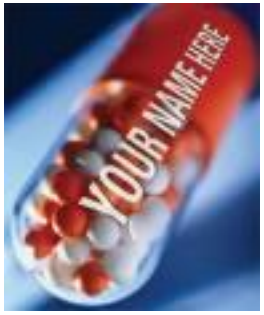

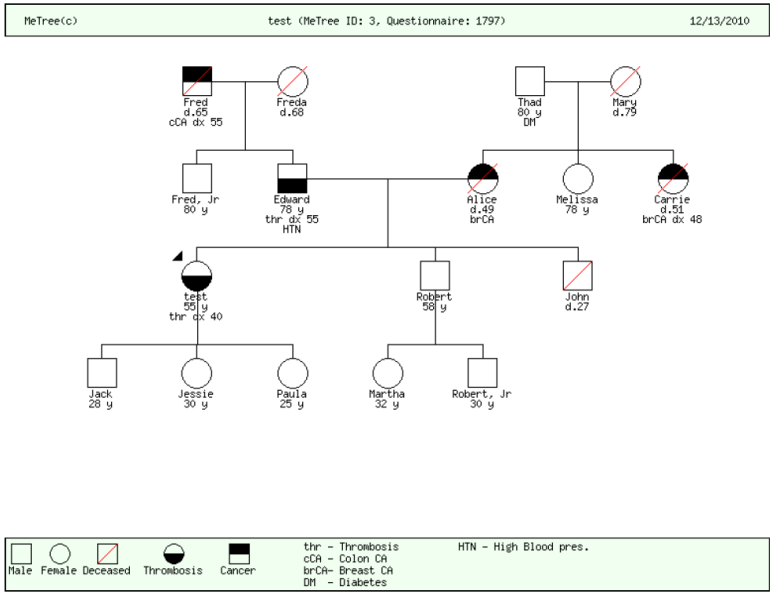

Family History Working Group Update: "Breakthroughs in Healthcare"

Genomic Medicine Meeting III

May 4, 2012

Family History WG Meetings

Goals:

- To develop and agenda to advance family health history use in the clinic
- To develop ideas for that may be responsive to an RFA or that could be initiated by this group

- Dec 2011 – GM II
- Jan 2012
- Feb 2012
- April 2012

Why?

"Bummer of a birthmark, Hal."

Who?

- David Adam
- Jonas Almeida
- Ebony Bookman
- Deanna Cross
- Adam Davis
- David Dimmock
- Corina Din-Lovinescu
- Andy Faucette
- Greg Feero
- Jennifer Geurtz
- Geoff Ginsburg
- Cathy McCarty

- Lori Orlando
- Diana Paltoo
- Teji Rakhra-Burris
- LH Rogers
- Maren Scheuner
- Maureen Smith
- Jeff Struewing
- Murugu Manickam
- Marc Williams
- Janet Williams
- Graham Wood
- David Valle

Where?

- Duke University
- Geisinger Health System (E)
- Essentia Institute for Rural Health
- Intermountain Health Care
- Johns Hopkins University
- Marshfield Clinic (E)
- Medical College of Wisconsin
- Morehouse
- NHGRI
- NHLBI
- Northwestern University (E)
- Ohio State University
- University of Alabama
- Veterans Administration

Dec 2011 - Topics

- Develop an outcomes research agenda
- Implementation science to integrate FH into the clinical workflow
- Advisory Group on FH
- Information interface and education of providers
- Explore electronic media tools to help patients and families create their own family
- Validation of family history information
- Building risk models with all the data

Dec 2011 - Topics

- Develop an outcomes research agenda
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- Validation of family history information
- Building risk models with all the data

The Bake Off

metree™

meTree™

Welcome to MeTree. This program will ask questions about your health and your family's health. Your answers will be used to give you personalized suggestions for your health care. Please answer as best you can.

TOUCH HERE TO START





MeTree

- Collects 3 generation family history
  48 diseases
- Decision support for 4 pilot diseases:
  Breast cancer
  Colon cancer
  Ovarian cancer
  Thrombosis
- Generates reports:
  Pedigree
  Provider report
  Tabular FH
  Patient report

How many biological (blood) nieces and nephews do you have? Please include any who have died.

How many daughters does Keith have?

(•) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) more than 5

How many sons does Keith have?

( ) 0 (•) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) more than 5

How many daughters does Tracy have?

(•) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) more than 5

How many sons does Tracy have?

(•) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) more than 5

Please fill in the information below.

	First name or nickname	Alive?	Age now **OR** at death (best guess)	I don't hav this info
Your biological mom	Gerry	Yes / No	72 year(s)	No Inf
Grandmother (mom's mother)	Olive	Yes / No	95 year(s)	No Inf
Grandfather (mom's father)	Charles	Yes / No	65 year(s)	No Inf
Aunt (mom's sister)	Ellie	Yes / No	70 year(s)	No Inf
Uncle (mom's brother)	Ray	Yes / No	65 year(s)	No Inf

Which family members did you talk to about your family history?

