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Improving Identification of Familial Hypercholesterolaemia (FH) in English Family Practice using Electronic Medical Records

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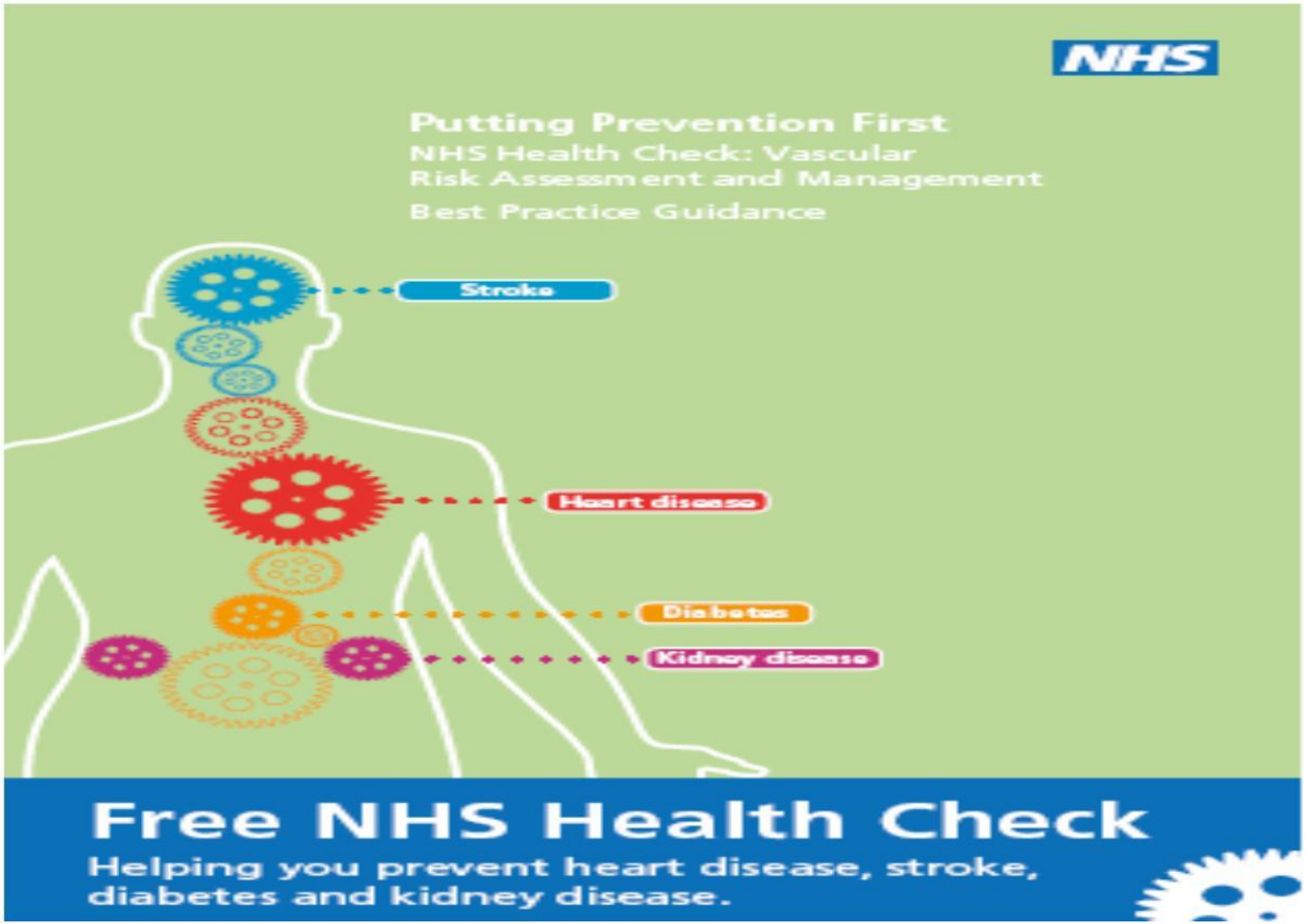
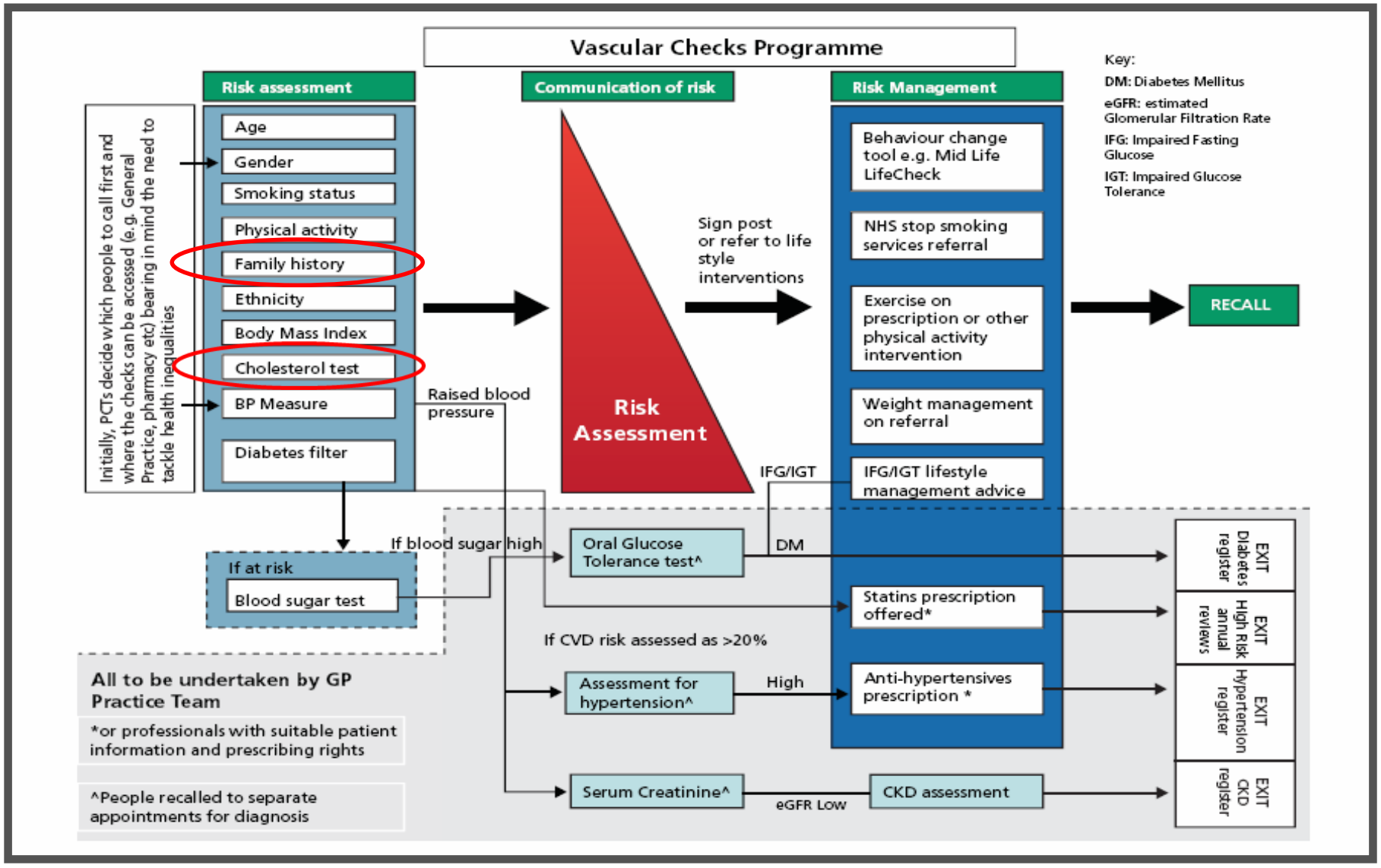
Opportunities in Primary Care Role

