British Heart Foundation

Familial Hypercholesterolaemia Service for South Yorkshire and North Derbyshire

Alison Moore 2019

FIGHT FOR EVERY HEARTBEAT

bhf.org.uk

Why diagnose FH?

- FH is the most common inherited condition causing lifetime burden of high LDL – thus high risk premature CVD

- If left untreated, 50% of men and 30% of women will develop clinically evident CVD by the age of 55 years

Prevalence now 1:270 est~ 6666 undiagnosed cases in our area

Identifying FH

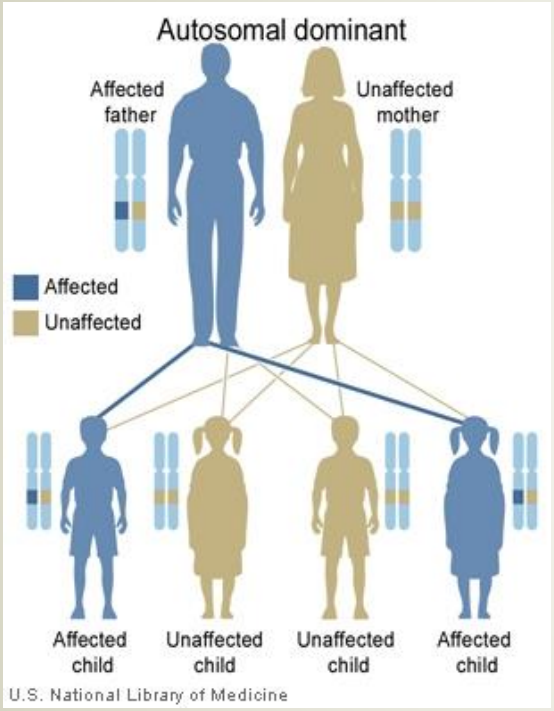
Simon Broome Criteria

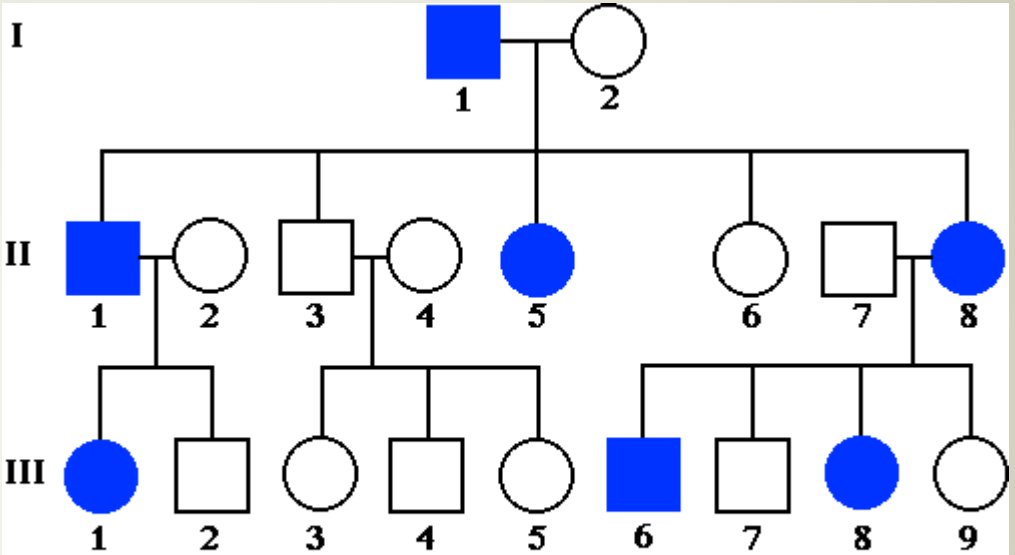
- Unexpectedly high total chol > 7.5mmol/L
- Arrange a fasted FULL lipid profile
- Raised LDL-C >4.9
- FCH more likely if trigs persistently increased
- Secondary causes excluded (hypothyroidism, nephrotic syndrome, chronic liver disease and diabetes)
- **Family history 1st & 2nd degree relatives**

  (CHD <60 years 1st degree/ CHD <50 2nd degree or TC >7.5 LDL >4.9)

- Do not perform risk assessment such as QRISK.