- [x] Gerry (mother)
- [ ] Roy (father)
- [ ] Tyler (son)
- [ ] Keith (brother)
- [ ] Tracy (brother)
- [ ] Ellie (aunt mom's side)
- [ ] Ray (uncle mom's side)
- [x] Roe (aunt dad's side)

PREVIOUS

Continued on next page

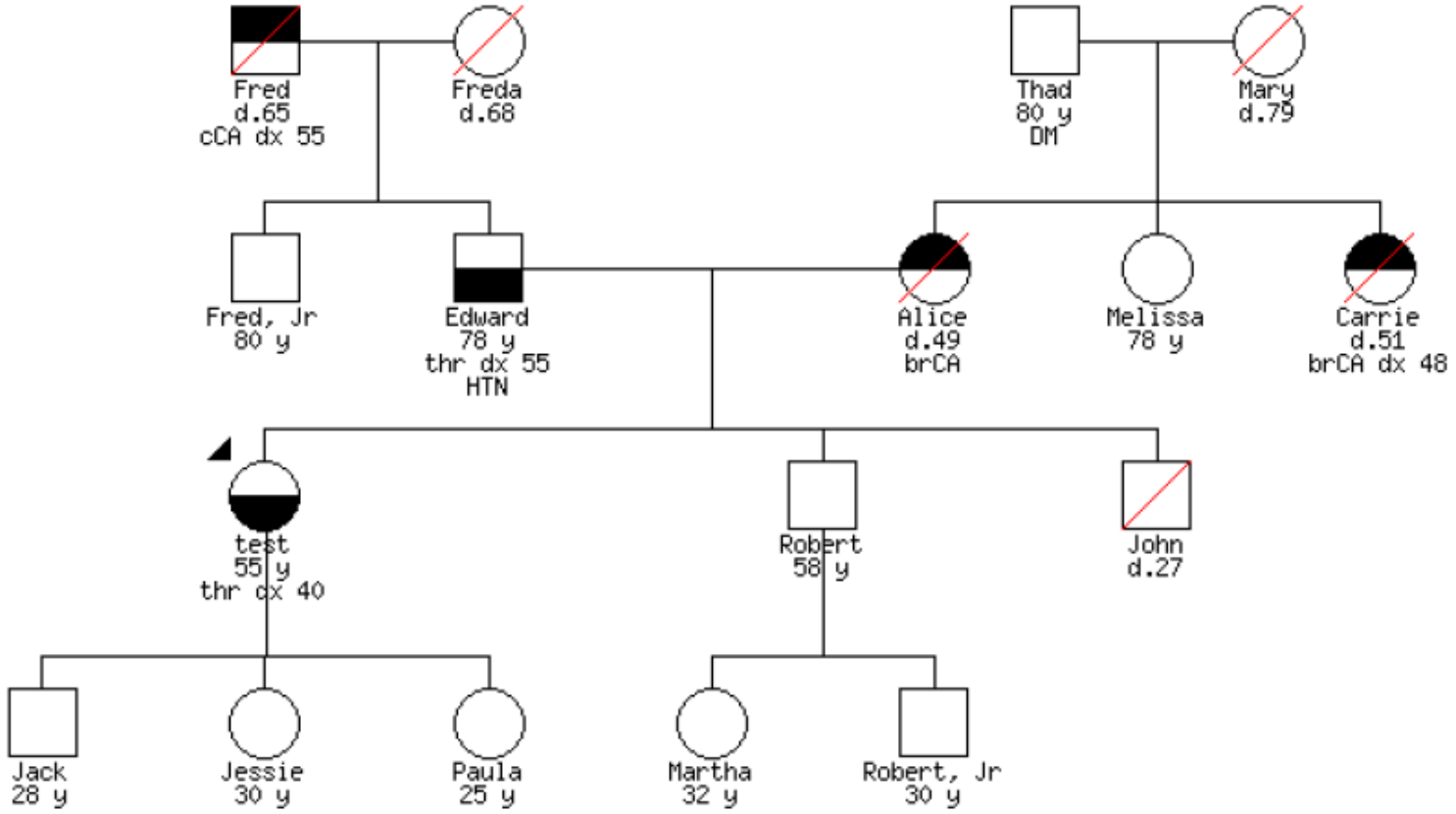
NEXT

What new information did you learn?

Select all that apply.

- [ ] I did not learn anything new
- [ ] Some relatives had diseases I did not know about
- [ ] I was mistaken about which diseases some relatives had
- [x] I learned how some relatives died
- [ ] More relatives had a disease than I had realized
- [ ] Some relative's diseases were more severe than I had realized
- [ ] Someone I thought was a blood relative is not
- [ ] I learned how old relatives were when they got a disease

MeTree(c) test (MeTree ID: 3, Questionnaire: 1797) 12/13/2010



Male Female Deceased Thrombosis Cancer

thr - Thrombosis
cCA - Colon CA
brCA- Breast CA
DM - Diabetes

HTN - High Blood pres.