- Integrate with national policy
- Computer records
  - Patient-specific reminders
  - Audit & Feedback
  - Software toolkits

National Health Checks 2009




Vascular Checks Programme
Risk assessment
Communication of risk
Risk Management
Key:
DM: Diabetes Mellitus
eGFR: estimated Glomerular Filtration Rate
IFG: Impaired Fasting Glucose
IGT: Impaired Glucose Tolerance
Initially, PCTs decide which people to call first and where the checks can be accessed (e.g. General Practice, pharmacy etc) bearing in mind the need to tackle health inequalities
Age
Gender
Smoking status
Physical activity
Family history
Ethnicity
Body Mass Index
Cholesterol test
BP Measure
Diabetes filter
Raised blood pressure
Risk Assessment
Sign post or refer to life style interventions
Behaviour change tool e.g. Mid Life LifeCheck
NHS stop smoking services referral
Exercise on prescription or other physical activity intervention
Weight management on referral
IFG/IGT lifestyle management advice
IFG/IGT
RECALL
If at risk
Blood sugar test
If blood sugar high
Oral Glucose Tolerance test^
DM
Statins prescription offered*
If CVD risk assessed as >20%
Assessment for hypertension^
High
Anti-hypertensives prescription *
Serum Creatinine^
eGFR Low
CKD assessment
EXIT Diabetes register
EXIT High Risk annual reviews
EXIT Hypertension register
EXIT CKD register
All to be undertaken by GP Practice Team
*or professionals with suitable patient information and prescribing rights
^People recalled to separate appointments for diagnosis

Threshold: There is no specific threshold for the vascular risk assessment and management programme or for primary prevention of vascular disease. However, if an individual's total cholesterol is **>7.5 mmol/l** it is important to consider familial hypercholesterolaemia – a genetic condition that causes a high cholesterol concentration in the blood – as set out in the NICE clinical guideline 71.

NHS
National Institute for
Health and Clinical Excellence

Familial hypercholesterolaemia

Implementing NICE guidance

2008

Clinical Guideline Implementation Tools

NICE clinical guideline 71

Identification of FH:

Simon Broome diagnostic criteria (adults)

Dx for possible FH:

- cholesterol > 7.5 mmol/l (LDL 4.9 mmol/l) ***Computer Search***

& at least one of the following:

- **FHx of MI** < 50 years in second-degree relative, or < 60 years in first-degree relative

OR

- **FHx of raised total cholesterol > 7.5 mmol/l** (LDL 4.9 mmol/l*) in adult first or second- degree relative OR > 6.7 mmol/l in child, brother or sister aged < 16 years.

Family History Questionnaire

t

Marks et al (2003) A review on the diagnosis, natural history, and treatment of familial hypercholesterolaemia. Atherosclerosis 168 (1): 1–14.
NICE clinical guideline 71 – Identification and management of familial hypercholesterolaemia

Identification and management familial hypercholesterolaemia: what does it mean to primary care?

Nadeem Qureshi, Steve E Humphries, Mary Seed, Philip Rowlands and Rubin Minhas, on behalf of the NICE Guideline Development Group

ABSTRACT

Familial hypercholesterolaemia is one of the most common dominantly inherited disorders to be identified in primary care, leading to raised serum cholesterol evident from the first year of life. Around 1 in 500 people are affected by this condition, but less than 15% of these are currently attending lipid clinics, suggesting that the vast majority are unrecognised in general practice. The recently released National Institute for Health and Clinical Excellence evidence-based guideline on the identification and management of familial hypercholesterolaemia provides an opportunity to bridge this gap. Primary care has a role in systematic and opportunistic case finding, such as recognising the relevance of a family history of

INTRODUCTION

This review summarises the recommendations that have an impact on primary care from the National Institute for Health and Clinical Excellence (NICE) guideline on the identification and management of familial hypercholesterolaemia.[1] Familial hypercholesterolaemia is an inherited disorder leading to raised serum cholesterol evident from the first year of life. This may present with signs indicative of raised cholesterol levels such as tendon xanthomata, and, if untreated, the development of premature coronary heart disease (CHD). The disorder has an autosomal dominant mode of

NICE Quality Standard for FH

Statement 1. Adults with a baseline total cholesterol above 7.5 mmol/l are **assessed for a clinical diagnosis** of familial hypercholesterolaemia (FH).