Dominant Inheritance





Pedigree 1. An idealized pedigree of a family with hypercholesterolemia, an autosomal dominant disease where the heterozygote has a reduced number of functional low density lipoprotein receptors.

Physical signs:

Tendon Xanthoma

Deposit of Cholesterol on the Tendons, can manifest as Achilles tendon thickening

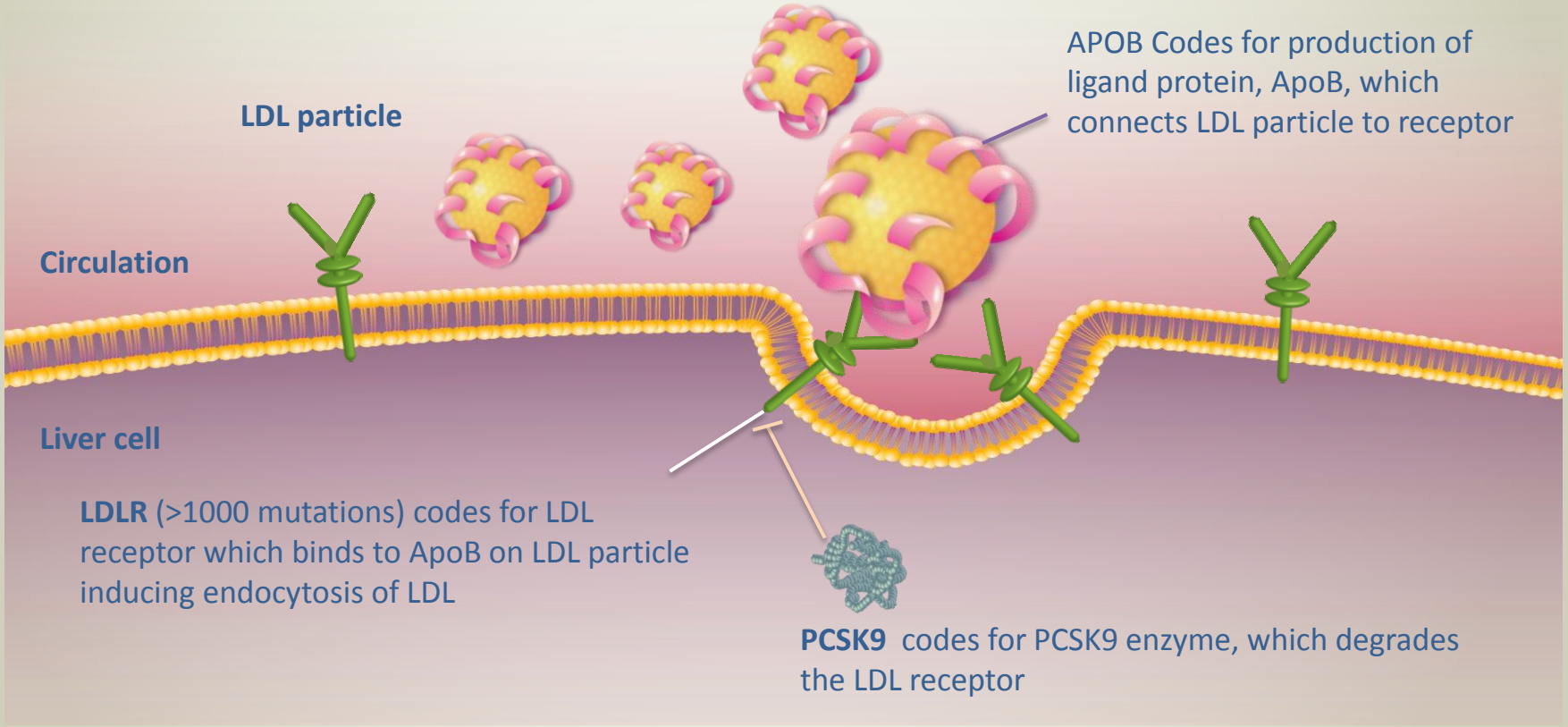
What to look for….