Tabular Family History Display Example (partial report):

MeTree ID: 12012

Relation	Firstname	Age	Cause of death	Breast/O varian	CRC	Other cancers	Thrombosis related	Chemo prev	General health
self		64				None / skin cancer /		stroke /	high bp / high cholesterol / stroke / None /
sister		51				None /			None /
brother		68				None /			high bp / None /
brother		62				None / skin cancer /			high bp / None /
brother		60				None /			high bp / None /
niece		20				None /			None /
niece		32				None /			None /
niece		26				None /			None /
niece		22				None /			None /
nephew		34				None / skin cancer /			None /
nephew		28				None /			high bp / high cholesterol / None /
father		91				None / skin cancer /			high bp / high cholesterol / None /
paternal Uncle		d85	heart /			None /			None /
paternal Grandfather		d69	unk /			None /			alzheimer's disease / None /

Sample Patient Report

Talk With Your Doctor About: | **Why?** | **More Information**

MeTree© Personalized Profile for test user (ID: 3) based on your answers to Questionnaire #1797 on 12/13/2010

Talk to your doctor about:	Why?	More information
Referral to a genetic counselor	There's an increased chance that cancer runs in your family for these reasons. **You have:** • At least 2 relatives with breast cancer. • At least 1 relative who was diagnosed with breast cancer at age 50 or younger.	• Talk with your doctor about how the chance of cancer running in your family affects your cancer screening plans. A genetic counselor will talk to you about: 1. Your chances of getting certain cancers. 2. Factors that can affect these chances. 3. Best ways for you to find or prevent cancer. 4. Testing for cancer genes. 5. Your family members' risks.
Regular colon cancer screening	Your chances of colon cancer increase with age. This is why most people should have regular screening beginning at age 50.	Several colon cancer screening tests have been shown to be effective. Talk with your doctor about the one that's right for you.

The information is based on facts you entered into MeTree©. It may not be accurate if facts are not correct. This program does not take into account all factors that may influence disease risk. Talk with your doctor about how other factors, such as health habits, influence disease risk. Based on your needs, a genetic counselor may suggest additional screenings that are not included in this report.

DukeMedicine

DUKE INSTITUTE FOR GENOME SCIENCES & POLICY

02/25/2010 MeTree Personalized Risk Profile

MeTree ID: #1234
Questionnaire: #9999
Patient X
DOB: 5/25/1965 Age: 41
BMI: 28

ACTIONABLE ITEMS

- Refer to genetic counseling for comprehensive INHERITED THROMBOPHILIA risk assessment & management [1]
- Refer to genetic counseling for comprehensive CANCER risk assessment & management [2,3,7-9]
- Coordinate risk management for HNPCC syndrome according to NCCN guidelines (www.nccn.org)
- Discuss chemoprevention for breast cancer (tamoxifen) [5,6]

INDICATIONS

Personal History

- Venous thrombosis in unusual location (head, neck, arm or abdomen).
- Patient meets Amsterdam II criteria for clinical diagnosis of HNPCC syndrome.
- Patient's 5-year breast cancer risk (Gail model estimate = ____%) exceeds cut-off of 1.65%.

Family History

- At least 1 first-degree relative was diagnosed with colorectal cancer < age 50.
- At least 3 relatives with HNPCC-related cancers (colorectal, uterine, gastric, ovarian, renal, small bowel, pancreatic, brain).

Contraindication(s)/Other Factors to Consider:

- Patient using oral estrogen or progesterone.
- Patient has had stroke.
- Patient has had blood clot(s).
- Refer to pedigree for additional indication(s) relating to thromboembolism

NOTE(S)

NOTE(S):

- Tamoxifen's effectiveness for breast cancer chemoprevention has not been tested in women who are under age 35, pregnant, breastfeeding, or taking hormone replacement therapy.
- Tamoxifen is associated with increased risk of endometrial cancer and thromboembolic events.
- Check patient's previous tamoxifen use.

MeTree© Assessment Tool recommendations are based on information supplied by patient. They may not represent a complete clinical assessment and are not intended to supplant physician discretion in risk management. Based on your needs, a genetic counselor may suggest additional screenings that are not included in this report.

1Chest Guidelines Chest 126; 3 September 2004 Supplement 401S
2U.S. Preventive Services Task Force. Ann Intern Med. 2005;143:355-61.
3Hampel H et al. J Med Genet. 2004;41:81-91.
4Smith RA et al. CA Cancer J Clin. 2006;56:161-79.
5Fisher B et al. J Natl Cancer Inst 1998;90:1371-88.
6Vogel VG et al. JAMA. 2006;295:E1-E15
7Berliner JL et al. J Genet Counsel. 2007;16:241-60.
8Levin B et al. CA Cancer J Clin. 2008;58:130-60.
9Vasen HF et al. Gastroenterology. 1999;116:1453-6.
10National Comprehensive Cancer Network. 2008. http://www.nccn.org/professionals/physician_gls/
11Saslow D et al., CA Cancer J Clin. 2007;57:75-89.
12Berry DA, et al., J Clin Oncol. 2002;20:2701-2712.

Recommended Actions

Indications

Points to Consider

Home

my HEALTH CARE

- Medical Records
- Appointments
- Our Family Health
- Advance Care Planning
- Help
- Health Resources
- Patient Guidelines / FAQs
- Hospital Bills
- Emergency Medical Card / Continuity of Care Record

Connecting Your Family For Better Health

Our Family Health is a new tool from Intermountain Healthcare, built to help you understand the diseases that run in your family. Click here to get started!

Use this new program to:

- Build your family tree.
- Add health and disease information to each person.
- Share with your family members and doctor.

What is family health history?

Your family health history is a collection of information about diseases that run in your family. It also includes the eating habits, activities, and environments that your family shares.

