Use of Family History Information in Pediatric Primary Care and Public Health:

A Family Physician's Perspective and Prescriptions

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February 23, 2006

Who am I?

- Current:

  Assistant Professor of Family and Community Medicine in a community hospital residency program associated with the Medical College of Wisconsin.

- Former

  Practicing physician in rural Vermont and northwestern Massachusetts

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Who am I?

I am informed in what I do both by my teaching role and my life as a rural practicing physician.

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What is a Family Physician?

"Family medicine is the medical specialty which provides continuing, comprehensive health care for the individual and family…The scope of family medicine encompasses all ages, both sexes, each organ system and every disease entity."

American Academy of Family Physicians, 2005

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Family Practice and Family History

- One of the beauties of family practice for me is knowing whole multi-generational families.
- I will often see a patient and know at least some of the family history.
- If I need to confirm a fact or get additional details (e.g. age of onset), it may already be in that family member's chart!

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Family Practice and Family History

- That history is mostly in my head
- Can fill it in as I access each patient's chart for family history; and
- Try to remember to access other charts as needed when a notable event occurs (new diagnosis of cancer or Alzheimer's)
- Electronic records are now no better than paper records at cross referencing such data.

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What are my biggest concerns about collecting family histories?

Biggest Concerns on Collection

Inaccurate information:

- Studies are usually on cancer
  - Accuracy is highest for breast cancer (94-97%)
  - Accuracy is highest for first degree relatives (85-95%), with decreasing accuracy as relations get further out (60% in 3°).
  - In general specificities are high, sensitivities are what drop off (**under reporting is the issue**).

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Biggest Concerns on Collection

- Unknown what would happen with less morbid conditions (e.g. diabetes, hypertension, asthma) but:
  - One study of parents reporting childhood illnesses and medications to a dentist:
    - Sensitivities mostly in the 50-86% range.
    - Specificities were all in the 94-100% range.
    - For this study, if you weight failures to report by prevalence, most of the weighted failure to report rates were 4-5%.

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Biggest Concerns on Collection

- Incomplete information
  - little collection of SAb's, nephews and nieces, only cause of DEATH collected)
- Collected information not useful
  - ages at DEATH, not ages of ONSET (8%-10% of charts in two studies have latter)
- Lack of reimbursement
- Updating of information

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Biggest Concerns on Confirmation

- Access to data
  - Permission not sought by proband or
  - Permission not granted or
  - Records unobtainable (can't find, not enough known) or
  - Affected family member is deceased
- Lack of reimbursement
  - Four to five contacts required to get peak data return

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Biggest Concerns on Interpretation

- Smaller families = less data
  - Combine with inaccuracies due to more distant relations and
- Lack of education
  - studies from Great Britain show correct interpretations by PCP's of cancer family histories run 20 to 60%. Only 26% knew all three primary risk factors (age at diagnosis, degree of relatedness, numbers of family members affected)
- Complex diseases could be much worse

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Interpreting Complex Disease Family Histories

- Issues will include
  - Multiple low penetrance genes
  - Multiple diseases in one family
    - May be interrelated (hypertension, diabetes, obesity, heart disease)
  - Environmental factors
  - Personal and familial factors (diet, exercise, tobacco abuse)

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What factors influence completeness and accuracy?

How can we improve these?

Factors for Completeness and Accuracy

- Type of disease
  - Severity
  - Public attitude
  - Options within a category like “lung disease”
  - Familiarity to general public (thrombophilias, rheumatic conditions bad)
- Time since diagnosis/death (accuracy of diagnosis and accuracy of recall)
- Disease patterns

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Factors for Completeness and Accuracy, Part II

- Closeness of relation
- Family relationships
- Culture?
- Access to data (death certificates, medical records)

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What makes completeness, accuracy better?

- Really not known
  - Emphasis by provider?
  - Specific questions?
  - Guided questions (electronic)?
  - Filling out history at other than office visit (paper or electronic)?

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“Prescriptions” for teaching

- Importance of family history
- Importance of age of onset
- How to do a good family history
- Underreporting (sensitivity) is the issue
- Interpretive skills: red flags

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"Prescriptions" for research

- Accuracy of family history in common complex diseases
- Ways to increase sensitivity
- Ways to convince public of importance
- Accuracy of age of onset recall
- Sensitivity variations:
  - Age (older patient, more underreporting in one study)
  - Cultural issues (non-white also increased underreporting in that study)

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Other "Prescriptions"

- Push electronic medical records that are "Family History Friendly"
- Electronic resources are going to be critically important to primary providers as the waves of genetic information get higher and more frequent
  - Reference resources
  - Interpretive resources
- Continue the public health emphasis
- Work with AMA's CPT coding folks and medical payers to reward providers with completed and updated family histories

The End!

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Family Practice and Family History

- To do better would require a “relations” field in an electronic record, where you could enter a name and a relationship (“Barbara Jones” “maternal grandmother”), and two things would happen:
  - The field would populate with all relations in the database using other charts as reference
  - Personal and family history in all those charts that is already entered would be mined and modified appropriately: if Barbara had breast cancer with onset at 45 y.o., the chart would have an entry for “maternal GM, breast cancer @ 45 y.o.”

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Family Practice and Family History

- I strongly suspect that such a field would be useful in Pediatrics as well, where siblings are routinely seen; probably also in Internal Medicine, and especially as life expectancy increases.

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Family Practice and Family History

- A complication would be where I know something about a patient that they do not want to tell family members about (carriage of an Alzheimer's gene, for example)
  - Solution: include a check box for each entry saying "Do Not Include in Family Histories". This returns us to current state, where it is in my head, but not the chart.

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