Corneal Arcus
Must be premature
<45 years

Xanthelasma
Common in mixed hypercholesterolaemia
not specific to FH

LDL Clearance

1000+ Mutations Identified to Date

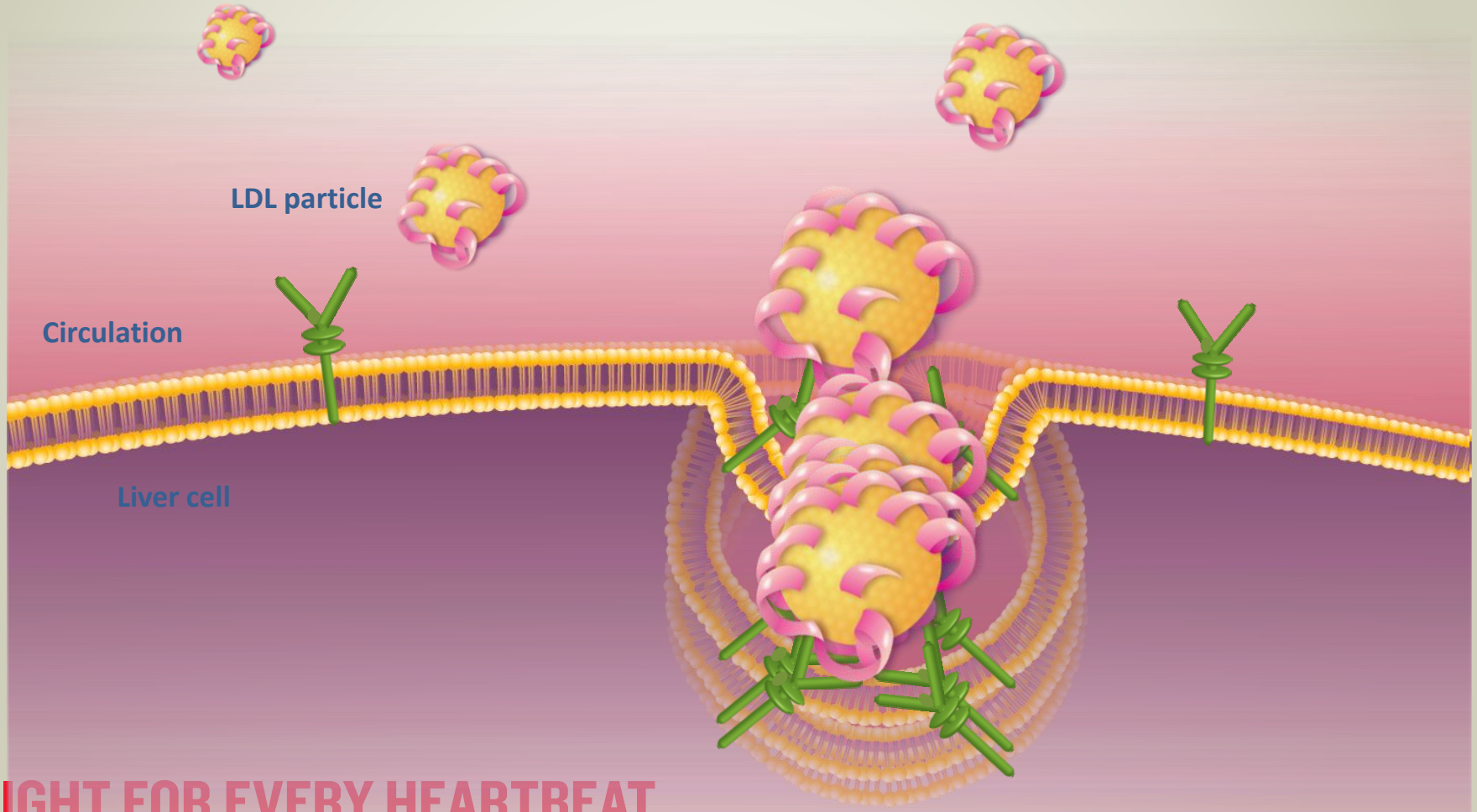


FIGHT FOR EVERY HEARTBEAT

bhf.org.uk

Rader DJ, et al. *J Clin Invest.* 2003;111:1795-1803.