How can family health history affect my health?

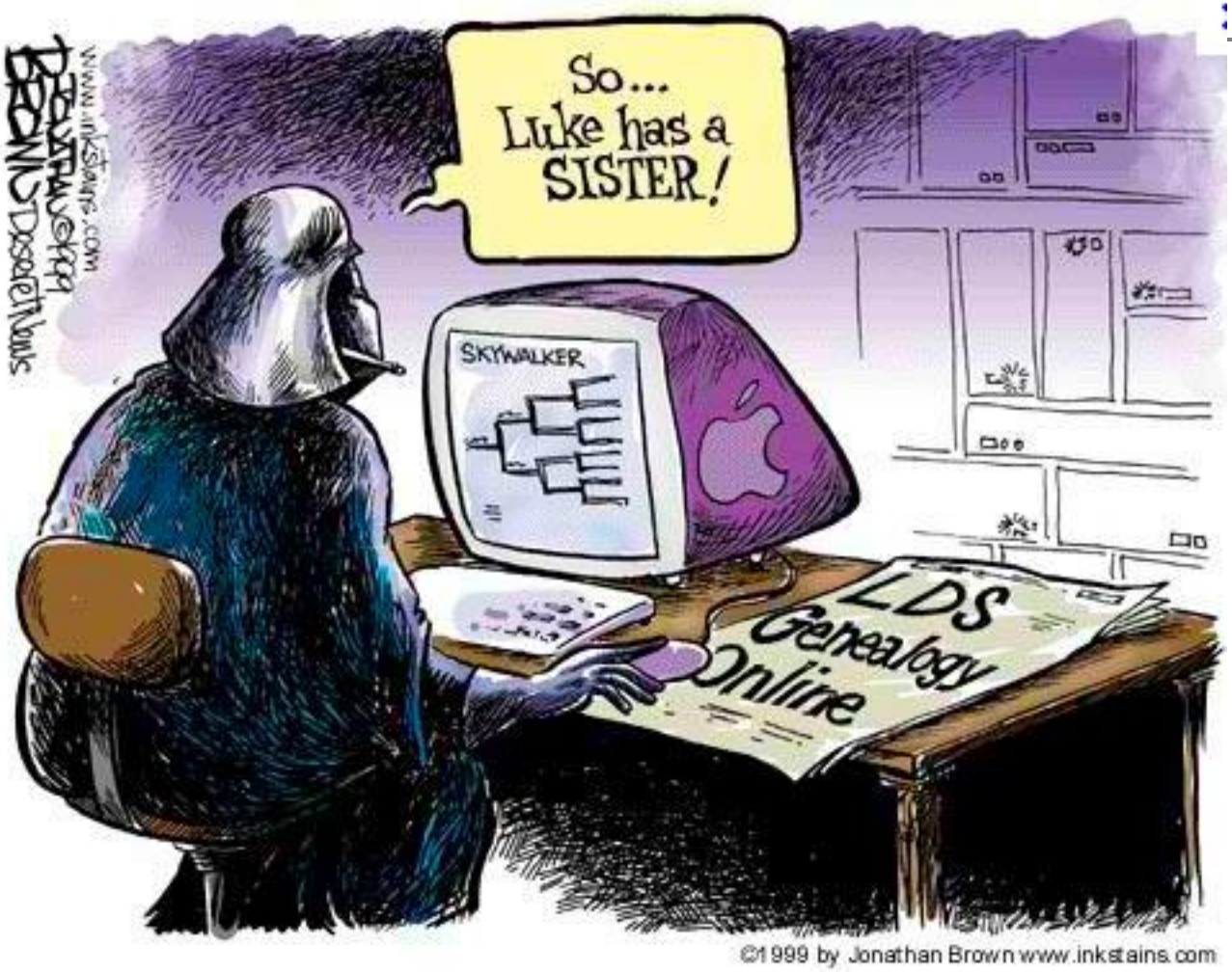
Some diseases and health issues are more likely than others to be passed down through families so it's important to know your own family health history. Knowing details about the diseases that run in your family can help you make healthy choices.

Your doctor can use your family health history to understand your risk for developing a disease. With it, they can personalize the care you receive. They can also recommend custom screenings, medications, and health habits to help you prevent disease.

Booklets from the Genetic Alliance to help you start and manage your family health history:

- A Guide to Family Health History
- A Guide for Understanding Genetics and Health

This program is a project sponsored by The Clinical Genetics Institute at LDS Hosiptal


So...
Luke has a
SISTER!
SKYWALKER
LDS
Genealogy
Online
©1999 by Jonathan Brown www.inkstains.com

Family Information

Let's get started... It's as easy as 1, 2 or 3. Choose an option below:

1. Quick Start

Let's get started with just me and my parents.

You can always add more people to the tree by clicking on the triangle arrows.

Start Now!

OR

2. Brothers, Sisters & Kids

Tell us a little about your family and we will help get your tree started. Only count immediate family members here, we'll add half-siblings and step-relatives later.

