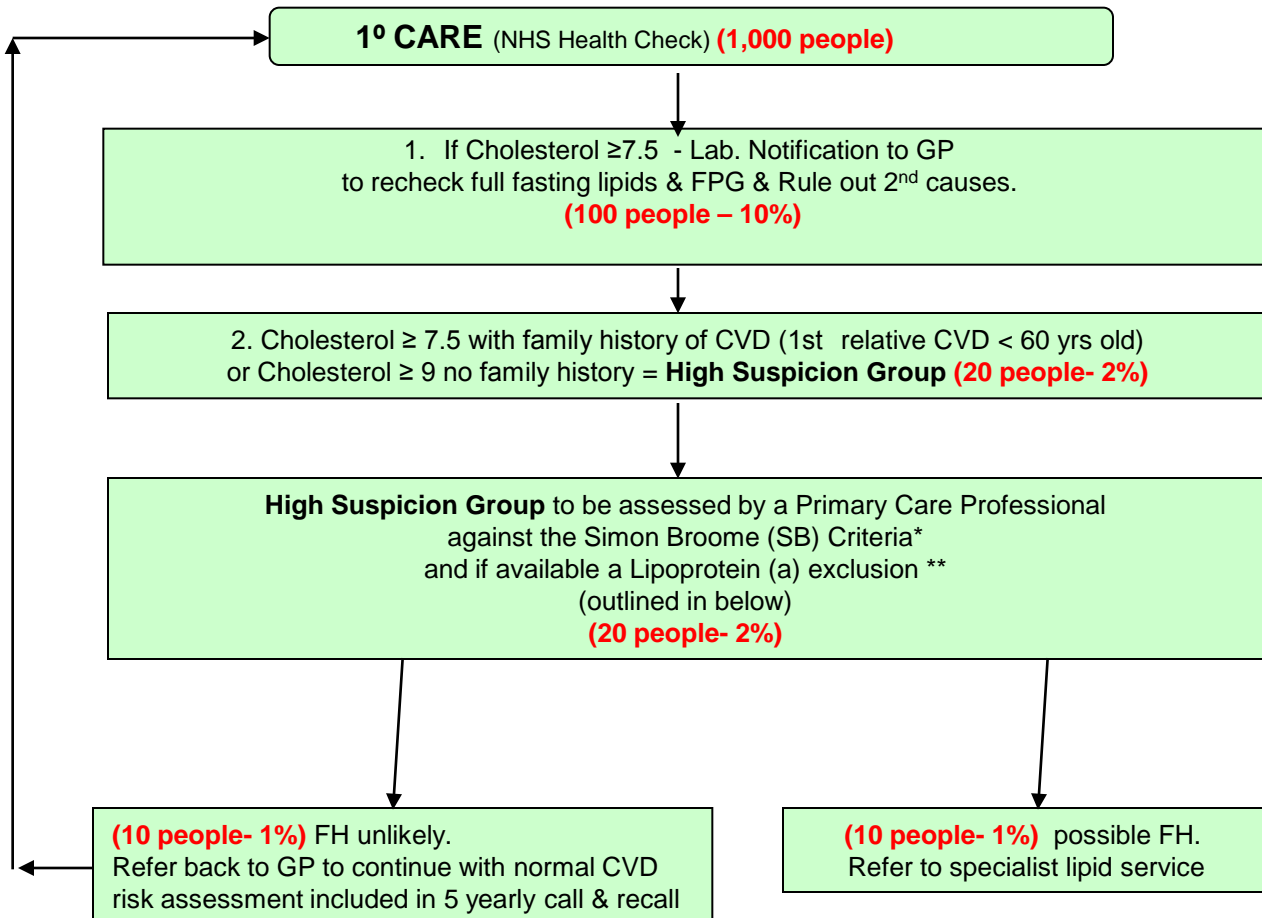


Familial Hypercholesterolaemia Pathway for Primary Care Assessment (Adults)



Simon Broome diagnostic criteria for index individuals

Diagnose a person with **definite** FH if they have:

- Adult total cholesterol > 7.5 mmol /L
- **and** tendon xanthomas (not arcus or xanthelasma) , **or**
- DNA-based evidence of a LDL-receptor, apo B-100, or a PCSK-9 mutation.

