Familial Hypercholesterolaemia:
Primary & Secondary Care working together

Dr David Milne, GP, Colchester
Co-Chair, HEART UK
FH Implementation Team
Declaration of Interests

• In the past I have accepted remuneration for writing articles, chairing meetings, giving presentations and sponsorship for attending medical meetings from Servier pharmaceuticals, MSD, Schering-Plough AstraZeneca, Sanofi-aventis and Bristol Myers Squibb. I have been on advisory boards for GE Healthcare and Schering-Plough.

• This presentation is wholly non promotional and although Heart UK (the Cholesterol Charity) have provided assistance & support in the preparation I accept full responsibility for its content.

David Milne Nov 2014
The biggest advance that could be made in medicine, is for full use to be made of existing knowledge.’

Lord Max Rosenheim 1954
Presentation outline

1. Background to the FH Problem
2. Current England initiatives
   1. Education
   2. FH Nurses
   3. Medway
   4. SHIP
3. The relevance of FH to primary care
4. What can you do locally?
5. Conclusion
Background to the FH Problem

• FH is a autosomal dominantly inherited genetic condition which causes high concentrations of LDL cholesterol in the blood.
• People with FH are at very high risk of CHD from a young age. (50% of affected men have IHD at 50 years of age). Yet it can be easily diagnosed and treated effectively.
• Prevalence is estimated at between 1 in 500 to 1 in 200. So 120,000 to 300,000 people in the UK have FH, but only ~15,000 patients identified.
• Thousands are unaware they have the condition
Background: NICE FH Guideline (CG71)

- NICE FH Guideline (2008) shows cascade testing, using cholesterol and DNA tests in affected families, followed by high intensity statin therapy, is cost-effective and delivers optimum health outcomes
- Cascade testing most cost effective at £2,700 per QALY
- After NICE Guideline published, HEART UK established FH Implementation Team (FHIT)
- There has been a failure to implement NICE Guidelines on FH in England
Why NICE Guideline not implemented

• Localised commissioning structure hampers development of FH services & access to genetic testing
  ➢ Too common for specialist commissioning
  ➢ Too rare for general awareness
• Lack of clinical awareness and understanding of FH
• Lack of lipid clinics & specialist services
• Lack of appropriate IT software for cascade screening & registering patient data
• Money is very tight
FH Report findings

• If 50% of patients with FH are diagnosed & treated optimally over 55 year period, £94.7 million (£1.97m per 1,000 cases) can be saved by NHS, or £1.7m per year.

• By not implementing cascade screening as recommended by NICE (identifying 50 per cent of potential relatives cases), NHS is losing £1.4m per year.

Saving Lives Saving Families
HEART UK recommendations

1. A national programme for FH in England under NHS England, featuring:
   - A dedicated network of HCPs
   - Clear referral pathways at the local level
   - FH nurses to rollout the cascade screening process
   - Measurement of outcomes as the programme is rolled out.

2. UK-wide national patient register and database

3. Improved capacity of lipid clinics

4. Increased education and training programmes.
Initiatives in England

• No ‘national’ FH programme in England, but CVD Outcomes Strategy ‘aspiration’ to find 50% of FH cases
• FH Quality Standard (QS41) – 8 Quality Statements for identification and management of FH patients
• HEART UK series of 4 FH articles in PCCJ (2013-2104)
• The BHF awarding grant funding for FH cascade testing nurses in England, Scotland and Northern Ireland
Educational articles in PCCJ
CPD associated with the articles

• Article 1 - CPD completed = 2561 clinicians
• Article 2 - CPD completed = 2167 clinicians
• Article 3 - CPD completed = 1732 clinicians (NB – online for only two months to-date)

Slade Carter - personal communication August 2014
BHF funding for FH Nurses – “Text Santa” 2013 - £1.3m

Applications from lipid services with support from CCG – funding for 18/12

- London Royal Brompton and Harefield NHS Trust
- London Royal Free NHS Foundation Trust
- London Guys and St Thomas'/St George's
- South Yorkshire Cardiothoracic Centre
- Manchester Central Manchester Foundation NHS Trust
- Bristol University Hospital
- Sunderland City Hospitals Sunderland Foundation Trust
- Scotland NHS Grampian
- Southampton Southampton General

Second round just announced - 24 November 2014 ‘Familial Hypercholesterolaemia: The Role of Commissioners and Service Providers in Improving Quality of Care and Patient Outcome’.
Other Initiatives - Updates

- Medway FH Audit
- SHIP project
- Isle of Wight
- FH Wales
Medway CCG FH Audit Project

- HEART UK working with Medway CCG (Dr Peter Green, Chief Clinical Officer at Medway CCG) and Sanofi
- 54 Practices – 280000 patients

Phase 1
- Audit + software (BMJ Informatica) – in consultation prompts

Phase 2
- Nurse Advisor to case find at risk individuals and screen
## Medway CCG FH Audit Project

### Table 1. Triggers and prompts within the Medway FH audit tool

<table>
<thead>
<tr>
<th>Trigger</th>
<th>Prompt</th>
</tr>
</thead>
<tbody>
<tr>
<td>Patients with FH or possible FH whose family has not been informed</td>
<td>Have relatives been informed regarding FH?</td>
</tr>
<tr>
<td>Patients with FH, possible FH or probable FH whose latest total cholesterol $&gt;$ 5 mmol/L</td>
<td>Up-titrate statins or consider referral</td>
</tr>
<tr>
<td>Patients whose latest cholesterol is $&gt;$ 7.5 mmol/L or LDL-C $&gt;$ 4.9 mmol/L who have had a positive genotype test</td>
<td>Diagnose FH</td>
</tr>
<tr>
<td>Patients whose latest cholesterol is $&gt;$ 7.5 mmol/L or LDL-C $&gt;$ 4.9 mmol/L and have a family history of premature CHD and/or hypercholesterolaemia and have not had a Simon Broome assessment</td>
<td>Consider possible FH</td>
</tr>
<tr>
<td>Patients whose latest cholesterol is $&gt;$ 7.5 mmol/L or LDL-C $&gt;$ 4.9 mmol/L, have not had a Simon Broome assessment and have a family history of CHD but no details of the age of the relatives</td>
<td>Ask patient if MI $&lt;$ 50 years of age in second degree relative or MI $&lt;$ 60 years of age in first degree relative <strong>Yes:</strong> Consider FH <strong>No:</strong> Assess using Simon Broome criteria</td>
</tr>
</tbody>
</table>

