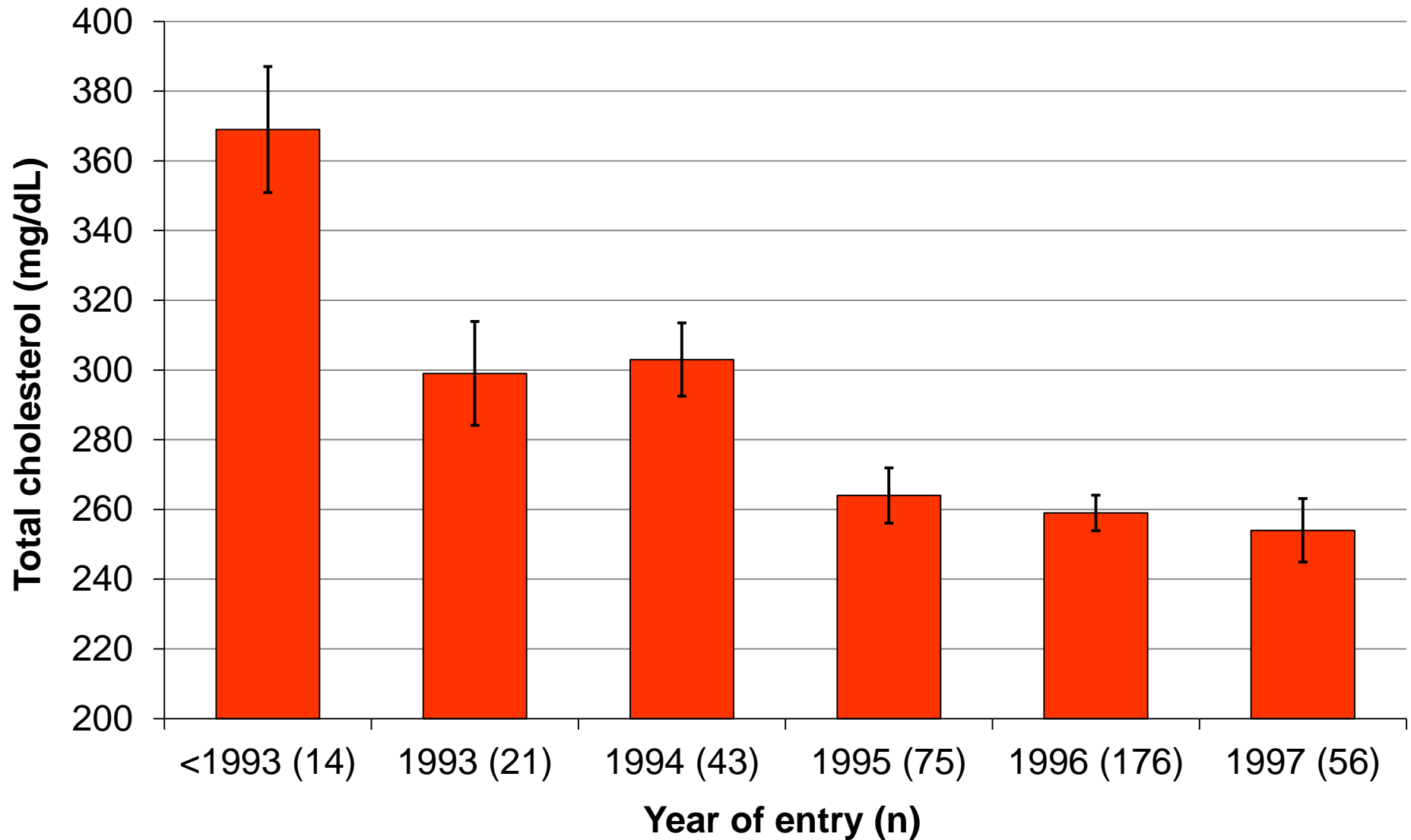
- Previously, the Dx usually came after MI
- Earlier, aggressive Rx → better outcomes
 - Universal screening of teenagers cost-effective

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



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.

