

WHAT TO DO

IF YOU SUSPECT THAT YOUR PATIENT HAS FH

Inform patients



- ▶ Inform your patient that they have a clinical diagnosis of FH.
- ▶ Provide them with information about FH, its treatment and its management. You may find some of the FHCARE educational materials helpful for this conversation. (scan the QR code to visit website)



Treat the condition



Treatment options include:

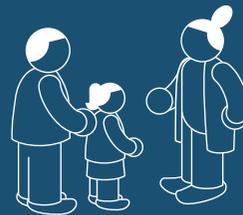
- 1 Lifestyle changes**
- Exercise
 - Eating a healthy low-fat diet

- 2 Medication, such as**
- Statins
 - Dietary cholesterol absorption inhibitors
 - Injectable medications that help the liver absorb more LDL-C

Recommend family cascade screening

Screening family members for FH is an effective way of identifying people with FH and treating them to prevent coronary heart disease. These individuals may otherwise go undetected.

- ▶ Explain to your patient that their family members are at higher risk for FH and heart disease
- ▶ Encourage your patient to ask their family members to get screened for FH



KEY FACTS ABOUT FH

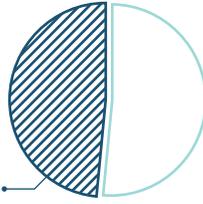
An estimated **1 in 250** people have FH¹  



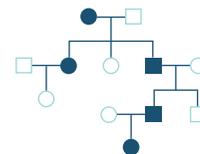

This means there are about 20,000 affected in Singapore.

In most countries, only **<10%** of people with FH receive a diagnosis²

 If untreated, people with FH have up to **20X higher risk** of early heart disease compared with those without FH³

Only **48%**  of people who know they have FH are on medication⁴

FH is easily detected: cascade screening of family members effectively identifies individuals at risk⁵



FH is readily treated: statins, for example, can reduce the risk of coronary heart disease in FH patients by **80 percent**⁶

Materials developed by:



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1 Nordestgaard BG, et al. *Eur Heart J*. 2017;38:1580-1583. 4 Benn M, et al. *J Clin Endocrinol Metab*. 2012;97:3956-3964.
2 Nordestgaard BG, et al. *Eur Heart J*. 2013;34:3478-3490. 5 Ademi Z, et al. *J Clin Lipidol*. 2014;8:390-400.
3 Goldberg AC, et al. *J Clin Lipidol*. 2011;5:S1-S8. 6 Versmissen J, et al. *BMJ*. 2008;337:a2423.



For healthcare professionals

DETECTION AND MANAGEMENT OF FAMILIAL HYPERCHOLESTEROLEMIA

Why screening and early detection are key for protecting your patients and their families

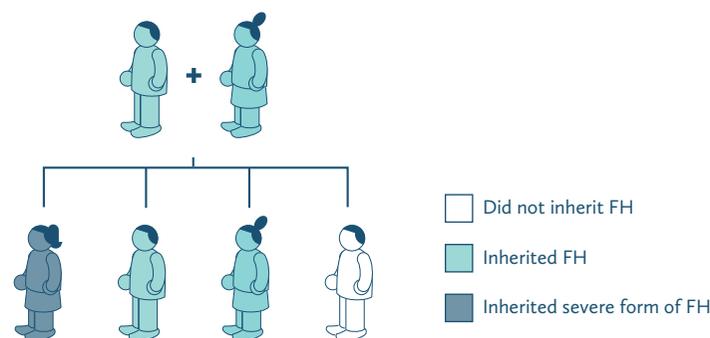


WHAT IS FAMILIAL HYPERCHOLESTEROLEMIA (FH)?

- ▶ Familial Hypercholesterolemia (FH) is a genetic disorder that causes high blood cholesterol levels.
- ▶ These patients are unable to metabolize excess low density lipoprotein cholesterol (LDL-C).
- ▶ If untreated, FH can lead to early heart attack and heart disease (<age 55 in men; <age 60 in women).¹

HOW IS FH INHERITED?

- ▶ FH can be caused by mutations in any of three main genes associated with the condition: *APOB*, *LDLR* and *PCSK9*.²
- ▶ The condition is autosomal dominant, meaning that just one copy of the disease variant is required.
- ▶ Parents, siblings and children of FH patients will have a 50% chance of also having the disease. If both parents have FH, there is a 25% chance that the child will have a more severe form of FH.



FH IS SEVERELY UNDERDIAGNOSED AND UNDERTREATED

- ▶ In most countries, < 10% of people with FH are diagnosed.³
- ▶ Many patients do not seek care until they have their first heart attack.

BUT FH IS EASILY DIAGNOSED AND READILY TREATED

There are currently three accepted diagnostic criteria for FH

- ▶ **MEDPED (US)**
- ▶ **Simon Broome (UK)**
- ▶ **Dutch Lipid Clinic Network (The Netherlands)**
The Dutch Lipid criteria has been shown to correlate well with genetic mutation status.^{4,5}

The following are considered in establishing a diagnosis of FH:

- ▶ **High blood cholesterol**
 - Total cholesterol of > 7.5mmol/L or 290mg/dL
 - LDL-cholesterol of > 4.9mmol/L or 190mg/dL (18% of people with LDL-C \geq 190mg/dL have FH)⁶
- ▶ **Physical signs** (e.g. tendon xanthomas)
- ▶ **Family history of high cholesterol or early heart attack**
- ▶ **Genetic testing** (detection of FH-causing mutation)



Lifestyle changes and medication can significantly reduce the risk of coronary heart disease in FH patients.

Scan QR code for more information about using the diagnostic criteria.

You can choose to diagnose and manage your own patients with FH.

Resources (see FHCARE Resources) are also available to support you in the diagnosis and treatment of your patients.

1 Goldberg AC, et al. *J Clin Lipidol*. 2011;5:S1-S8.

2 Berberich AJ, Hegele RA. *Nat Rev Cardiol*. 2019;16:9-20.

3 Nordestgaard BG, et al. *Eur Heart J*. 2013;34:3478-3490.

4 Palacios L, et al. *Atherosclerosis*. 2012;221:137-142.

5 Mickiewicz A, et al. *Atherosclerosis*. 2016;249:52-58.

6 Benn M, et al. *J Clin Endo Metab*. 2012;97:3956-3964.

FHCARE RESOURCES

If you think your patient has FH, the FHCARE team (a team of healthcare professionals) can

- ▶ Provide advice on diagnosis and treatment
- ▶ Confirm the diagnosis and perform genetic testing to determine the specific FH mutation
- ▶ Enrol your patient in our FH registry, which stores medical and genetic data and can help advance research in the disease
- ▶ Perform cascade screening of your patient's family members

How to refer your patient, and what happens next:

STEP 1

You will need to obtain patient consent for referral and provide required information to FHCARE.

Contact FHCARE

By E-mail: cholesterol.info@ktph.com.sg

By Phone: (+65) 6602 2346 / 9674 5167 / 9825 9793

Website: www.myheart.org/FH



STEP 2

FHCARE team will

- ▶ Confirm the diagnosis, enrol patient in FH registry and perform genetic testing
- ▶ Send diagnosis results back to you, and work together with you on a personalized care plan for your patient.

STEP 3

You can contact FHCARE for further discussion and advice during continued treatment.