# of Brothers: 0

# of Sisters: 0

# of Sons: 0

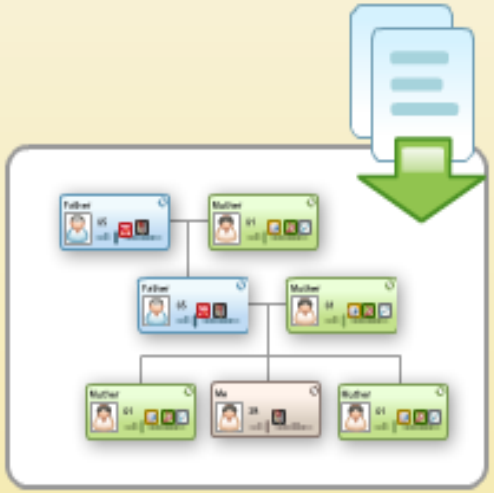
# of Daughters: 0

Let's Go!

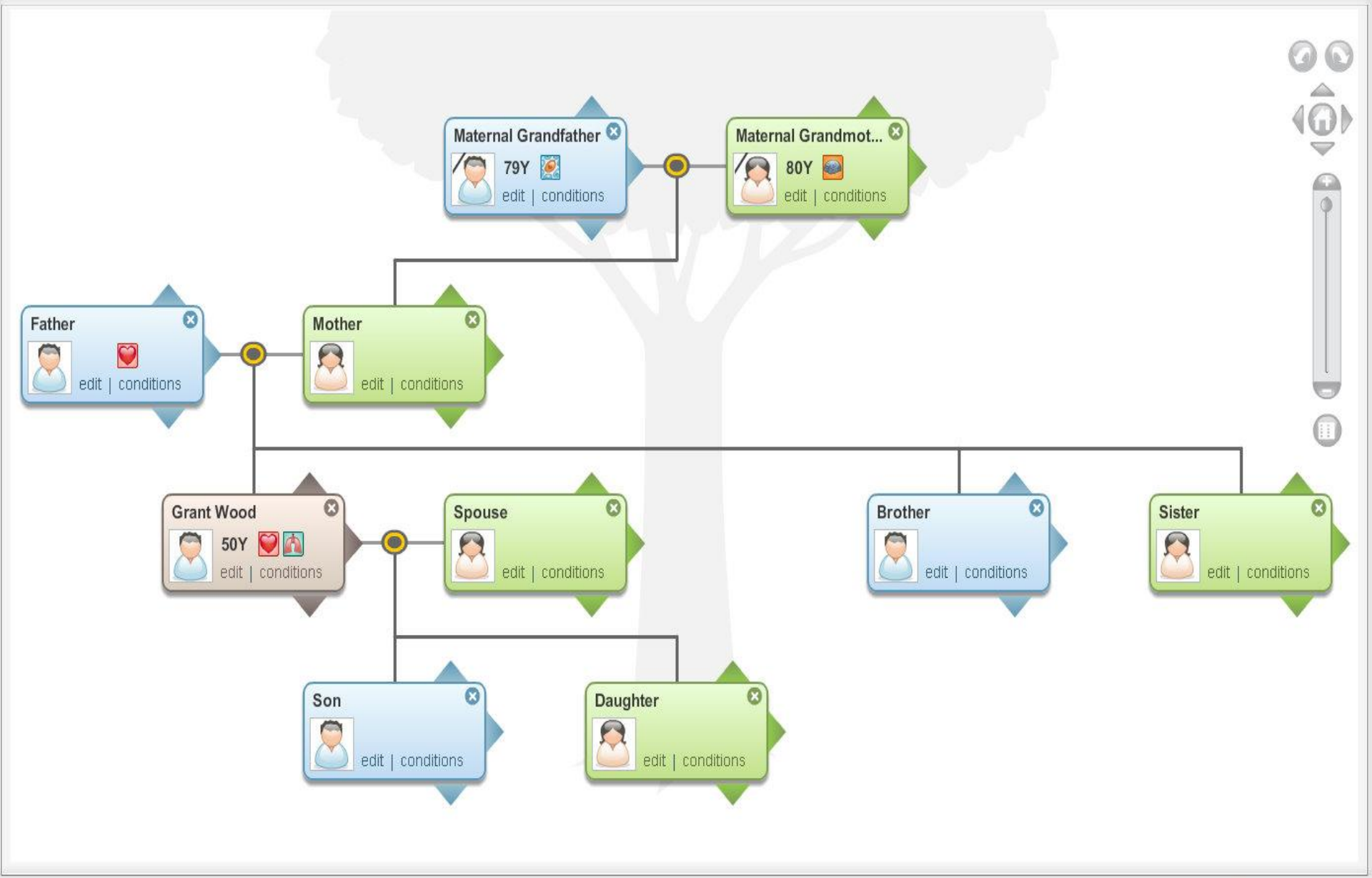
OR

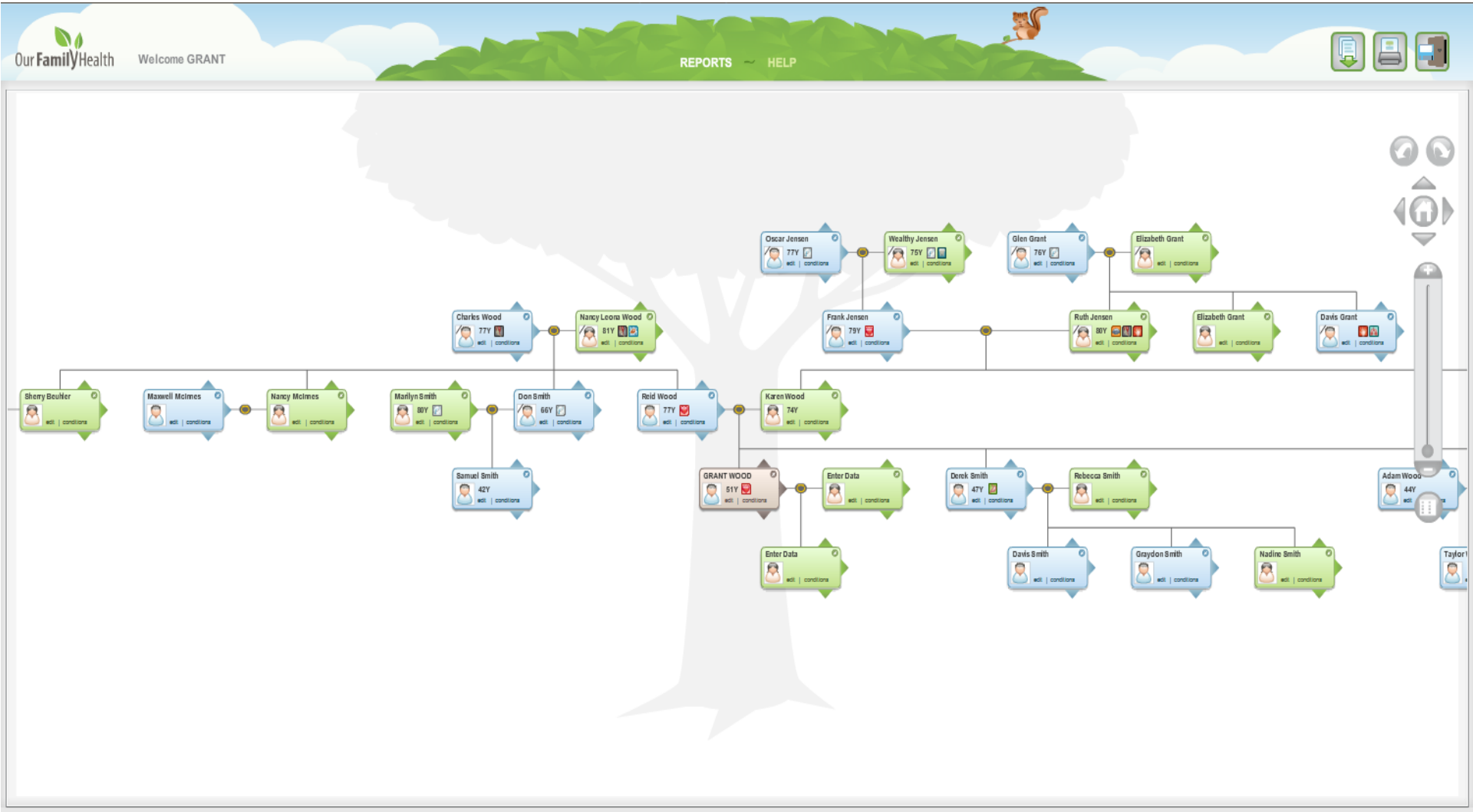
3. Start With Genealogy Data

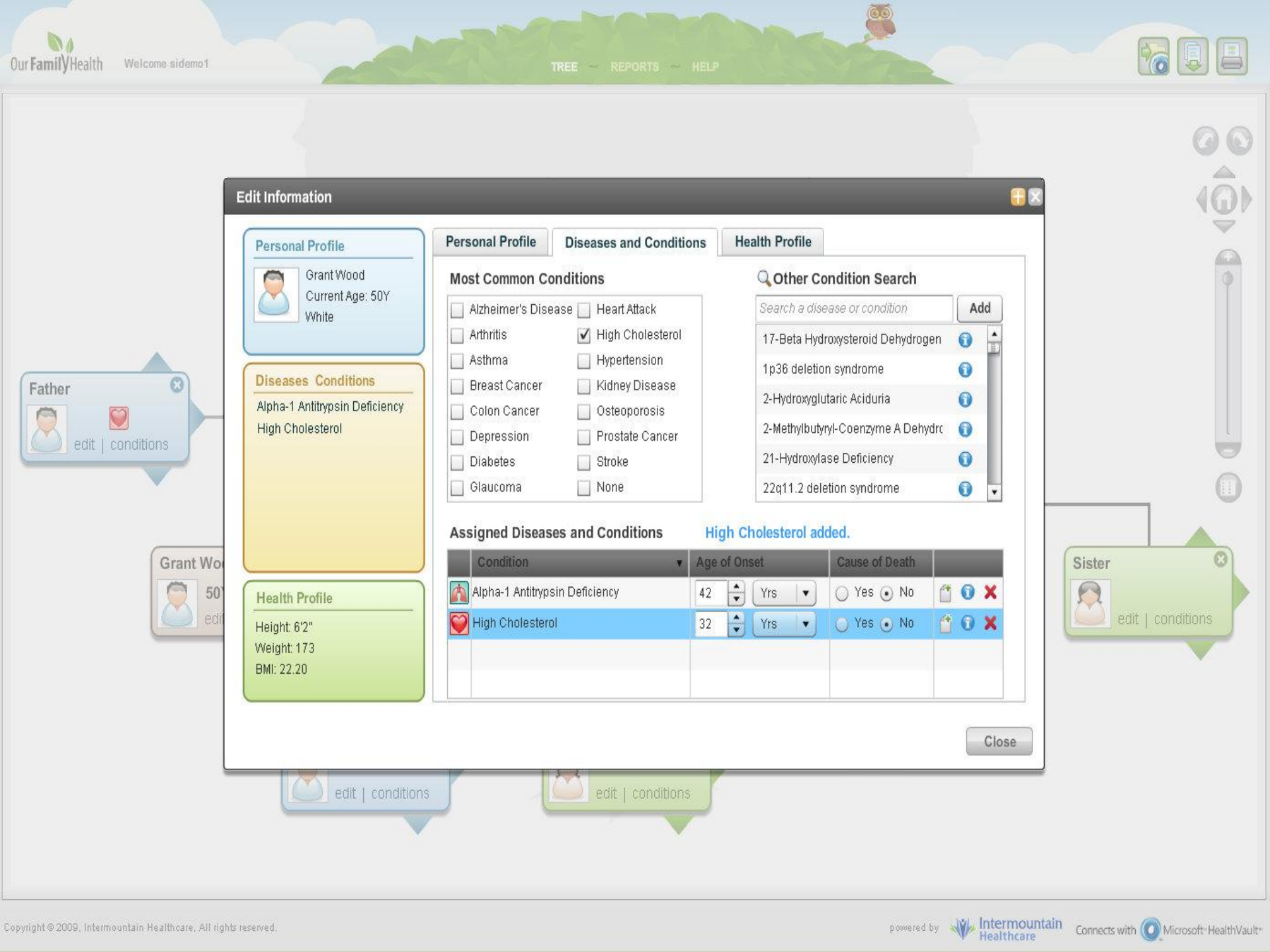
Many people store their family histories on the computer in GEDCOM files. If you have a GEDCOM file from another site you may import it here to get a quick start.



Import GEDCOM File




Our Family Health
Welcome GRANT
REPORTS
HELP
Oscar Jensen
77Y
Wealthy Jensen
75Y
Glen Grant
76Y
Elizabeth Grant
Charles Wood
77Y
Nancy Leona Wood
81Y
Frank Jensen
79Y
Ruth Jensen
80Y
Elizabeth Grant
Davis Grant
Sherry Beuhler
Maxwell Mcimes
Nancy Mcimes
Marilyn Smith
80Y
Don Smith
66Y
Reid Wood
77Y
Karen Wood
74Y
Samuel Smith
42Y
GRANT WOOD
51Y
Enter Data
Derek Smith
47Y
Rebecca Smith
Adam Wood
44Y
Enter Data
Davis Smith
Graydon Smith
Nadine Smith


Our FamilyHealth
Welcome sidemo1
TREE ~ REPORTS ~ HELP
Edit Information