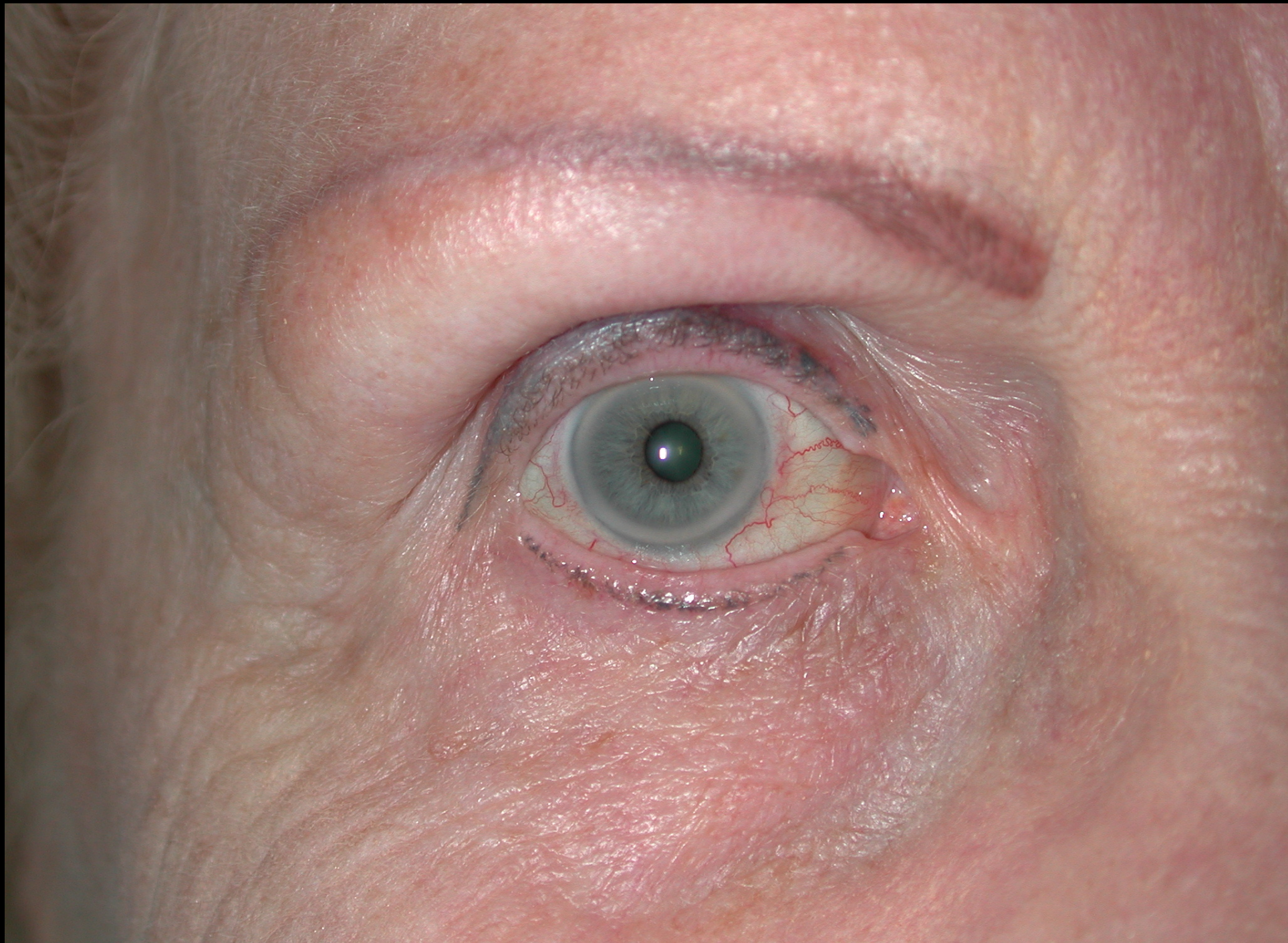


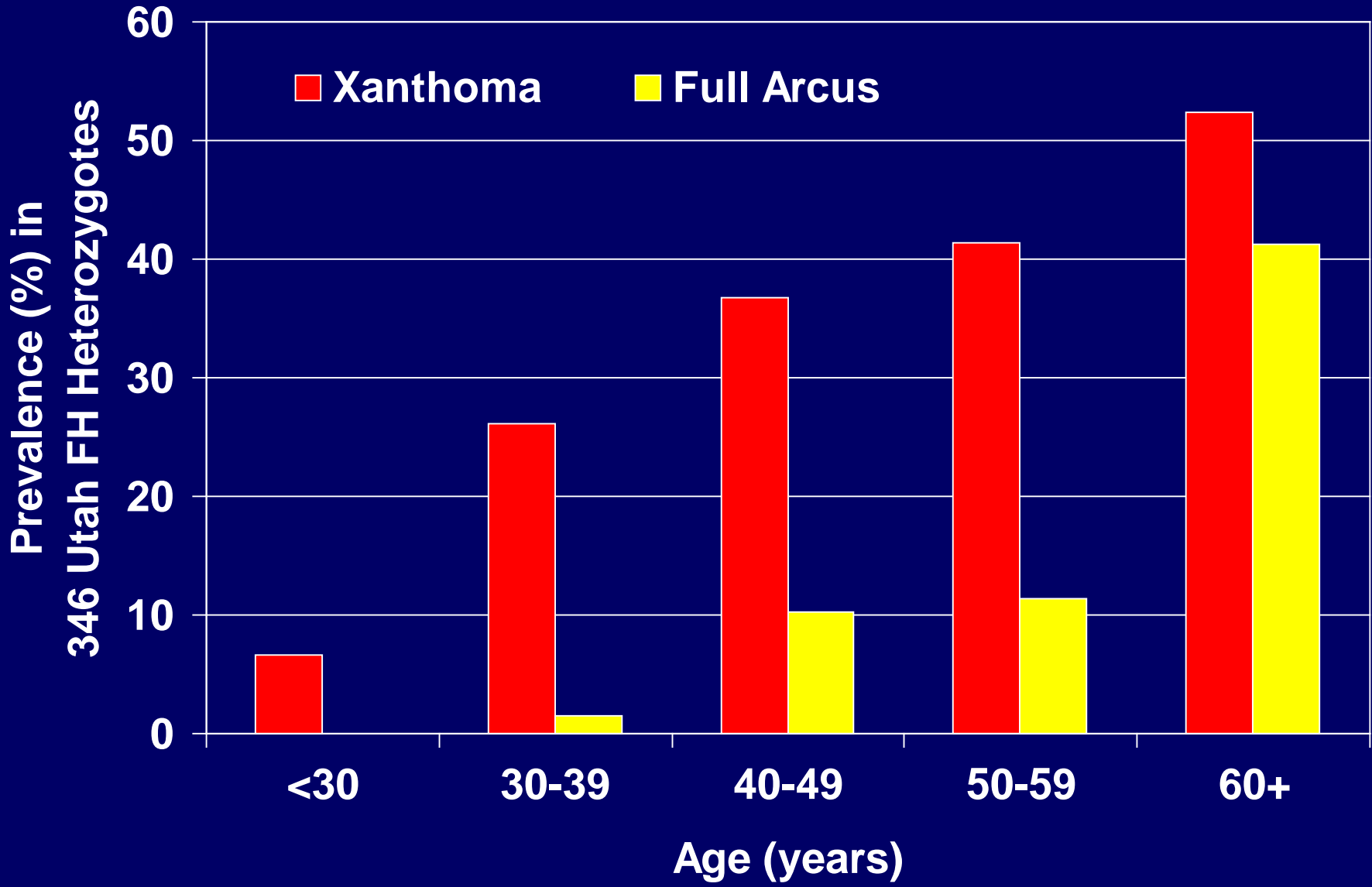


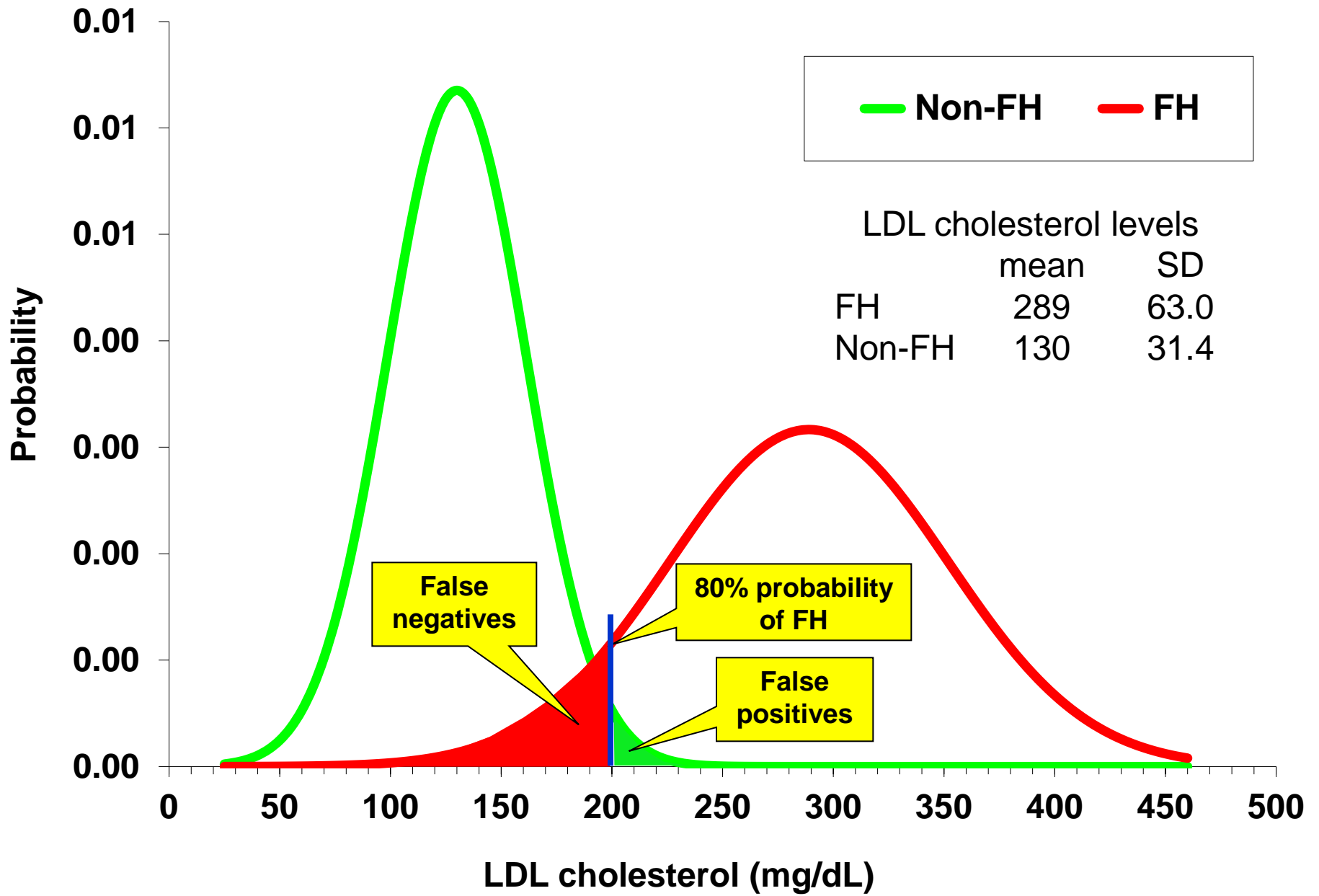










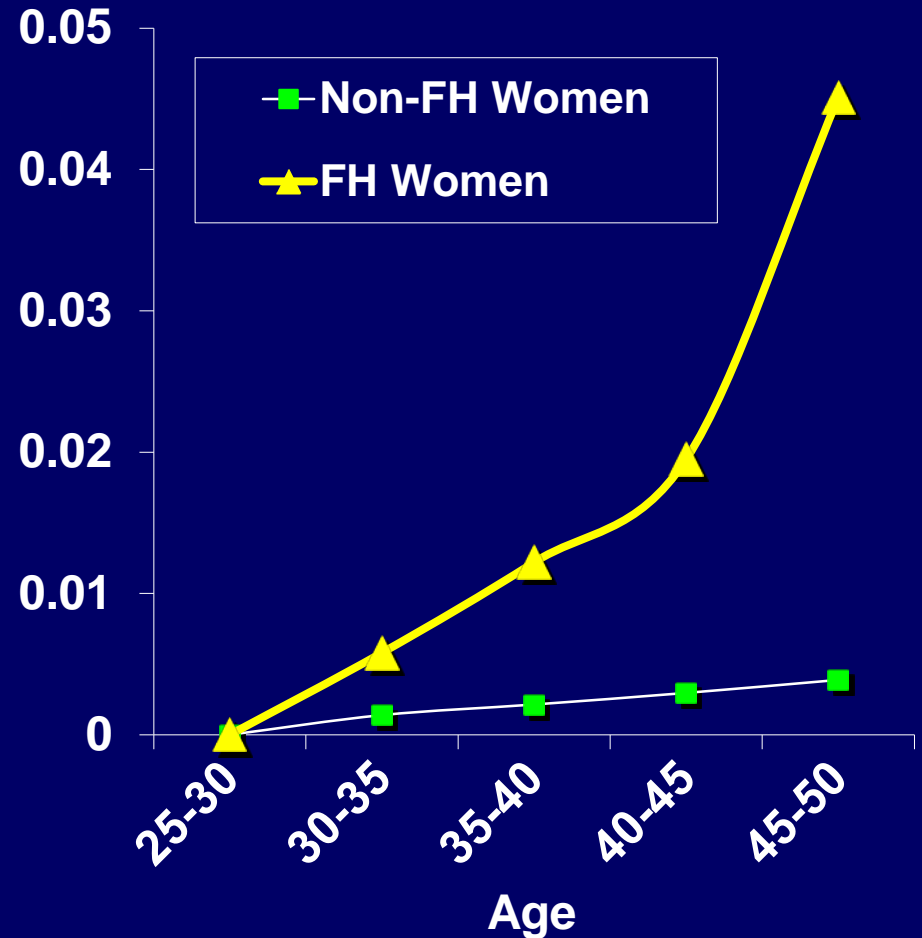
