

Appendix F
Diagnostic criteria
for
probands (Simon Broome)
and relatives

1 Simon Broome diagnostic criteria for probands

Definite familial hypercholesterolaemia is defined as:

- total cholesterol above 6.7mmol/l or LDL cholesterol above 4.0mmol/l in a child aged under 16 years or
total cholesterol above 7.5mmol/l or LDL cholesterol above 4.9mmol/l in an adult (levels either pre- treatment or highest on treatment)

plus

- tendon xanthomas in patient, or in 1st degree relative (parent, sibling, child), or in 2nd degree relative (grandparent, uncle, aunt)

or

- DNA-based evidence of an LDL receptor mutation, familial defective apo B-100, or a *PCSK9* mutation.

Possible familial hypercholesterolaemia is defined as above plus one of the criteria below:

- family history of myocardial infarction: below age of 50 years in 2nd degree relative or below age 60 years in 1st degree relative
- family history of raised total cholesterol: above 7.5mmol/l in adult 1st or 2nd degree relative or above 6.7mmol/l in child or sibling aged under 16 years.

2 Diagnostic criteria for relatives

2.1 LDL diagnostic tables

LDL-C diagnostic table for first degree relatives					
0 To 14	15 To 24	25 To 34	35 To 44	45 To 54	55 and Older
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
4.4	4.4	4.4	4.4	4.4	4.4
4.3	4.3	4.3	4.3	4.3	4.3
4.2	4.2	4.2	4.2	4.2	4.2
4.1	4.1	4.1	4.1	4.1	4.1
4.0	4.0	4.0	4.0	4.0	4.0
3.9	3.9	3.9	3.9	3.9	3.9
3.8	3.8	3.8	3.8	3.8	3.8
3.7	3.7	3.7	3.7	3.7	3.7
3.6	3.6	3.6	3.6	3.6	3.6
3.5	3.5	3.5	3.5	3.5	3.5
3.4	3.4	3.4	3.4	3.4	3.4
3.3	3.3	3.3	3.3	3.3	3.3
3.2	3.2	3.2	3.2	3.2	3.2
3.1	3.1	3.1	3.1	3.1	3.1
3.0	3.0	3.0	3.0	3.0	3.0

Red = Likely FH

Grey = Uncertain

Green = Unlikely FH

2.2 TC diagnostic tables

TC diagnostic table for first degree relatives					
0 To 14	15 To 24	25 To 34	35 To 44	45 To 54	55 and Older
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

Red = Likely FH

Grey = Uncertain

Green = Unlikely FH