Family History: a Disease Prevention Tool for Public Health Practice

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CDC, Office of Genomics & Disease Prevention
DNPA Teleconference
October 13, 2005
I. Defining and measuring family history

II. Rationale for using family history for disease prevention

III. New tools and initiatives – Family Healthware and My Family Health Portrait

IV. Incorporating family history into state programs
Risk factors for chronic diseases

- Genetics
- Environmental Exposures
- Behaviors

Interaction

Genetics

Behaviors

Environmental Exposures
Risk factors for common diseases

- Behaviors
  - Modifiable
- Genetics
  - Not Modifiable
- Environmental Exposures
  - Modifiable

Family History
What is family health history?

A family’s combination of shared genes, environment, behavior, and culture

A risk factor
How is family history measured?

Have any of your blood relatives ever been diagnosed with asthma?

Yes  No

Has your “relative” ever been diagnosed as having coronary heart disease?

At what age was “relative” diagnosed?

Is “relative” still alive?

What did “relative” die of?

Relative = parents, grandparents, aunts, uncles, siblings, children, nieces, nephews, cousins…. 
Recording your family history
ASHG.org

For each relative, try to write down as many of these items as possible:

Age or date of birth (and, for all family members who have passed on, age at death and cause of death). When the information is unavailable, write down your best guess (for example, "40's").

Medical problems such as:

- Cancer
- Heart disease
- Diabetes
- Asthma
- Mental illness
- Birth defects such as spina bifida, cleft lip, heart defects, others
- Learning problems, mental retardation
- Vision loss/hearing loss at a young age
- Others

Note the ages at which the conditions occurred.

For family members with known medical problems, jot down if they smoked, their diet and exercise habits, and if they were overweight.
Family Tree (Pedigree)

- Old age 80
- Committed suicide 20
- Committed suicide 20
- Cancer 70
- Heart attack 60
- Old age 80
- Old age 70
- Cancer 60
- Thalassemia carrier
- Hysterectomy 40
- Arrythmia 85
- Arrythmia 68
- Thalassemia carrier
- Hysterectomy 40
- Old age 80
- Old age 70
- Cancer 60
- Thalassemia carrier
- Endometriosis 40
- Old age 20
- Committed suicide 20
- Old age 80
- Old age 70
- Cancer 60
- Thalassemia carrier
- Endometriosis 40
### Table 2

Suggested guidelines for risk stratification based on family history

<table>
<thead>
<tr>
<th>Risk Level</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>High risk</td>
<td>1. Premature disease in a 1st-degree relative</td>
</tr>
<tr>
<td></td>
<td>2. Premature disease in a 2nd-degree relative (coronary artery disease only)</td>
</tr>
<tr>
<td></td>
<td>3. Two affected 1st-degree relatives</td>
</tr>
<tr>
<td></td>
<td>4. One 1st-degree relative with late or unknown disease onset and an affected 2nd-degree relative with premature disease from the same lineage</td>
</tr>
<tr>
<td></td>
<td>5. Two 2nd-degree maternal or paternal relatives with at least one having premature onset of disease</td>
</tr>
<tr>
<td></td>
<td>6. Three or more affected maternal or paternal relatives</td>
</tr>
<tr>
<td></td>
<td>7. Presence of a “moderate risk” family history on both sides of the pedigree</td>
</tr>
<tr>
<td>Moderate risk</td>
<td>1. One 1st-degree relative with late or unknown onset of disease</td>
</tr>
<tr>
<td></td>
<td>2. Two 2nd-degree relatives from the same lineage with late or unknown disease onset</td>
</tr>
<tr>
<td>Average risk</td>
<td>1. No affected relatives</td>
</tr>
<tr>
<td></td>
<td>2. Only one affected 2nd-degree relative from one or both sides of the pedigree</td>
</tr>
<tr>
<td></td>
<td>3. No known family history</td>
</tr>
<tr>
<td></td>
<td>4. Adopted person with unknown family history</td>
</tr>
</tbody>
</table>

*From Scheuner et al.*

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**Algorithms for classifying risk**

Scheuner M et al.  
*Am J Med Genet*  
Risk Stratification/Pedigree Analysis

Weak

Moderate

Strong

Family history features associated with increased risk

• Early age at disease onset
• Two or more closely related affected relatives
• Two or more generations with affected relatives
• Multifocal disease/severe phenotype
• Disease in the less often affected sex
• Presence of related conditions
• Patterns suggestive of a known Mendelian disorders
What is the evidence for the association of family history with chronic disease?