Statement 2. People with a clinical diagnosis of familial hypercholesterolaemia (FH) are **referred for specialist assessment.**

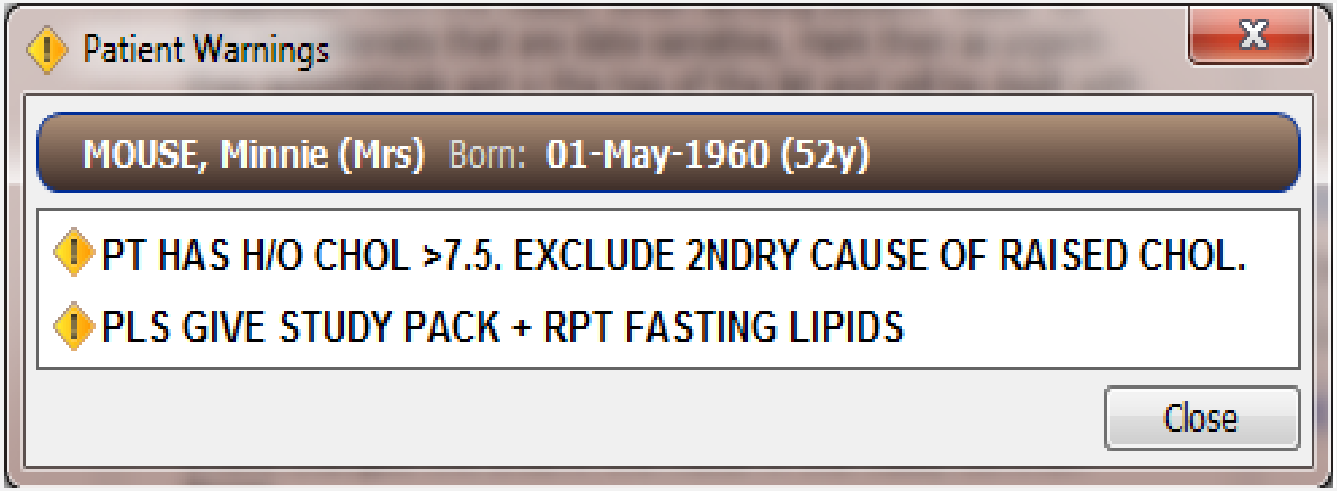
How can identification of FH be enhanced in general practice?

FAMCHOL Study

Objectives:

- Assess the feasibility of incorporating computerised patient specific reminders (PSRs) for FH if Chol > 7.5
- Evaluate the recruitment & retention of participants

Patient specific reminders



FAMCHOL study

Feasibility of improving identification of familial hypercholesterolaemia in general practice: intervention development study

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No. of eligible patients aged > 18 years: **31377**

No. with cholesterol > 7.5 mmol/l: **927**

No. participate in the study: **124** (13%)

No. of possible FH identified: **27** (3%)

Unclear if possible FH, due to lack of information on family history: **16**

FAMCHOL study

Feasibility of improving identification of familial hypercholesterolaemia in general practice: intervention development study

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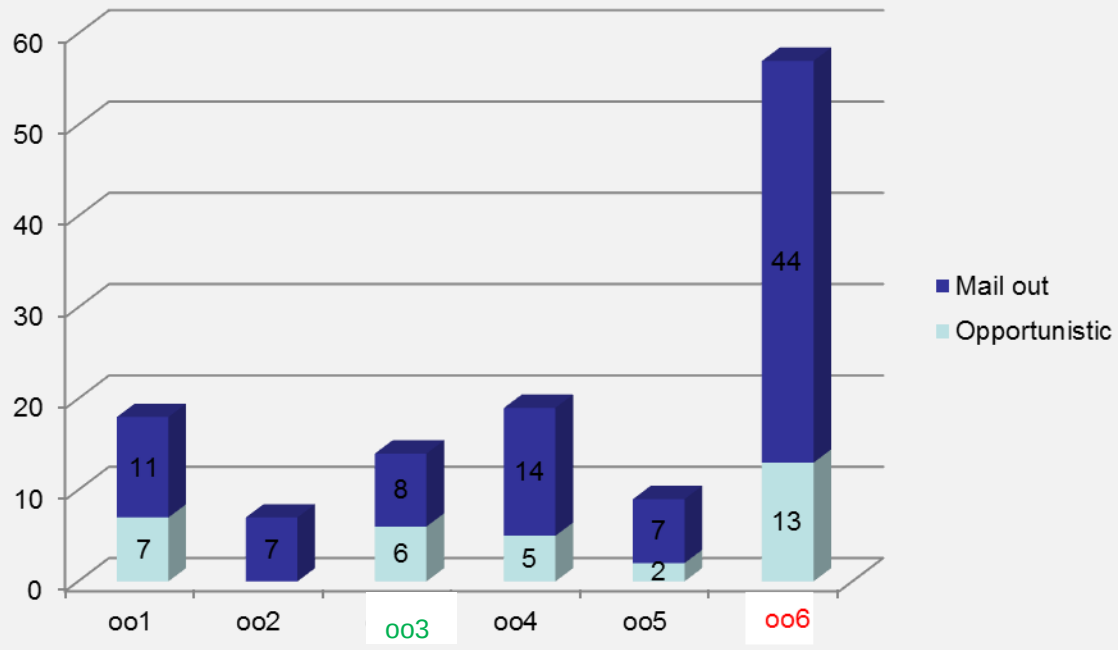
Practice recruitment



Practice geographical area

001 – Inner city
002 – Inner city
003 – Suburban
004 – Inner city
005 – Inner city
006 - Rural

Total recruitment	
Opportunistic	Mail out
33 (14%)	91(10%)
Total = 124 (11%)	

