FH Pilot Study Results

AHSN NENC CVD Prevention Programme Launch

Dr Peter Carey

On behalf of the AHSN NENC FH Steering Group

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Background

- FH Genetic testing in NENC since mid 2014
- Historically higher uptake in North and West of the region
- As part of negotiations around ongoing funding for regional FH genetic services Durham and Darlington keen to develop more testing in Primary Care
- Medway project had previously shown that systematic searches of Primary Care records could improve rates of FH diagnosis on clinical criteria
- NICE now recommend systematic searches based on:-
  - Chol >7.5 mmol/L if <30 yrs AND
  - Chol >9.0 mmol/L if >30 yrs
- Aim was to identify ~20 patients suitable for FH genetic testing
  - 6-7 indexes with a confirmed mutation
  - Further 12-14 relatives through cascade testing

Phase 1 - FAMCAT

- 4 Practices – population = 45,123; 180 Expected FH
- 6 Patients with prior FH DNA diagnosis
  - 1 undergoing testing → negative
- 303 very high risk – 43 (14%) invited to screening
  21 (49%) attended
  12 eligible for testing
- 3 significant results 1 positive
  2 VUS
- 28 1st Degree + 77 2nd Degree = Expect 33 +ve cascade testing

Re-development of Pathway

- FAMCAT tool used hard drive of GP IT system and not integrated with practice medical records – impractical to use as presented a security challenge
- Large number of patients not eligible for FH testing due to underlying secondary causes (↑TG) or incomplete data
- Time consuming triaging process to identify low numbers of eligible patients
- Development of search tool incorporating NICE criteria (excluding ↑ TG) and personal history of premature CVD < 50 yr
- Feedback from patients led to changes to patient literature and pre visit telephone call to discuss rationale for appointment

Phase 2 - CDRC

- 5 Practices – population = 49,321; 197 Expected FH
- 9 Patients with prior FH DNA diagnosis
- 126 very high risk – 71 (56 %) invited to screening
  54 (76 %) attended
  53 eligible for testing - 2 declined
- 24 significant results 22 positive
  2 VUS
- 19 Indexes – 6 members of 1 family
- 50 1st Degree + 89 2nd Degree = Expect 47 +ve cascade testing
- All practices had positive patients with minimum of 2 patients per practice

Summary

- Developed an optimized FH pathway based on systematic Primary Care record searches
- Incorporated patient feedback to improve awareness
- Allows guided FH genetic testing and cascade testing of relatives
- Developed for use via SystmONE and EMIS
- Aim :- meet NHS Long Term Plan target of improving FH diagnosis rates to 25% over next 5 years