<table>
<thead>
<tr>
<th>Disease</th>
<th>Relative Risk</th>
</tr>
</thead>
<tbody>
<tr>
<td>Heart disease</td>
<td>2.0 – 5.4</td>
</tr>
<tr>
<td>Breast cancer</td>
<td>2.1 – 3.9</td>
</tr>
<tr>
<td>Colorectal cancer</td>
<td>1.7 – 4.9</td>
</tr>
<tr>
<td>Prostate cancer</td>
<td>3.2 – 11.0</td>
</tr>
<tr>
<td>Melanoma</td>
<td>2.7 – 4.3</td>
</tr>
<tr>
<td>Diabetes</td>
<td>2.4 – 4.0</td>
</tr>
<tr>
<td>Osteoporosis</td>
<td>2.0 – 2.4</td>
</tr>
<tr>
<td>Asthma</td>
<td>3.0 – 7.0</td>
</tr>
</tbody>
</table>

Am J Prev Med
February 2003
Risk factors for common diseases – Family History
Risk factors for common diseases – Family History

Type 2 Diabetes
Usefulness of Cardiovascular Family History Data for Population-Based Preventive Medicine and Medical Research (The Health Family Tree Study and the NHLBI Family Heart Study)

Roger R. Williams, *MD*, Steven C. Hunt, PhD, Gerardo Heiss, *MD*, PhD, Michael A. Province, PhD, Jeannette T. Bensen, MS, Millicent Higgins, MD, Robert M. Chamberlain, PhD, Joan Ware, MSPH, and Paul N. Hopkins, MD, MSPH

Detailed medical family history data have been proposed to be effective in identifying high-risk families for targeted intervention. With use of a validated and standardized quantitative family risk score (FRS), the degree of familial aggregation of coronary heart disease (CHD), stroke, hypertension, and diabetes was obtained from 122,155 Utah families and 6,578 Texas families in the large, population-based Health Family Tree Study, and 1,442 families in the NHLBI Family Heart Study in Massachusetts, Minnesota, North Carolina, and Utah. Utah families with a positive family history of CHD (FRS ≥ 0.5) represented only 14% of the general population but accounted for 72% of persons with early CHD (men before age 55 years, women before age 65 years) and 48% of CHD at all ages. For strokes, 11% of families with FRS ≥ 0.5 accounted for 86% of early strokes (< 75 years) and 68% of all strokes. Analyses of > 5,000 families sampled each year in Utah for 14 years demonstrated a gradual decrease in the frequency of a strong positive family history of CHD (~26%/decade) and stroke (~15%/decade) that paralleled a decrease in incidence rates (r = 0.86, p < 0.001 for CHD; r = 0.66, p < 0.01 for stroke). Because of the collaboration of schools, health departments, and medical schools, the Health Family Tree Study proved to be a highly cost-efficient method for identifying 17,064 CHD-prone families and 13,106 stroke-prone families (at a cost of about $27 per high-risk family) in whom well-established preventive measures can be encouraged. We conclude that most early cardiovascular events in a population occur in families with a positive family history of cardiovascular disease. Family history collection is a validated and relatively inexpensive tool for family-based preventive medicine and medical research. ©2001 by Excerpta Medica, Inc.

(Am J Cardiol 2001;87:129–135)
Effects of a Controlled Family-based Health Education/Counseling Intervention

Marika Salminen, MSc; Tero Vahlberg, MSc; Ansa Ojanlatva, PhD, CHES, CSE
Sirkka-Liisa Kivelä, MD, PhD

**Objective:** To describe the effects of a controlled family-based health education/counseling intervention on health behaviors of children with a familial history of cardiovascular diseases (FH-CVDs). **Methods:** The intervention group (IG, n=432) received 5 counseling sessions. The control groups 1 (CG1, n=200) and 2 (CG2, n=423) received no counseling. Outcome measures comprised changes in diet, exercise, and cigarette smoking. **Results:** The changes in the use of fats and salt, and in exercise, were more favorable in IG than in CG1 and/or CG2. **Conclusion:** Health education/counseling produced positive effects on diet and nutrition in particular and in part in exercise.

**Key words:** adolescent, child, early intervention, health behaviors, primary prevention

*Am J Health Behav. 2005;29(5):395-406*
Family History

How can we use family health history to…

- assess risk for common diseases
- influence early disease detection
- target and prioritize prevention strategies
Why focus on family history...