Familial Hypercholesterolaemia CHART Audit

Total no. of eligible patients in Practice	2932

No. of patients with Total Cholesterol > 7.5 ever	54
No. of those patients with Primary Cause recorded	11
No. of those patients with Secondary Cause recorded	15
No. of those patients with Possible Drug Cause recorded	15
No. of those patients with Thyroid or Diabetes Rx recorded	14
No. of those patients whose most recent cholesterol before 1/3/2013 < 4	5
No. of those patients with no primary exclusions	43

Potential participants	
No. of patients recruited	19
No. of patients with Latest Total Cholesterol > 7.5	9
No. of patients with Total Cholesterol in last 6 months	10
No. of patients with TSH recorded	20
No. of patients with Dietary Advice recorded	5
No. of patients with Weight Management advice recorded	0
No. of patients with GPPAQ exercise advice recorded	6
No. of patients with Smoking Cessation advice recorded	14
No. of patients with Statin prescribed	14

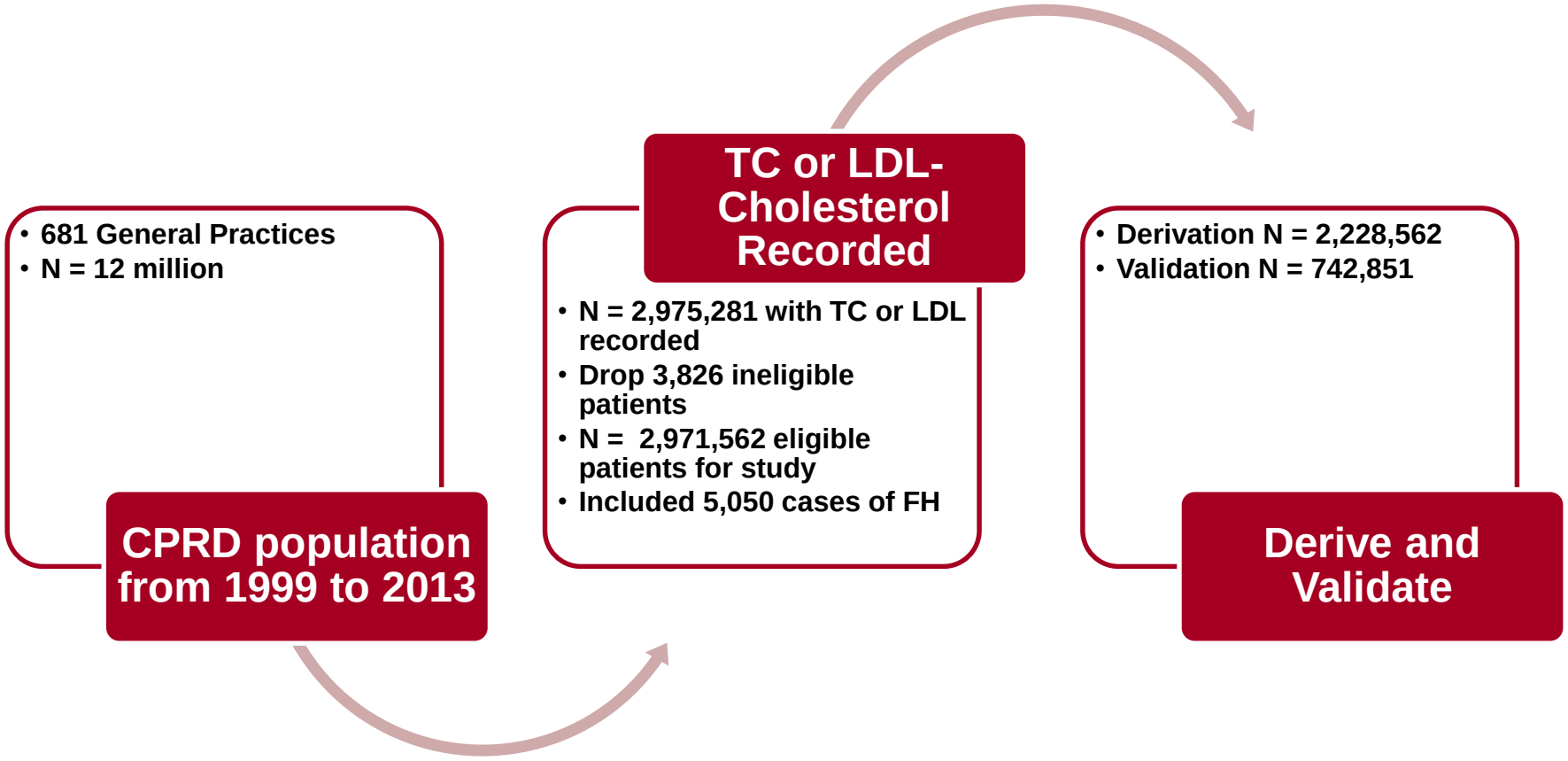
Improving Identification of Familial Hypercholesterolemia in General Practice Computer Medical Records

Rationale

(1) Current tools lack sensitivity and specificity → Results in both misdiagnosis and under-diagnosis → Misses 93,500 to 102,000 cases of FH who could benefit from lipid lowering treatment to reduce morbidity/mortality related to coronary heart disease

(2) From feasibility study large number of case identified using Serum Cholesterol > 7.5 filter: ? rationalise