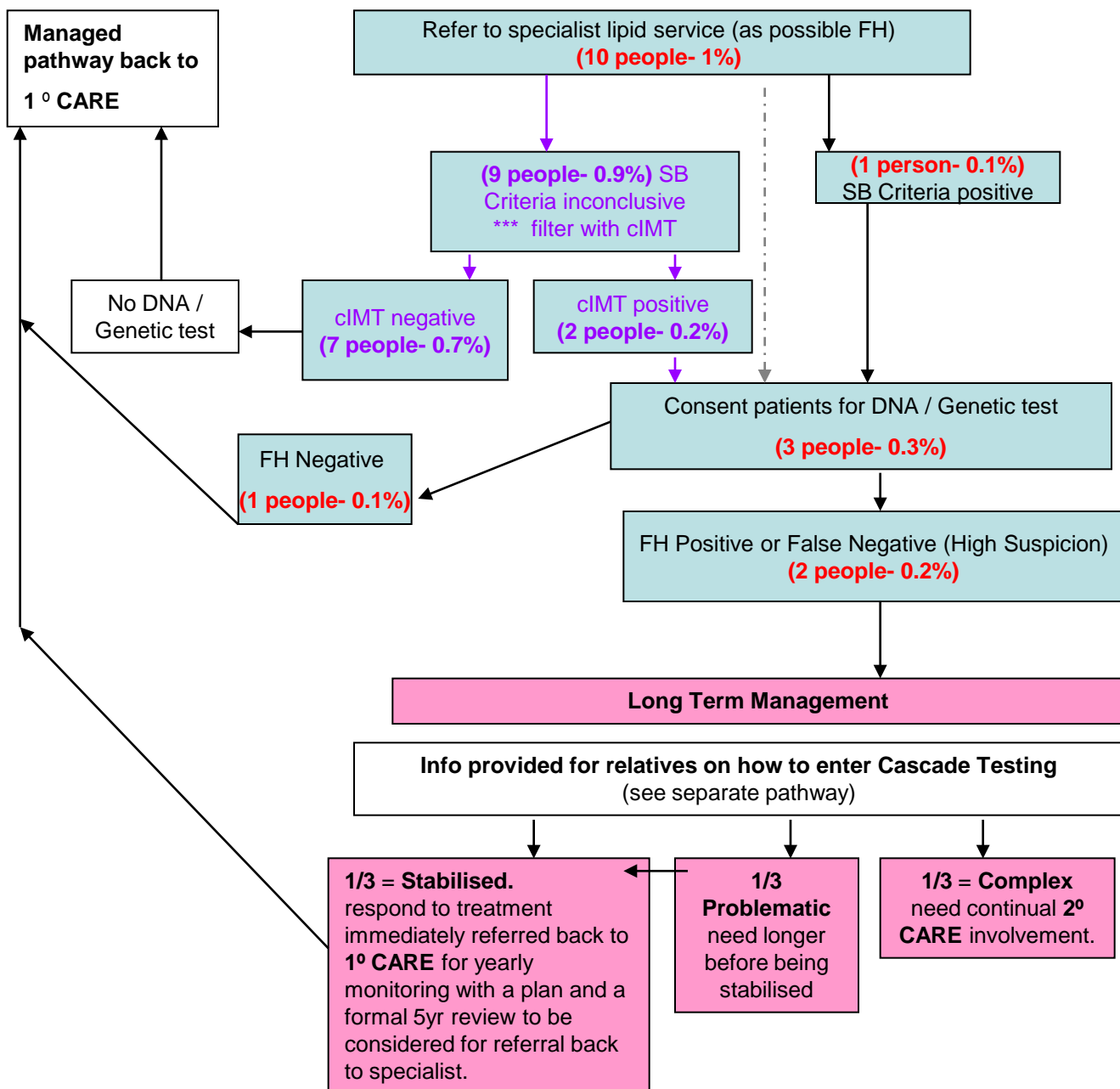
Diagnose a person with **possible** FH if they have:

- adult total cholesterol > 7.5 mmol /L **and** at least one of the following;
- Family history of coronary heart disease : age < 60 years in 1st degree relative **or** age < 50 years in 2nd degree relative **or**
- Family history of total cholesterol > 7.5 mmol/l in adult 1st- or 2nd -degree relative (> 6.7 mmol/l in child < 16 years).
- * Pg 7 - NICE Quick reference guide - Familial hypercholesterolaemia: August 2008

** Lipoprotein (a) tests to track family heart disease:

- Raised Lp(a) > 0.3 g/L associated with increased inherited risk of CHD, CVA or Peripheral arterial disease (PAD)
- Prognostic factor in patients with FH

Familial Hypercholesterolaemia Pathway in Secondary Care Assessment & Long Term Management (Adults)