Personal Profile
Grant Wood
Current Age: 50Y
White
Diseases Conditions
Alpha-1 Antitrypsin Deficiency
High Cholesterol
Health Profile
Height: 6'2"
Weight: 173
BMI: 22.20
Personal Profile
Diseases and Conditions
Health Profile
Most Common Conditions
Alzheimer's Disease
Arthritis
Asthma
Breast Cancer
Colon Cancer
Depression
Diabetes
Glaucoma
Heart Attack
High Cholesterol
Hypertension
Kidney Disease
Osteoporosis
Prostate Cancer
Stroke
None
Other Condition Search
Search a disease or condition
Add
17-Beta Hydroxysteroid Dehydrogen
1p36 deletion syndrome
2-Hydroxyglutaric Aciduria
2-Methylbutyryl-Coenzyme A Dehydrc
21-Hydroxylase Deficiency
22q11.2 deletion syndrome
Assigned Diseases and Conditions
High Cholesterol added.
Condition
Age of Onset
Cause of Death
Alpha-1 Antitrypsin Deficiency
42
Yrs
Yes
No
High Cholesterol
32
Yrs
Yes
No
Close
Father
edit | conditions
Grant Wo
Sister
edit | conditions
edit | conditions
edit | conditions
Copyright © 2009, Intermountain Healthcare, All rights reserved.
powered by Intermountain Healthcare
Connects with Microsoft HealthVault

GRANT WOOD - Family Health History Report - Family View

Last updated: April 30, 2012

Intermountain Healthcare
Our FamilY Health

Relationship	Name	Age*	Alive?	Disease Name	Age of Onset	Notes
Me and My Immediate Family						
Me	GRANT WOOD	51	Y	High Cholesterol	18	
Spouse						
Father	Reid Wood	77	Y	High Cholesterol	18	
				Heart Attack	66	
Mother	Karen Wood	74	Y			
Brother	Derek Wood	47	Y	High Cholesterol		
Brother	Adam Wood	44	Y			
Brother	Aaron Wood	41	Y			
Brother	Daniel Wood	34	Y			
Sister-in-law	Kym Wood					
Sister-in-law	Rebecca Wood					
Sister-in-law	Esther Wood		Y			
Sister-in-law	Laurie Wood		Y			
Sister	Heather Brown	52	Y			
Sister	Stephanie Wood	50	Y	Celiac Disease		
Sister	Elizabeth Fleshner	36	Y			
Brother-in-law	Robert Brown					
Brother-in-law	Heath Fleshner		Y			

*Indicates current age if alive, age at death if deceased

Please remember to keep your history up to date.