Methodology



Diagnostic Variables	Adjusted Odds Ratio (AOR)	95% Confidence Interval	
		Lower Confidence Limit	Upper Confidence Limit
Gender			
Male	Ref	--	--
Female	1.24	1.16	1.33
Highest TC or LDL recorded (mmol/L)			
Ideal (TC ≤ 5 OR LDL ≤ 3.3)	Ref	--	--
High (TC > 5 to ≤ 6.5 OR LDL > 3.3 to ≤ 4.1)	2.60	2.26	3.00
Very High (TC > 6.5 to ≤ 7.5 OR LDL > 4.1 to ≤ 4.9)	8.29	7.17	9.59
Extremely High (TC > 7.5 OR LDL > 4.9)	42.74	37.24	49.07
Age during cholesterol measurement (years)	0.95	0.94	0.96
Triglycerides during cholesterol measurement (mmol/L)			
Ideal (< 1.7)	Ref	--	--
Borderline High (≥ 1.7 to < 2.3)	0.96	0.88	1.05
High (≥ 2.3 to < 5.6)	0.83	0.76	0.91
Very High (≥ 5.6)	0.68	0.57	0.82
Not Recorded	0.43	0.38	0.50
Lipid lowering drug usage during cholesterol measurement			
No lipid lowering drugs prescribed	Ref	--	--
Prescribed fibrate, bile acid sequestrant, or nicotinic acid	4.51	3.51	5.80
Prescribed low potency statins[1]	2.67	2.11	3.37
Prescribed medium potency statin[2]	3.98	3.53	4.48
Prescribed high potency statins[3]	8.30	7.31	9.43
Family history of familial hypercholesterolemia			
None recorded/No	Ref	--	--
Yes	9.13	8.12	10.26
Family history of myocardial infarction			
None recorded/No	Ref	--	--
Yes	1.81	1.61	2.05
Family history of raised cholesterol			
None recorded/No	Ref	--	--
Yes	3.20	2.75	3.72
Any diagnosis of diabetes			
No	Ref	--	--
Yes	0.37	0.32	0.42
Any diagnosis of kidney disease			
No	Ref	--	--
Yes	0.71	0.62	0.81

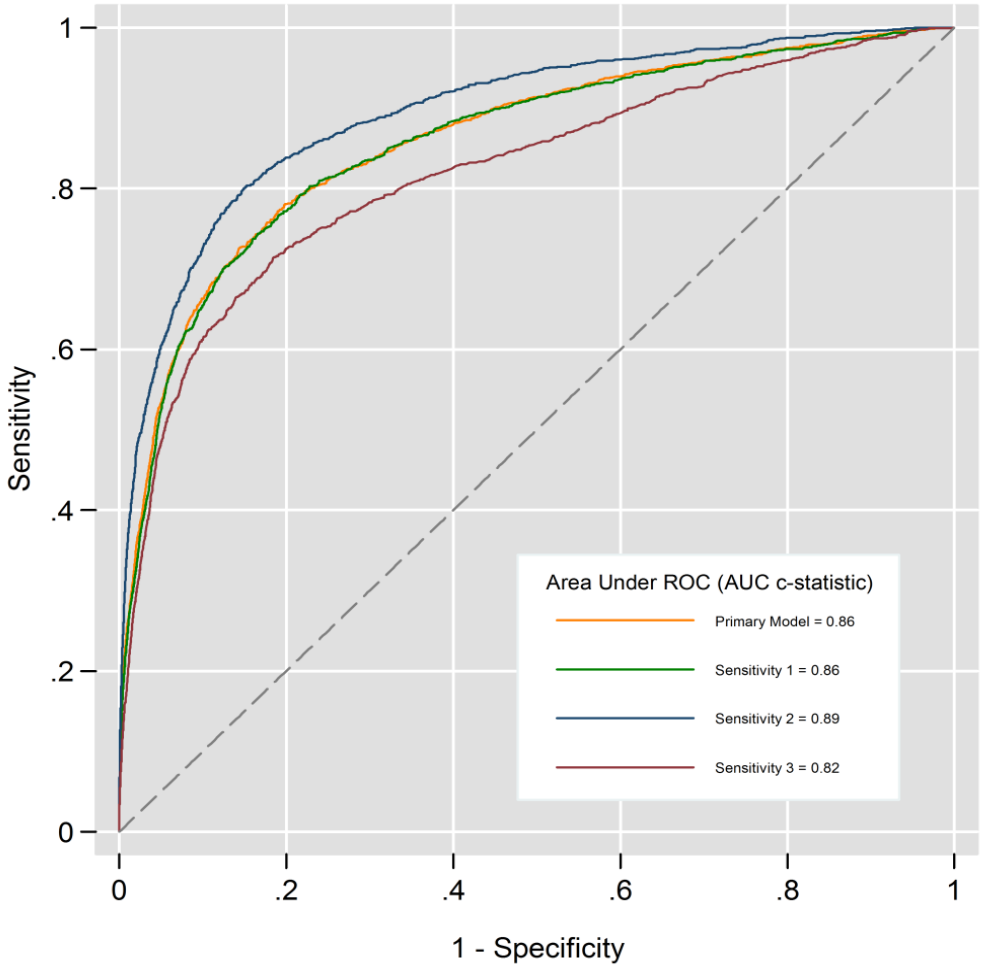
[1] Fluvastatin or Pravastatin ≤ 40 mg/day; Simvastatin ≤ 10 mg/day

[2] Fluvastatin or Pravastatin 80 mg/day; Simvastatin 20 mg/day or 40 mg/day; Atorvastatin ≤ 10 mg/day; Rosuvastatin 5 mg

[3] Simvastatin 80 mg; Atorvastatin ≥ 20 mg/day; Rosuvastatin ≥ 10 mg/day

Model Performance	AUC c-statistic*	Standard Error+	95% Confidence Interval	R^2
Primary Analysis				
Simon-Broome	0.749	0.007	0.735 - 0.763	0.105
FAMCAT	0.860	0.006	0.848 - 0.871	0.179
Sensitivity Analysis				
FAMCAT excluding secondary disease causes[1]	0.858	0.006	0.845 - 0.869	0.173
FAMCAT with comprehensive family history of MI[2]	0.894	0.005	0.884 - 0.904	0.232
FAMCAT excluding family history variables[3]	0.820	0.007	0.807 - 0.834	0.137

***Harrell's c concordance index**
+Bootstrap standard errors using jack-knife procedure
[1] Excluded kidney disease and diabetes
[2] Assumes 80.3% of familial hypercholesterolemia cases and 9.3% of non-cases have positive family history of myocardial infarction
[3] Excluded family history of myocardial infarction, raised cholesterol and familial hypercholesterolemia



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Data extracted on 17/04/14 using Reference date 17/04/14