....as a public health strategy when it’s pretty obvious what we need to do to prevent common chronic diseases?

- Exercise more
- Eat a healthier diet
- Stop smoking
- Drink alcohol in moderation
- Take an aspirin per day
- See your doctor
- Get screened
- Etc.
Prevention Efforts - Obesity

Prevalence of overweight and obesity among U.S. adults, age 20-74

<table>
<thead>
<tr>
<th></th>
<th></th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Overweight or obese</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>(BMI &gt;=25.0)</td>
<td>47</td>
<td>56</td>
<td>65</td>
</tr>
<tr>
<td>Obese (BMI &gt;=30.0)</td>
<td>15</td>
<td>23</td>
<td>31</td>
</tr>
</tbody>
</table>
Prevention Efforts - Lack of Physical Activity

59% of adults do not engage in vigorous leisure-time physical activity lasting 10 minutes or more per week

Source: NHIS, 2002
Prevention Efforts - Smoking

Source: BRFSS
Prevention Efforts – Poor Diet

Fruit & Vegetable Consumption
Nationwide - 2002

Source: BRFSS
Prevention Efforts– Colorectal cancer screening

- Only 45% of men and 41% of women aged ≥50 years had undergone a flexible sigmoidoscopy or colonoscopy within the previous 10 years or had used a FOBT home test kit within the preceding year
  CDC’s 2000 NHIS

- Approximately 41.8 million average-risk persons aged ≥50 years have not been screened for colorectal cancer according to national guidelines
  CDC’s National Survey of Endoscopic Capacity
What is the added value of family history?

- One size fits all population approach to prevention has limits

- Augment with targeted and personalized prevention strategies focused on higher-risk families

- Awareness of familiar risk may be a motivating factor for behavior change and screening uptake

- Family-centered approaches to risk reduction may have longer impact

- Earlier or more frequent screening based on familial risk may be cost effective
Goals of Family History Initiatives

• Increase awareness among the public and health professionals of the value of family history for disease prevention and health promotion

• Provide tools to gather information, assess risk, and guide prevention strategies; and educational materials to facilitate communication about familial risk between patients and providers

• Increase genomics and health literacy

• Prepare the public and health professionals for the coming era in which genomics will be an integral part of regular health care
U.S. Surgeon General’s Family History Initiative
U.S. Surgeon General’s Family History Initiative

- November 8, 2004
  - Formal announcement of the initiative
  - Release of My Family Health Portrait

- Thanksgiving Day, 2004
  First Annual National Family History Day
Welcome to Your Family Health Portrait

Your Family Health Portrait allows you to create a personalized family health history based on information you provide about you and your family’s experience with the leading diseases.

Your Family Health Portrait should only be used in consultation with a healthcare professional. It can be a valuable tool for discussion, risk assessment, and medical advice.

New Users Start Here ▶
Returning Users ▶

Information you provide is stored on your computer’s hard drive and is only as secure as that drive. Please take appropriate precautions to protect sensitive information. For general questions about security and privacy, click here.

http://www.hhs.gov/familyhistory/
La iniciativa de Historial Familiar del Cirujano General
U.S. Surgeon General’s Family History Initiative

1. My Family Health Portrait – new easy to use web-based tool

2. Resource packets for health departments and community groups
   - Fact sheets
   - FAQs
   - Case Studies
   - Web resources
   - Electronic and hard copies of FHx tools
   - Presentations
   - Brochures and posters
Welcome to Family Healthware

Family Healthware is a tool that collects information on your:

- lifestyle behaviors
- use of screening tests
- family history of six major diseases

and produces a personalized report that:

- analyzes your family history as a risk factor for disease
- recommends screening, lifestyle and other changes to improve your health.

New Users
Begin your Family Healthware assessment today...

Create My Account

Returning Users
Username: 

Password: 

Forgot your username or password?

Family Healthware is not designed to replace medical advice and discussions with a health professional. You should talk to your health professional before making a decision about your medical care.
Incorporating family history into programs

• Collect family history data at state and local levels
  - BRFSS
  - Health interview surveys
  - Focus groups

• Include family history data in disease registries (cancer, birth defects)

• Increase family history awareness in the community

• Incorporate family history into prevention/risk reduction activities
Family history is a risk factor for diseases throughout all stages of life.