Normally, LDL Receptors on hepatocytes help to clear LDL Particles from plasma

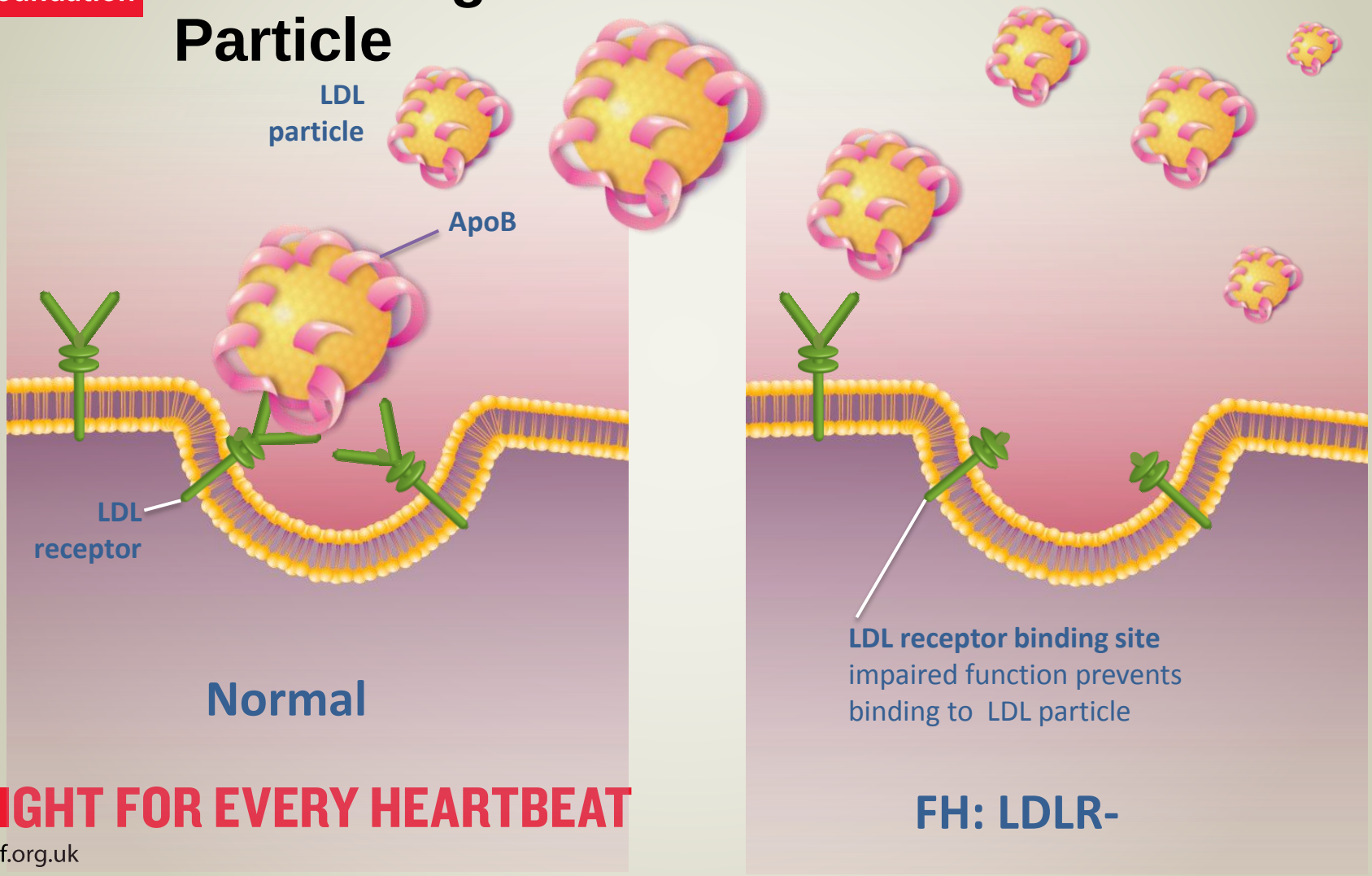


FIGHT FOR EVERY HEARTBEAT

bhf.org.uk

Rader DJ, et al. *J Clin Invest.* 2003;111:1795-1803.

LDL Receptor (LDLR) Mutations Alter LDL Receptors, Preventing Attachment to Particle



FIGHT FOR EVERY HEARTBEAT

bhf.org.uk

Rader DJ, et al. *J Clin Invest.* 2003;111:1795-1803.

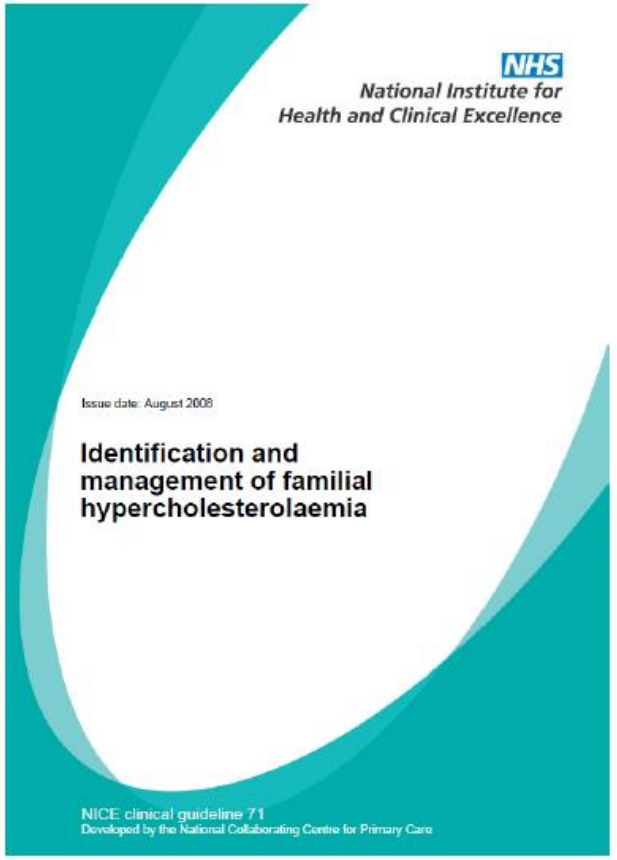
Familial hypercholesterolaemia

NICE CG71, 2008

Issued: August 2013

NICE quality standard 41

guidance.nice.org.uk/qs41



Specialist referral

See FH pathway

Drug treatment in adults

AIM to reduce LDL-C by 50% and beyond (EAS 2.5mmol/1.8 CHD)

- Initiate high intensity statin atorvastatin/rosuvastatin/ezetimibe
- Avoid during pregnancy
- PCSK9 inhibitors injections
- Annual review

Drug treatment in children

Consider statin therapy by the age 10.
Paediatric referral to SCH

Cascade screening

Relatives of people with a confirmed diagnosis of monogenic FH are offered DNA testing through a nationwide, systematic cascade process- PASS database

Take home messages

- FH = high risk of CVD and is under diagnosed
- Consider FH if TC>7.5 in adults
- DO NOT use Qrisk for these patients
- They need treatment to achieve at least 50% LDL reduction (aim <2.5)
- FHICC will organise cascade testing
- FH patients need annual review
- Barnsley FH clinic