PRIMIS CHART care and health analysis in real time The University of Nottingham

JKJ

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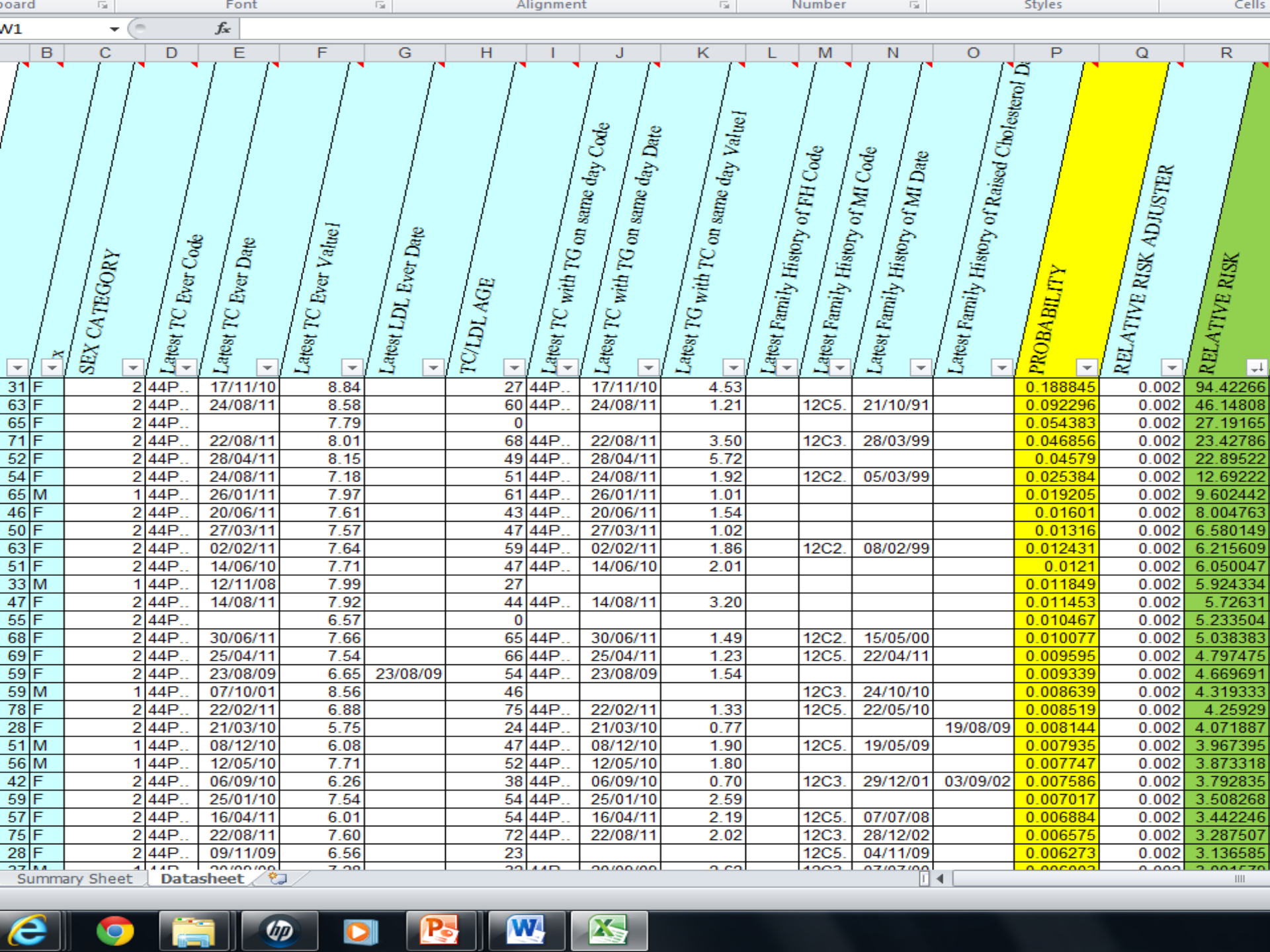
Total patients in practice	5296
↓	
Of which are aged 16-120	4459
↓	
Of which have a TC or LDL recording	1265

	Probability ≤ 1/1000	*Probability ≤ 1/500 to > 1/1000*	Probability > 1/500
Total Number	**998**	**157**	**110**
Men	485	61	39
Women	513	96	71
Total Patients with TC > 7.5 mmol/L OR LDL above > 4.9 mmol/L	1	0	29
Total Patients on lipid lowering drug treatment	**224**	**64**	**45**
Patients on high potency statins	161	55	41
Patients on medium potency statins	55	7	4
Patients on low potency statins	7	0	0
Patients on other lipid lowering drugs	1	2	0
Total patients with at least one of following family histories	180	60	61
Patients with family history of myocardial infarction	180	60	57
Patients with family history of cholesterol	0	0	0
Patients with family history of familial hypercholesterolaemia	0	0	5

	Probability ≤ 1/1000	*Probability ≤ 1/500 to > 1/1000*	Probability > 1/500
Total Men	**485**	**61**	**39**
16-24	2	1	0
25-34	7	1	2
35-44	33	8	10
45-64	62	18	10
55-64	114	15	8
65-74	127	14	9
75-84	113	2	0
85 and over	27	2	0
Total Women	**513**	**96**	**71**
16-24	2	4	0
25-34	11	5	6
35-44	30	11	9
45-64	75	21	14
55-64	82	22	22
65-74	113	21	12
75-84	132	12	7
85 and over	68	0	1