- Birth defects
- Blood disorders
- Diabetes
- Depression
- Alzheimer’s disease
- Osteoporosis
- Infants
- Children
- Adolescents
- Adults
- Older adults
- Asthma
- Autism
- Cancer
- Heart disease
Establish disease-specific prevalence estimates of FHx

Prevalence of Familial CHD

- **Strong**
  - 31% (n=1273)
- **Moderate**
  - 12% (n=471)
- **Weak**
  - 57% (n=2291)

Scheuner et al. Healthstyles 2003
Develop and validate familial risk stratification algorithms

<table>
<thead>
<tr>
<th>Risk Category</th>
<th>n (%); Criteria Met per Subject</th>
<th>ORa</th>
<th>95% CI</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Strong Familial Risk</strong></td>
<td>1273 (31%); 1.86 criteria met</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>At least . . .</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Two 1st deg early CHD, same</td>
<td>4.1</td>
<td>2.5-6.7</td>
<td></td>
</tr>
<tr>
<td>lineage</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>One 1st deg early CHD + one</td>
<td>2.6</td>
<td>1.6-4.1</td>
<td></td>
</tr>
<tr>
<td>1st deg late CHD, same lineage</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>One 1st deg early CHD</td>
<td>3.6</td>
<td>2.6-4.9</td>
<td></td>
</tr>
<tr>
<td>Two 1st deg late CHD, same</td>
<td>2.5</td>
<td>1.4-4.4</td>
<td></td>
</tr>
<tr>
<td>lineage</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>One 1st deg late CHD + one</td>
<td>2.4</td>
<td>1.6-3.7</td>
<td></td>
</tr>
<tr>
<td>2nd deg early CHD, same lineage</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>One 1st deg late CHD + two</td>
<td>2.6</td>
<td>1.6-4.3</td>
<td></td>
</tr>
<tr>
<td>2nd deg late CHD, same lineage</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Two 2nd deg early CHD, same</td>
<td>3.3</td>
<td>2.3-4.6</td>
<td></td>
</tr>
<tr>
<td>lineage</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>One 2nd deg early CHD + two</td>
<td>3.6</td>
<td>2.4-5.5</td>
<td></td>
</tr>
<tr>
<td>2nd deg late CHD, same lineage</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Moderate Familial Risk</strong></td>
<td>471 (12%); 1.25 criteria met</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Only . . .</td>
<td></td>
<td></td>
</tr>
<tr>
<td>One 1st deg late CHD + one</td>
<td>1.6</td>
<td>0.8-3.0</td>
<td></td>
</tr>
<tr>
<td>2nd deg late CHD, same lineage</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Mother and father late CHD</td>
<td>2.2</td>
<td>1.2-4.0</td>
<td></td>
</tr>
<tr>
<td>One 1st deg late CHD</td>
<td>2.1</td>
<td>1.5-2.9</td>
<td></td>
</tr>
<tr>
<td>One 2nd deg early CHD + one</td>
<td>1.1</td>
<td>0.5-2.2</td>
<td></td>
</tr>
<tr>
<td>2nd deg late CHD, same lineage</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Two 2nd deg late CHD, same</td>
<td>2.5</td>
<td>1.7-3.6</td>
<td></td>
</tr>
<tr>
<td>lineage</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Weak Familial Risk</strong></td>
<td>2291 (57%); 1.01 criteria met</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Only . . .</td>
<td></td>
<td></td>
</tr>
<tr>
<td>One 2nd deg early CHD (one</td>
<td>1.5</td>
<td>0.6-4.1</td>
<td></td>
</tr>
<tr>
<td>or both sides of family)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>One 2nd deg late CHD (one</td>
<td>0.9</td>
<td>0.3-3.0</td>
<td></td>
</tr>
<tr>
<td>or both sides of family)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>No family history of CHD</td>
<td>1.0</td>
<td></td>
<td>(referent group)</td>
</tr>
</tbody>
</table>
Determine if FHx modifies the association between disease and other risk factors

Diabetes Risk Factors by Family History
Risk Category

<table>
<thead>
<tr>
<th>Risk factors*</th>
<th>Average risk (n=2791)</th>
<th>Moderate risk (n=843)</th>
<th>High risk (n=711)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Obesity (BMI &gt;=30)</td>
<td>14%</td>
<td>19%</td>
<td>27%</td>
</tr>
<tr>
<td>Did not meet exercise guidelines</td>
<td>62%</td>
<td>63%</td>
<td>60%</td>
</tr>
</tbody>
</table>

*Mantel-Haenszel Chi square test for equal proportions significant at p <0.001 for all variables

