Towards optimising collaborative FH Care and Research in the Amsterdam Region

APH meeting Personalized Medicine
23 November 2017

Carla van El VUMC Community Genetics

Pauline Slottje VUMC Primary Care ANH Database
APH PM 10,000 euro collaboration AMC VUMC

Project team:

- Carla van El - Community Genetics VUMC
- Pauline Slottje, Dick Walstock – Department & Academic Network of General Practice (ANH VUmc)
- Kees Hovingh, Merel Hartgers, AMC Vascular medicine
- Henk van Weert, Dept of General Practice AMC
- Manon Houter, Janneke Wittekoek - LEEFH
- Mirte Schaafsma, VUMC research assistant
What is the problem?

• Familial Hypercholesterolemia (FH) affects 1:240
• mostly mutation in LDL receptor \textit{LDLR}, \textit{APOB} or \textit{PCSK9} gene
• elevated plasma Low-density lipoprotein cholesterol levels cause premature cardiovascular events
• First degree family members of patients have a 50% chance of carrying the same mutation
• Treatable via statins
• End of population screening programme 2014\textgreater
Half of carriers (70,000) not detected
Population screening programme 1994-2014

- Strong role AMC care, research and database
- Active approach of family members: via index patient contacted by Foundation for Tracing Familial Hypercholesterolaemia (StOEH)

<table>
<thead>
<tr>
<th></th>
<th>94-95</th>
<th>2006</th>
<th>2007</th>
<th>2008</th>
<th>2009</th>
<th>2010</th>
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<th>2012</th>
<th>2013</th>
<th>2014</th>
<th>2015</th>
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<td>1793</td>
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<td>1796</td>
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<td>1492</td>
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<td>1964</td>
<td>1699</td>
<td>1095</td>
<td>1121</td>
<td>-</td>
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<tr>
<td>Index patients</td>
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<td>363</td>
<td>448</td>
<td>338</td>
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<td>260</td>
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<td>Family screening</td>
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<td>4285</td>
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<td>3593</td>
<td>2098</td>
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<td>FH positive</td>
<td>10074</td>
<td>2271</td>
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<td>1596</td>
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<td>1685</td>
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<td>1361</td>
<td>869</td>
<td>155</td>
<td>219</td>
<td>23161</td>
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<tr>
<td>% of positive</td>
<td>33%</td>
<td>37.10%</td>
<td>36.20%</td>
<td>37.20%</td>
<td>36.40%</td>
<td>36.20%</td>
<td>34.70%</td>
<td>37.90%</td>
<td>41.40%</td>
<td>57%</td>
<td>55%</td>
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<td>Detected (include index patients)</td>
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<td>1670</td>
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<td>390</td>
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</table>
After 2014 - role of the GP in FH care

- Diagnosis still via GP
- Referral to Vascular medicine or LEEFH (National Expertise Center:
  - database (check known families)
  - inform patients
  - encourage patient to contact family
Methods

- Prior search in GP database on coding for FH

- 3 focus groups (including additional training)
  - ANH VUMC (Amsterdam network primary care studies)
  - HAG net AMC (Primary care studies)
  - ROHA (Regional Organisation GPs Amsterdam)

- Interviews experts and physician assistants (praktijkondersteuners)
Focus group: Role of the GP in FH care

- Presentation on FH epidemiology, diagnosis, treatment (AMC)
- Presentation on LEEFH (National Expertise Center)

4 themes
- Coding in GP database - mirror information
- Knowledge and awareness - FH diagnosis and family risk
- Role of GP – focus on patient and commitment family?
- Support care GP – role physician assistants (POH)
Aim of project

• explore barriers and strategies for improvement FH care in primary care
• intensified information and communication between primary and secondary care strengthening a clinical network
• improve registration in electronic medical records
• strengthen the infrastructure for research on FH in the Amsterdam region

>> Write action plan how to optimise FH care
>> consider new research and potentially options for prediction modelling