Familial Hypercholesterolaemia Primary Care Project

Hardwick Hall – 18th October 2018

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Chair NENC Lipid Specialists Advisory Group

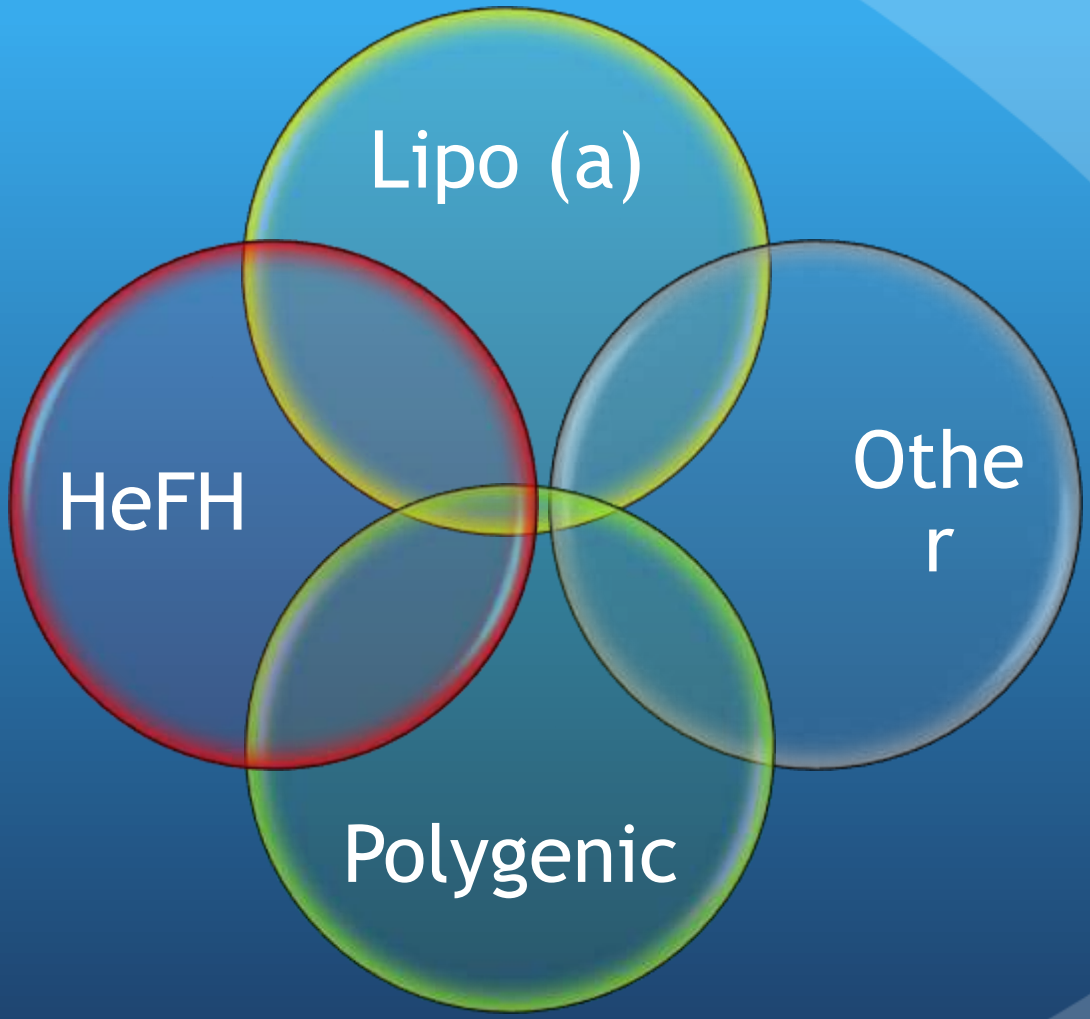
Completing Interests

- I have undertaken research for Amgen
- I have participated in an Advisory Board for Sanofi

A common Lipid Clinic referral query:- "Could this patient have FH??"

- This can present in many ways
- High cholesterol
  - Chol >7.5 mmol/L or LDL > 4.9 mmol/L
  - ? Other Simon Broome criteria??
  - Mixed dyslipidaemia
- Family History of Cardiovascular disease
  - Premature MI (< 60yr 1^{o} / <50yr 2^{o})
  - History of sudden death
  - Premature CeVD

It’s not always as simple as you think!



Time to reframe the question!

- Could this patient have an identifiable pathogenic mutation to explain their high cholesterol and / or their family history of cardiovascular disease, that would allow us to screen their relatives and prevent further cardiovascular events?

The Project

- Use the PRIMUS tool to identify (and stratify) patients with possible FH from Primary Care database
- Use FH Specialist Nurses to work in Primary Care and undertake genetic testing (DLCS>6)
- Streamline referrals to local Lipid Clinics
- Focus on areas with less developed FH services
- Identify more individuals and families with FH

Agenda

- **FH: A national Overview**
  - Prof Huon Grey
- **The value of FH Genetic diagnosis and Cascade testing services**
  - Dr Dermot Neely
- **FH in the NE – the story so far**
  - Dr Guy Pilkington / Dr Jonathan Smith
- **The Role of the FH Nurse**
  - Susan Musson / Lorna Ingoe
- Summary and Close