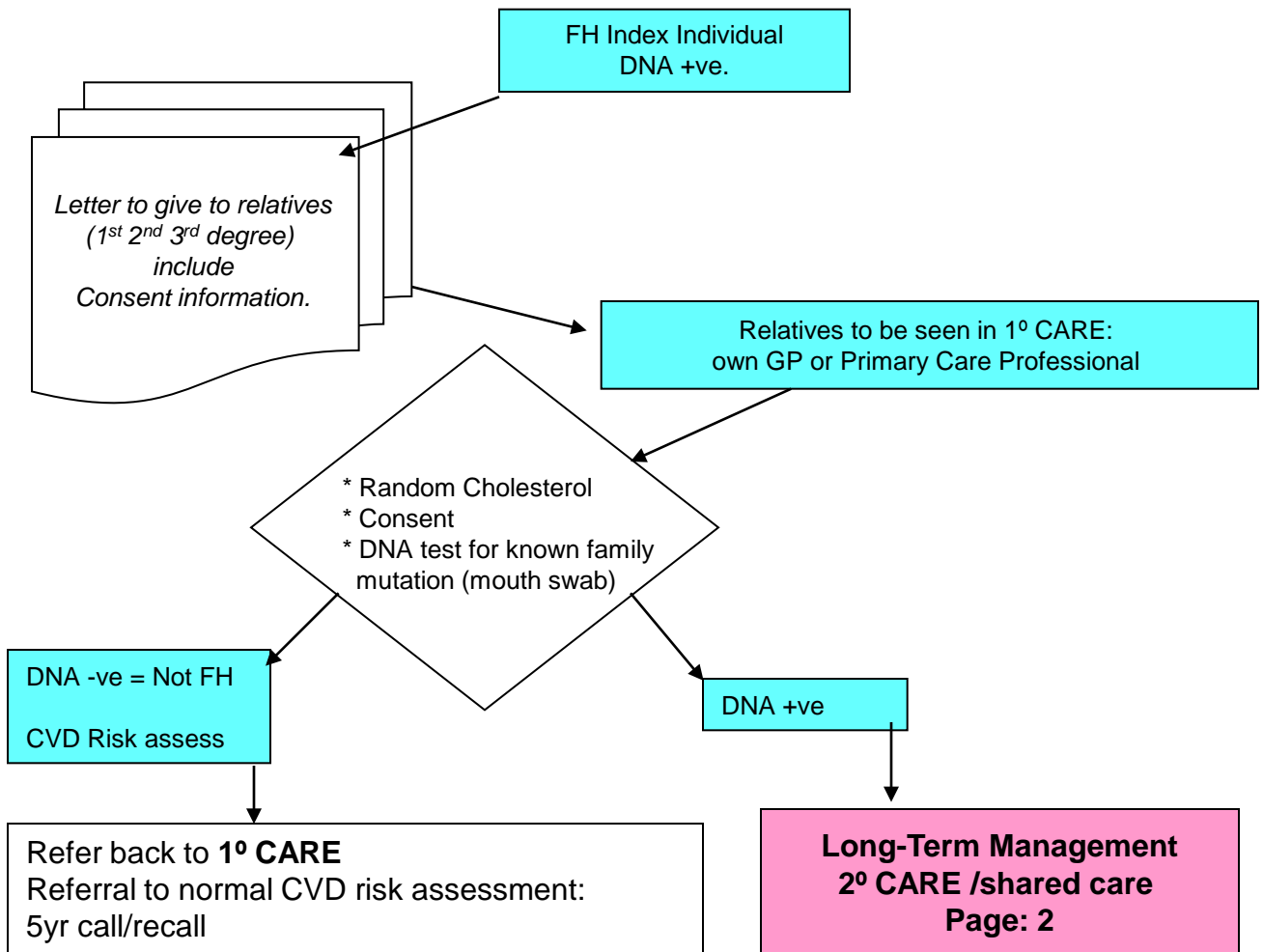


*** Ultrasound carotid intima-media thickness (c-IMT) screening out (2° CARE or poly clinic stage)
- Non-invasive test to assess burden of atherosclerosis
- Reported as age & sex specific centile: >75th centile is assumed high risk. Most FH patients >90th centile.

NB. Need to develop a Shared Care Register of FH (kept in 2° CARE)

Familial Hypercholesterolaemia Cascade Testing Pathway for Primary Care Assessment & Secondary Care Long Term Management (Adults)

Cascade Testing Pathway



Paediatric (children <16 years)

When an index individual has children <16 years related to them, the testing and long term management should be carried out in a suitable Paediatric setting with transition protocol to adult services.

Paediatric Lipid Clinics in South London (May 2010):

St George's Service: Consultant Chemical Pathology, St George's Hospital, Blackshaw Road, London SW17 0QT
Dept: 0208 725 5923

South London Service: Paediatric Lipid Clinic, Department of Inherited Metabolic Disease, Level 6, Evelina Children's Hospital, St Thomas' Hospital, Westminster Bridge Road, London SE1 7EH.