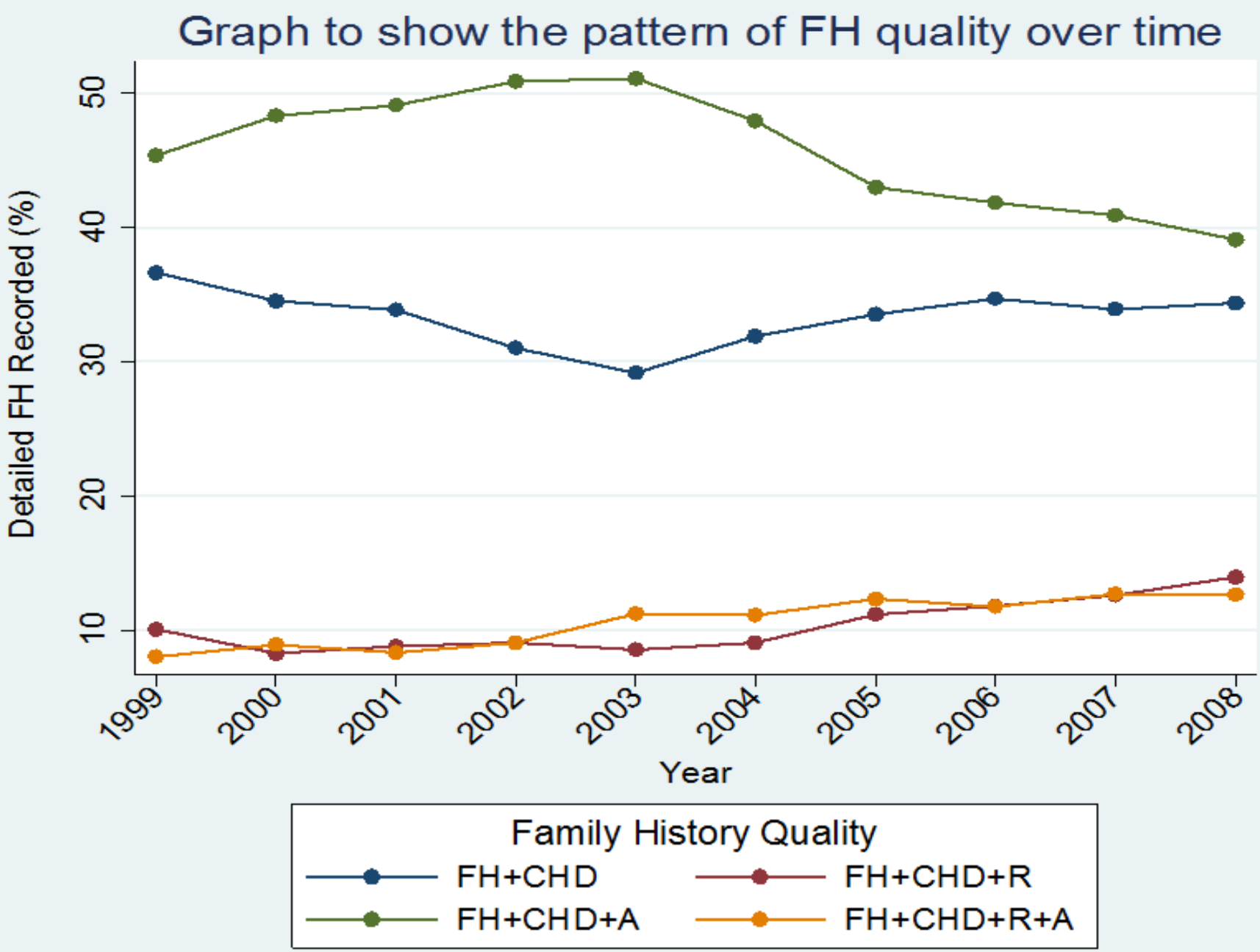


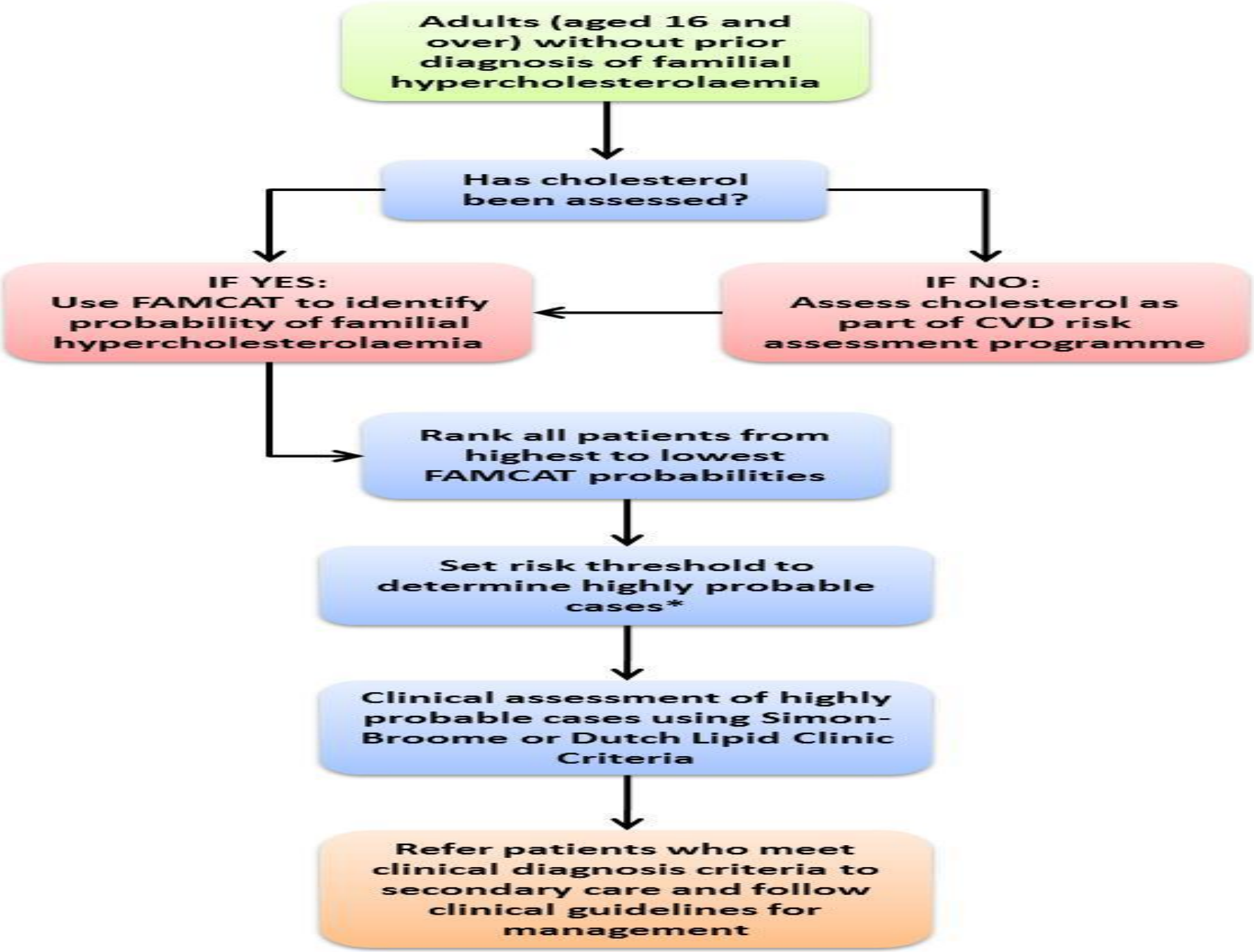
Font Alignment Number Styles Cells

W1

	B	C	D	E	F	G	H	I	J	K	L	M	N	O	P	Q	R
	Sex	SEX CATEGORY	Latest TC Ever Code	Latest TC Ever Date	Latest TC Ever Value1	Latest LDL Ever Date	TC/LDL AGE	Latest TC with TG on same day Code	Latest TC with TG on same day Date	Latest TG with TC on same day Value1	Latest Family History of FH Code	Latest Family History of MI Code	Latest Family History of MI Date	Latest Family History of Raised Cholesterol D	PROBABILITY	RELATIVE RISK ADJUSTER	RELATIVE RISK
31	F	2	44P..	17/11/10	8.84		27	44P..	17/11/10	4.53					0.188845	0.002	94.42266
63	F	2	44P..	24/08/11	8.58		60	44P..	24/08/11	1.21		12C5.	21/10/91		0.092296	0.002	46.14808
65	F	2	44P..		7.79		0								0.054383	0.002	27.19165
71	F	2	44P..	22/08/11	8.01		68	44P..	22/08/11	3.50		12C3.	28/03/99		0.046856	0.002	23.42786
52	F	2	44P..	28/04/11	8.15		49	44P..	28/04/11	5.72					0.04579	0.002	22.89522
54	F	2	44P..	24/08/11	7.18		51	44P..	24/08/11	1.92		12C2.	05/03/99		0.025384	0.002	12.69222
65	M	1	44P..	26/01/11	7.97		61	44P..	26/01/11	1.01					0.019205	0.002	9.602442
46	F	2	44P..	20/06/11	7.61		43	44P..	20/06/11	1.54					0.01601	0.002	8.004763
50	F	2	44P..	27/03/11	7.57		47	44P..	27/03/11	1.02					0.01316	0.002	6.580149
63	F	2	44P..	02/02/11	7.64		59	44P..	02/02/11	1.86		12C2.	08/02/99		0.012431	0.002	6.215609
51	F	2	44P..	14/06/10	7.71		47	44P..	14/06/10	2.01					0.0121	0.002	6.050047
33	M	1	44P..	12/11/08	7.99		27								0.011849	0.002	5.924334
47	F	2	44P..	14/08/11	7.92		44	44P..	14/08/11	3.20					0.011453	0.002	5.72631
55	F	2	44P..		6.57		0								0.010467	0.002	5.233504
68	F	2	44P..	30/06/11	7.66		65	44P..	30/06/11	1.49		12C2.	15/05/00		0.010077	0.002	5.038383
69	F	2	44P..	25/04/11	7.54		66	44P..	25/04/11	1.23		12C5.	22/04/11		0.009595	0.002	4.797475