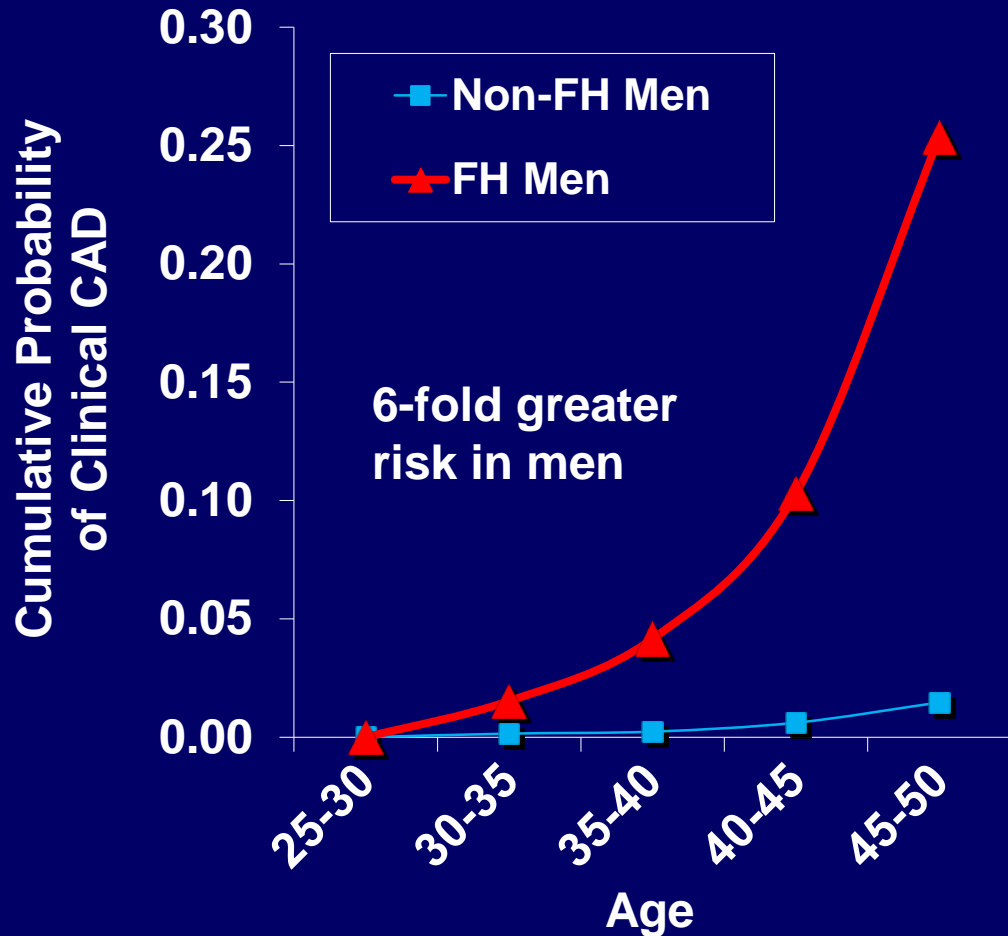


Practical LDL Criteria for Diagnosing Heterozygous FH in U.S. Population

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

	Degree of Relation to Closest FH				
Age	First	Second	Third	General	“100%”
<20	220 (155)	230 (165)	240 (170)	270 (200)	(240)
20-29	240 (170)	250 (180)	260 (185)	290 (220)	(260)
30-39	270 (190)	280 (200)	290 (210)	340 (240)	(280)
40+	290 (205)	300 (215)	310 (225)	360 (260)	(300)

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



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!