Systematic Review of Family History Questionnaires in Primary Care

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Family History in General Practice

- An important risk factor for conditions including cancers, cardiovascular disease, diabetes, mental illness, osteoporosis
- Family history of a common chronic disease is associated with a 2 to 5-fold relative risk of developing the condition

How Can We Use Family History?

- Reflects shared genetic, environmental and behavioural risks
  - Not used systematically in general practice
- Potential use as a screening tool for tailored risk assessment and preventive management
- Identify sub-groups with specific genetic conditions (e.g. breast cancer, familial hypercholesterolaemia)

Family History As a Risk Predictor

- Increases with the number of affected relatives
- Increases with early age of onset
- Increases with particular ethnic backgrounds:
  - E.g. Ashkenazi Jewish – breast/ovarian cancer
  - E.g. South East Asian - haemoglobinopathies
  - E.g. Aboriginal/TSI – type 2 diabetes

Risk assessment is a complex task!

Family doctors require on average ~30 minutes to record a typical 3 generational pedigree
**What’s Needed?**
- Simple family history questionnaire to triage patients
- Applicable to entire population
- Highlights who needs a more detailed risk assessment
  - Pedigree
  - Referral to specialist or additional screening tests
- Time efficient and user friendly

**Aim**
- Systematically review previously developed family history questionnaires
- Develop an Australian Family History Questionnaire validation study

**Systematic Review**
- Medline, Google Scholar
  - Search terms: family history screening, family history questionnaire, chronic disease, primary care
  - MeSH terms: genetic predisposition to disease, medical family history taking

**Systematic Review**
- Inclusion criteria:
  - Questions used to determine family history of disease
  - Evaluation of either the FHQ itself, or, the result of its use (e.g. behaviour change)
  - Online search of “grey literature”

**Data Extracted**
- Paper vs. electronic tool
- Consumer vs. professional completion
- Pedigree production
- Condition specific
- Risk assessment
- FHQ wording/phrasing

**Data Extracted: Evaluation of Questionnaires**
- Validation of tools
- Types of evaluation:
  - Validation of family history data against gold standard (e.g. cancer register, full pedigree)
  - Effect on behaviours of undergoing screening (e.g. uptake of screening such as mammography)
Results
- Grey online literature
  - 26 websites identified
  - Many more. Number identified only restricted by time to search...
  - E.g. CDC “My Family Health Portrait”
  - E.g. Harvard “Your Disease Risk”
- No formal evaluation

Results – Published Literature
- To date 23 published articles from which data has been extracted:
  - 12 excluded
    - Mainly not meeting validation criteria
  - 11 met inclusion criteria

Validation of Questionnaires

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<tbody>
<tr>
<td>Family history questionnaires validated</td>
<td>8</td>
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<tr>
<td>Validation regarding effect of use</td>
<td>1</td>
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<tr>
<td>Both</td>
<td>3</td>
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Results: Overall Features
- None had a scoring system that could be used directly as a screening tool
- Mainly focused on single condition
- Mainly patient self-completion tools
- Mainly paper-based...

Results: Wording of Questionnaires
- Ethnicity:
  - Please tick the box that best describes your parents’ ethnic group.
    vs.
  - What ethnic group do you most identify with?

Results: Wording of Questionnaires
- Family history
  - Has anyone in your family ever suffered from...
  - Do you have any close relatives (mother, father, brother, sister, son, daughter) with...
  - Do you have any first degree relatives who have been diagnosed with...
  - Do you have any immediate relatives with...
  - Have any of your parents, brothers, sisters or children developed...
Where to From Here?

- Inform the development of a simple family history screening tool for use in primary care.
- Familial cancers, cardiovascular disease, diabetes.
- Validated in a general practice population in Perth.
- 500 patients to be recruited.
- 3 generational pedigree to be collected and used as the "gold standard" and compared to the scored family history tool.

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