North London Service: Paediatric lipids & cardiology, Cardiothoracic Unit, Great Ormond Street Hospital, Great Ormond Street, London WC1N 3JH Tel: 020 7404 5094

Family screening for Familial Hypercholesterolaemia

Total cholesterol cut-offs tables for women & men

women

Age					
0 To 14	15 To 24	25 To 34	35 To 44	45 To 54	55 and Older
8.0	8.0	8.0	8.0	8.0	8.0
7.9	7.9	7.9	7.9	7.9	7.9
7.8	7.8	7.8	7.8	7.8	7.8
7.7	7.7	7.7	7.7	7.7	7.7
7.6	7.6	7.6	7.6	7.6	7.6
7.5	7.5	7.5	7.5	7.5	7.5
7.4	7.4	7.4	7.4	7.4	7.4
7.3	7.3	7.3	7.3	7.3	7.3
7.2	7.2	7.2	7.2	7.2	7.2
7.1	7.1	7.1	7.1	7.1	7.1
7.0	7.0	7.0	7.0	7.0	7.0
6.9	6.9	6.9	6.9	6.9	6.9
6.8	6.8	6.8	6.8	6.8	6.8
6.7	6.7	6.7	6.7	6.7	6.7
6.6	6.6	6.6	6.6	6.6	6.6
6.5	6.5	6.5	6.5	6.5	6.5
6.4	6.4	6.4	6.4	6.4	6.4
6.3	6.3	6.3	6.3	6.3	6.3
6.2	6.2	6.2	6.2	6.2	6.2
6.1	6.1	6.1	6.1	6.1	6.1
6.0	6.0	6.0	6.0	6.0	6.0
5.9	5.9	5.9	5.9	5.9	5.9
5.8	5.8	5.8	5.8	5.8	5.8
5.7	5.7	5.7	5.7	5.7	5.7
5.6	5.6	5.6	5.6	5.6	5.6
5.5	5.5	5.5	5.5	5.5	5.5
5.4	5.4	5.4	5.4	5.4	5.4
5.3	5.3	5.3	5.3	5.3	5.3
5.2	5.2	5.2	5.2	5.2	5.2
5.1	5.1	5.1	5.1	5.1	5.1
5.0	5.0	5.0	5.0	5.0	5.0
4.9	4.9	4.9	4.9	4.9	4.9
4.8	4.8	4.8	4.8	4.8	4.8
4.7	4.7	4.7	4.7	4.7	4.7
4.6	4.6	4.6	4.6	4.6	4.6
4.5	4.5	4.5	4.5	4.5	4.5

men

Age					
0 To 14	15 To 24	25 To 34	35 To 44	45 To 54	55 and Older
8.0	8.0	8.0	8.0	8.0	8.0
7.9	7.9	7.9	7.9	7.9	7.9
7.8	7.8	7.8	7.8	7.8	7.8
7.7	7.7	7.7	7.7	7.7	7.7
7.6	7.6	7.6	7.6	7.6	7.6
7.5	7.5	7.5	7.5	7.5	7.5
7.4	7.4	7.4	7.4	7.4	7.4
7.3	7.3	7.3	7.3	7.3	7.3
7.2	7.2	7.2	7.2	7.2	7.2
7.1	7.1	7.1	7.1	7.1	7.1
7.0	7.0	7.0	7.0	7.0	7.0
6.9	6.9	6.9	6.9	6.9	6.9
6.8	6.8	6.8	6.8	6.8	6.8
6.7	6.7	6.7	6.7	6.7	6.7
6.6	6.6	6.6	6.6	6.6	6.6
6.5	6.5	6.5	6.5	6.5	6.5
6.4	6.4	6.4	6.4	6.4	6.4
6.3	6.3	6.3	6.3	6.3	6.3
6.2	6.2	6.2	6.2	6.2	6.2
6.1	6.1	6.1	6.1	6.1	6.1
6.0	6.0	6.0	6.0	6.0	6.0
5.9	5.9	5.9	5.9	5.9	5.9
5.8	5.8	5.8	5.8	5.8	5.8
5.7	5.7	5.7	5.7	5.7	5.7
5.6	5.6	5.6	5.6	5.6	5.6
5.5	5.5	5.5	5.5	5.5	5.5
5.4	5.4	5.4	5.4	5.4	5.4
5.3	5.3	5.3	5.3	5.3	5.3
5.2	5.2	5.2	5.2	5.2	5.2
5.1	5.1	5.1	5.1	5.1	5.1
5.0	5.0	5.0	5.0	5.0	5.0
4.9	4.9	4.9	4.9	4.9	4.9
4.8	4.8	4.8	4.8	4.8	4.8
4.7	4.7	4.7	4.7	4.7	4.7
4.6	4.6	4.6	4.6	4.6	4.6
4.5	4.5	4.5	4.5	4.5	4.5

KEY: Red= positive: Grey= uncertain: Green = negative:

Ultrasound carotid intima-media thickness (C-IMT) in patients with FH (red) and controls (blue)

De Groot et al : Circulation 2004; 109 suppl 3: 33-8

