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.

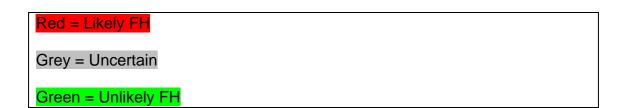
Possible familial hypercholesterolaemia is defined <u>as above plus one</u> of the criteria below:

- 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			



Familial hypercholesterolaemia: full guideline DRAFT (February 2008) Appendix F: Diagnostic criteria for probands (Simon Broome) and relatives



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		



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