Hariri et al. Healthstyles 2004
Assess associations between familial risk and risk-reducing behaviors

Risk-reducing and Risk-aware Behaviors by Family History Category

- Collected information for family history
- Made lifestyle changes to prevent DM
- Tested in past 2 years
- Talked about diabetes with family

Percent

Average  Moderate  High

Hariri et al. Healthstyles 2004
Examine knowledge, attitudes, and practice of collecting family histories – individuals and clinicians

Doctor informed respondent of familial DM risk by behaviors

Qureshi et al. Healthstyles 2004
## Family history data collection

- Need to standardize questions and modules

<table>
<thead>
<tr>
<th>Have any of your blood relatives ever been diagnosed with asthma?</th>
</tr>
</thead>
<tbody>
<tr>
<td>Yes</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Have any of your first degree relatives (mother, father, siblings, children) ever been diagnosed with asthma?</th>
</tr>
</thead>
<tbody>
<tr>
<td>Yes</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>How many of your first degree relatives were diagnosed with asthma?</th>
</tr>
</thead>
<tbody>
<tr>
<td>One</td>
</tr>
</tbody>
</table>
1. Have you ever been diagnosed by your doctor as having coronary heart disease (e.g., myocardial infarction, coronary bypass graft surgery or angioplasty)?

2. Has your mother ever been diagnosed as having coronary heart disease?

3. Has your father ever been diagnosed as having coronary heart disease?

Yes, at or before aged 60      Yes, after age 60      NO      DK
4. How many of your brothers and sisters have been diagnosed with coronary heart disease **at or before age 60**?

5. How many of your brothers and sisters have been diagnosed with coronary heart disease **after age 60**?

6. How many of your mother’s relatives (her sisters, brothers and parents) were diagnosed with coronary heart disease **at or before age 60**?

7. How many of your mother’s relatives (her sisters, brothers and parents) were diagnosed with coronary heart disease **after age 60**?

8. How many of your father’s relatives (his sisters, brothers and parents) were diagnosed with coronary heart disease **at or before age 60**?

9. How many of your father’s relatives (his sisters, brothers and parents) were diagnosed with coronary heart disease **after age 60**?

<table>
<thead>
<tr>
<th>None</th>
<th>One</th>
<th>Two or more</th>
<th>DK</th>
</tr>
</thead>
</table>
Increasing awareness about family history in the community

- Convene community presentation/discussions – churches, libraries, schools, hospitals, health fairs, etc

- Develop resource packets - videos, powerpoint presentations, case studies, fact sheets, family history tools, etc

- Partner with local companies (Olin Mills, Sears, Penny’s, etc) to include family history themed materials in their studios.

- Develop materials for doctors offices, hospitals and clinics
Increasing awareness about family history in the community

- Sponsor poster contest for students
- Develop continuing education modules for health dept personnel
- Write articles for local newspapers (include local case studies)
- Identify a champion to work with
- Your ideas???
How can you collect your family history?

- Ask questions
- Talk at family gatherings
- Draw a family tree
- Write down the information
- Look at death certificates, family medical records, etc.
What information do you need?

- Major medical conditions and causes of death
- Age when disease diagnosed and age at death
- Ethnic background
- Lifestyle factors like heavy drinking and smoking

*Example:*

Uncle Bill – smoked cigarettes since he was a teenager, had a heart attack at age 52
What relatives should be included?

- you
- children
- siblings
- parents
- grandparents
- aunts and uncles
- nieces and nephews
What should you do with the information?

- Organize the information
- Share it with your doctor
- Keep it updated
- Pass it on to your children
What will your doctor do with the information?

- Determine your risk for disease based on:
  - Number of family members with the disease
  - The age when they were diagnosed
  - How you are related to the family member

- Consider other disease risk factors

- Recommend screening tests and lifestyle changes
Incorporate family history in prevention/risk reduction activities

Example - Assessment of BMI in schools:

- Identify affected sibships (families at highest risk i.e., those with more than one overweight child in the school system)

- Conduct assessment of additional risk factors and health problems including family history of chronic diseases associated with obesity (cardiovascular disease, diabetes, cancer)

- Work with high risk families to change their unhealthy behaviors and increase screening uptake. Engage local health departments, schools, YMCA's, etc.
For more information

Paula Yoon
Contact: pyoon@cdc.gov

Office of Genomics and Disease Prevention
Website: http://www.cdc.gov/genomics