



Key Messages for Patients from the National Lipid Association Expert Panel on Familial Hypercholesterolemia



In May 2011, the National Lipid Association Expert Panel on Familial Hypercholesterolemia published an Executive Summary with FH recommendations in the *Journal of Clinical Lipidology*. In addition, the panel published five supporting articles in a supplement that accompanied the journal issue.

Please see the following slides for key messages from the panel's FH recommendations. To access the Executive Summary or supplement articles, please visit the journal website at www.lipidjournal.com or call the NLA Office at (904) 998-0853.

How Common is FH and what is the Associated Risk?

- ♥ FH is estimated to affect as many as 1 in 300 or as few as 1 in 500 people in many populations, making FH among the most common of serious genetic disorders.
- ♥ In special populations (such as French Canadians and Dutch Afrikaners), the prevalence of FH may be as high as 1 in 100.
- ♥ There are approximately 620,000 FH patients living in the United States.
- ♥ The risk of premature coronary heart disease (CHD) is elevated about 20-fold in untreated FH patients.

Genetics (inheritance) of FH

- ♥ Currently, known causes of FH include mutations or changes in a gene that codes for the LDL-receptor. The defective LDL-receptor cannot take up LDL-Cholesterol (known as “bad” cholesterol) from the blood into the cell. Excess cholesterol is deposited in blood vessel walls and sometimes in tendons, the skin, and the eyes.
- ♥ Currently, there are more than 1,600 known mutations of the LDLR gene documented to cause FH, accounting for about 85 to 90% of FH cases.



Screening for FH

- ♥ Cholesterol screening should be considered beginning at age 2 for children with a family history of premature cardiovascular disease or elevated cholesterol. All individuals should be screened by age 20.
- ♥ FH should be suspected when untreated fasting LDL cholesterol or non-HDL cholesterol levels are at or above the following:

Adults (≥ 20 years):

- I. LDL cholesterol ≥ 190 mg/dL; or
- II. non-HDL cholesterol ≥ 220 mg/dL; (*your non-HDL cholesterol measurement is calculated as total cholesterol minus HDL cholesterol*)

Children, adolescents and young adults (≤ 20 years):

- I. LDL cholesterol ≥ 160 mg/dL; or
- II. non-HDL cholesterol ≥ 190 mg/dL.

Screening for FH (continued)

- ♥ For all individuals with these levels, a family history of high cholesterol and heart disease in first-degree relatives should be collected. The likelihood of FH is higher in individuals who have a family history of high cholesterol or of premature CHD (onset in men before age 55 years and women before age 65 years).

Diagnosis of FH

- ♥ A physician or lipid specialist can make a diagnosis for FH.
- ♥ The physician or lipid specialist may notice physical symptoms of FH including xanthomas (lumps caused by deposits of cholesterol within tendons) on the Achilles tendon, the knuckles and the elbows. Yellow deposits in the skin around the eyes and/or a white deposit of cholesterol in the shape of a circle (at 6 and 12 o'clock) may also be seen at the edge of the colored part of the eye.
- ♥ Importantly, the absence of these physical findings does not rule out FH.
- ♥ Diagnosis of FH can be made by a lipid specialist applying a set of criteria [i.e., U.S. Make Early Diagnosis Prevent Early Death (MEDPED), Dutch Lipid Clinic Network, Simon-Broome Registry].

Treatment

Individuals with FH have a very high lifetime risk of CHD.

- ♥ FH requires lifelong treatment and regular follow-up.
- ♥ Early treatment is highly beneficial and critical to delay the premature onset of CHD.
- ♥ Children and pregnant women with FH should seek specific advice regarding their treatment regimens from their physician.



Risk Factors

- ♥ Other risk factors for FH are the same as in the general population.
- ♥ To minimize their risk, patients with FH must pay special attention to smoking cessation and lifestyle management, including regular physical activity, a healthy diet and weight control.

Public Awareness

- ♥ Each person should be aware of his/her cholesterol health and risk for coronary heart disease (CHD).
- ♥ For more information:
 - I. Patients can visit the website www.learnyourlipids.com;
 - II. Practitioners can visit the NLA website, www.lipid.org; and
 - III. The Executive Summary on FH may be obtained via the *Journal of Clinical Lipidology* website at www.lipidjournal.com.