59	F	2	44P..	23/08/09	6.65	23/08/09	54	44P..	23/08/09	1.54					0.009339	0.002	4.669691
59	M	1	44P..	07/10/01	8.56		46					12C3.	24/10/10		0.008639	0.002	4.319333
78	F	2	44P..	22/02/11	6.88		75	44P..	22/02/11	1.33		12C5.	22/05/10		0.008519	0.002	4.25929
28	F	2	44P..	21/03/10	5.75		24	44P..	21/03/10	0.77				19/08/09	0.008144	0.002	4.071887
51	M	1	44P..	08/12/10	6.08		47	44P..	08/12/10	1.90		12C5.	19/05/09		0.007935	0.002	3.967395
56	M	1	44P..	12/05/10	7.71		52	44P..	12/05/10	1.80					0.007747	0.002	3.873318
42	F	2	44P..	06/09/10	6.26		38	44P..	06/09/10	0.70		12C3.	29/12/01	03/09/02	0.007586	0.002	3.792835
59	F	2	44P..	25/01/10	7.54		54	44P..	25/01/10	2.59					0.007017	0.002	3.508268
57	F	2	44P..	16/04/11	6.01		54	44P..	16/04/11	2.19		12C5.	07/07/08		0.006884	0.002	3.442246
75	F	2	44P..	22/08/11	7.60		72	44P..	22/08/11	2.02		12C3.	28/12/02		0.006575	0.002	3.287507
28	F	2	44P..	09/11/09	6.56		23					12C5.	04/11/09		0.006273	0.002	3.136585

Summary Sheet | Datasheet


Graph to show the pattern of FH quality over time
Detailed FH Recorded (%)
10
20
30
40
50
1999
2000
2001
2002
2003
2004
2005
2006
2007
2008
Year
Family History Quality
FH+CHD
FH+CHD+R
FH+CHD+A
FH+CHD+R+A


Adults (aged 16 and over) without prior diagnosis of familial hypercholesterolaemia
Has cholesterol been assessed?
IF YES: Use FAMCAT to identify probability of familial hypercholesterolaemia
IF NO: Assess cholesterol as part of CVD risk assessment programme
Rank all patients from highest to lowest FAMCAT probabilities
Set risk threshold to determine highly probable cases*
Clinical assessment of highly probable cases using Simon-Broome or Dutch Lipid Clinic Criteria
Refer patients who meet clinical diagnosis criteria to secondary care and follow clinical guidelines for management