Note: Prompts contain further information along with relevant read codes, which can be added directly into the patient record from the prompt screen.
Figure 1: Summary of the Medway FH audit results at baseline, after two years and after the introduction of the FH Nurse Advisor Programme

FH (definite, probable and possible)
* Patients were considered to be ‘at risk and unscreened’ if they had a total cholesterol >7.5 mmol/L and/or LDL-C >4.9 mmol/L and had not been assessed using the Simon Broome criteria.
SHIP

- Southampton
- Hampshire
- I of Wight
- Portsmouth
SHIP FH cascade testing service

- Leads: Prof Chris Byrne, Melanie Watson
- Funded by consortium of 8 CCGs (= 2.5 million people)
- FH clinic, includes mobile specialist nurse and genetic counsellor, with genetic tests in Bristol
- Diagnosis determined by hospital lipid specialist in consultation with nurse and genetic counsellor
- Project demonstrates capacity of CCGs to pool resources to est. FH service for large population
3 models of care: same entry

Potential FH case identified by GP, lipidologist or cardiologist

FH Co-ordinator assesses

Diagnostic criteria met: Genetic testing

Diagnostic criteria not met: Referred back

Diagnostic criteria met: Genetic testing

Diagnostic criteria not met: Management on phenotype
SHIP costing demonstrate testing much less costly in 2013 than in 2008

Costs and benefits over 10yrs

- 43% less expensive
The economic case

Cost per QALY of FH cascade testing compared to other interventions judged to be cost effective

- FH: adding DNA test to cascade testing
- Cervical screening
- Cochlear implants
- Rituximab for lymphoma

NICE threshold for cost effectiveness: £20,000
Isle of Wight – update 2014

• Lead: Dr Ali Al-Bahrani, Consultant Chem Pathologist
• Eclipse software program to search patients with very high cholesterol
• Started with one surgery, now 12 out of 17
• Letters sent to patients, inviting appt with Dr Al-Bahrani
• Dr Al-Bahrani then assesses and treats where necessary, incl referral for genetic test
• So far - 37 FH positive
• Will move on to cascading through families
Other initiatives: National register?

• BHF funding for FH nurses

• AstraZeneca purchased 50 licences from PASS for use in England, Northern Ireland & Scotland

• HEART UK working with AZ to establish criteria for licences being awarded, to help ensure high standards

• First step to establishing national register for FH patients
The Welsh model

Other parts of the UK are doing better than England, with dedicated FH standards or programmes

• FH All Wales Cascade Testing Service features diagnostic service for FH & family cascade testing, with Welsh Assembly funding

• Multidisciplinary service links with lipid clinic provision, clinical genetics, paediatrics & laboratory testing. Service hosted by Cardiff and Vale Health Board.
Familial Hypercholesterolaemia (FH) is a common inherited condition affecting around 1 in 500 of the UK population. It is caused by an abnormal gene which results in very high cholesterol levels in the blood.

People with FH, if untreated, are at an increased risk of early coronary heart disease. The FH Wales Service identifies and treats individuals and families in Wales.

The FH Service for Wales aims to diagnose individuals and families affected by FH so that treatment can prevent early onset heart disease.
The Welsh model

- Service enables availability within Wales for referral to specialist lipid services and for genetic testing for FH
- Provides pathway for system of family testing for FH, including Welsh national database using PASS software.
- Based on Dutch experience, the service aims to identify 60 per cent of those affected in Wales over 10 years.
- At start up in 2010 – 97 FH patients in Wales
- So far - 1183 identified with 492 genetic positive
The relevance of FH to primary care

- FH not specified in QOF, but primary care is key to identifying and treating people with FH
- NHS Health Checks - people with total cholesterol >7.5 be considered as possible FH.
- Clinical databases of past Cholesterol recordings
- Tie in with NHS Outcomes Framework: Domain 1 prevent people dying prematurely; Domain 2 enhance quality of life for people with LTCs; Domain 4 positive experience of care
- Primary care gatekeepers for secondary care referral and possible genetic testing
What can you do locally?

• Think of the possibility of FH in your own practice
• Ensure referral and care pathway to lipid clinics well established & promoted in primary care
• Improve access to genetic testing – work with genetics service to set criteria & keep costs reasonable
• Include FH in local health promotion & awareness campaigns
Conclusion

• The failure to implement the FH Guideline is
  ➢ Costing Lives
  ➢ Costing Money

• A missed opportunity to capture current
generations, but also future generations, as FH will
be known in affected families. The potential
savings and long term health gains are therefore
boundless

The biggest advance that could be made in medicine, is for
full use to be made of existing knowledge.’

Lord Max Rosenheim 1954
Contacts

HEART UK website: www.heartuk.org.uk

Visit the Heart UK stand - Jules Payne CEO & Helen Walsh
David Milne david.milne1@nhs.net
5 minutes to get to Peter Green’s talk! OR stay & listen to Professor Gorog