Function	MeTree®	Our Family Health™
Patient-entered	+	+
Web-enabled	+	+
Includes all AHIC elements	Under evaluation	+
HL-7 v.2.x compatible	+	+
Uploads data to risk stratification algorithms	+	Explore whether tool would do this or would data come from EDW after storage
Stores FHH in EDW	+	+
Exports information to EHR	Pilot underway	+ but not implemented
Imports clinical data on patient	Pilot underway	+ limited to age and sex at present
Links to patient education materials	+	+ using context specific approach
Clinical Decision Support for patient	+	+ limited to providing information on Smoking Quit Line at present
Clinical Decision Support for providers	+	Under evaluation

MeTree
vs
Our Family Health

Idea # 1: STTR/SBIR with EPIC

Overall Goal: To evaluate the ability to integrate family history software and decision support tools with an electronic medical record

- Standards
- Identify the gaps
- Examine the workflow
- Meaningful use (Medicare/Medicaid EHR Incentive Program)
- Collection methods
- Validate the information
- Integration of third party applications
- Interoperability
- Representation of the FH in the EMR – making it readable

- Recommendation: To link to Clinical Decision Support Consortium
- http://www.partners.org/cird/cdsc/
- Open CDS www.opencds.org

Idea # 2: Social Networking/Computing for FHH Data Acquisition

Overall Goal: To use social media to capture family history data and transmit to providers

- Application Program Interface established – Jonas Almeida UAB
- Informatics research prototype
- Cloud computing to collect and manage FHH
- Hosting and control of the process entirely by the patient
- Can use the Surgeon General's FHH Tool
- Connected distribution of FHH among those being described
- GitHub and Google Code – deliver the FHH application to the patient's Google.com account
- Partnership with provider organization to access information

http://www.youtube.com/watch?v=KwNBgyO2gzI

Idea #3: FHH Intervention and Outcomes

Overall Goals:

- To optimize the collection of patient-entered electronic FHH data and its export to clinical decision support tools and into the EHR.
- To measure and demonstrate improved outcomes as a result of an FHH intervention at various stakeholder levels

- Settings:
  - Primary care and emergency department
  - Rural practices
  - Underserved practices
  - Educational (teaching hospital) practices
- “Does an intervention work under usual conditions?”
- Probable study design: Pragmatic cluster randomized trial

2012:
FHH Intervention for CVD Risk Assessment

IMPROVING PATIENT CARE | ORIGINAL RESEARCH

Effect of Adding Systematic Family History Enquiry to Cardiovascular Disease Risk Assessment in Primary Care

A Matched-Pair, Cluster Randomized Trial

Nadeem Qureshi, DM; Sarah Armstrong, PhD; Paula Dhiman, MSc; Paula Saukko, PhD; Joan Middlemass, MPhil; Philip H. Evans, MPhil; and Joe Kai, MD, for the ADDFAM (Added Value of Family History in CVD Risk Assessment) Study Group*

Background: Evidence of the value of systematically collecting family history in primary care is limited.

Objective: To evaluate the feasibility of systematically collecting family history of coronary heart disease in primary care and the effect of incorporating these data into cardiovascular risk assessment.

Design: Pragmatic, matched-pair, cluster randomized, controlled trial. (International Standardized Randomized Controlled Trial Number Register: ISRCTN 17943542).

Setting: 24 family practices in the United Kingdom.

Participants: 748 persons aged 30 to 65 years with no previously diagnosed cardiovascular risk, seen between July 2007 and March 2009.

Intervention: Participants in control practices had the usual Framingham-based cardiovascular risk assessment with and without use of existing family history information in their medical records. Participants in intervention practices also completed a questionnaire to systematically collect their family history. All participants were informed of their risk status. Participants with high cardiovascular risk were invited for a consultation.

Measurements: The primary outcome was the proportion of participants with high cardiovascular risk (10-year risk ≥20%). Other measures included questionnaire completion rate and anxiety score.

Results: 98% of participants completed the family history questionnaire. The mean increase in proportion of participants classified as having high cardiovascular risk was 4.8 percentage points in the intervention practices, compared with 0.3 percentage point in control practices when family history from patient records was incorporated. The 4.5–percentage point difference between groups (95% CI, 1.7 to 7.2 percentage points) remained significant after adjustment for participant and practice characteristics ($P = 0.007$). Anxiety scores were similar between groups.

Limitations: Relatively few participants were from ethnic minority or less-educated groups. The potential to explore behavioral change and clinical outcomes was limited. Many data were missing for anxiety scores.

Conclusion: Systematically collecting family history increases the proportion of persons identified as having high cardiovascular risk for further targeted prevention and seems to have little or no effect on anxiety.

Primary Funding Source: Genetics Health Services Research program of the United Kingdom Department of Health.

Ann Intern Med. 2012;156:253-262. www.annals.org

For author affiliations, see end of text.

* For additional members of the ADDFAM study group, see **Appendix 1** (available at www.annals.org).

Major Findings

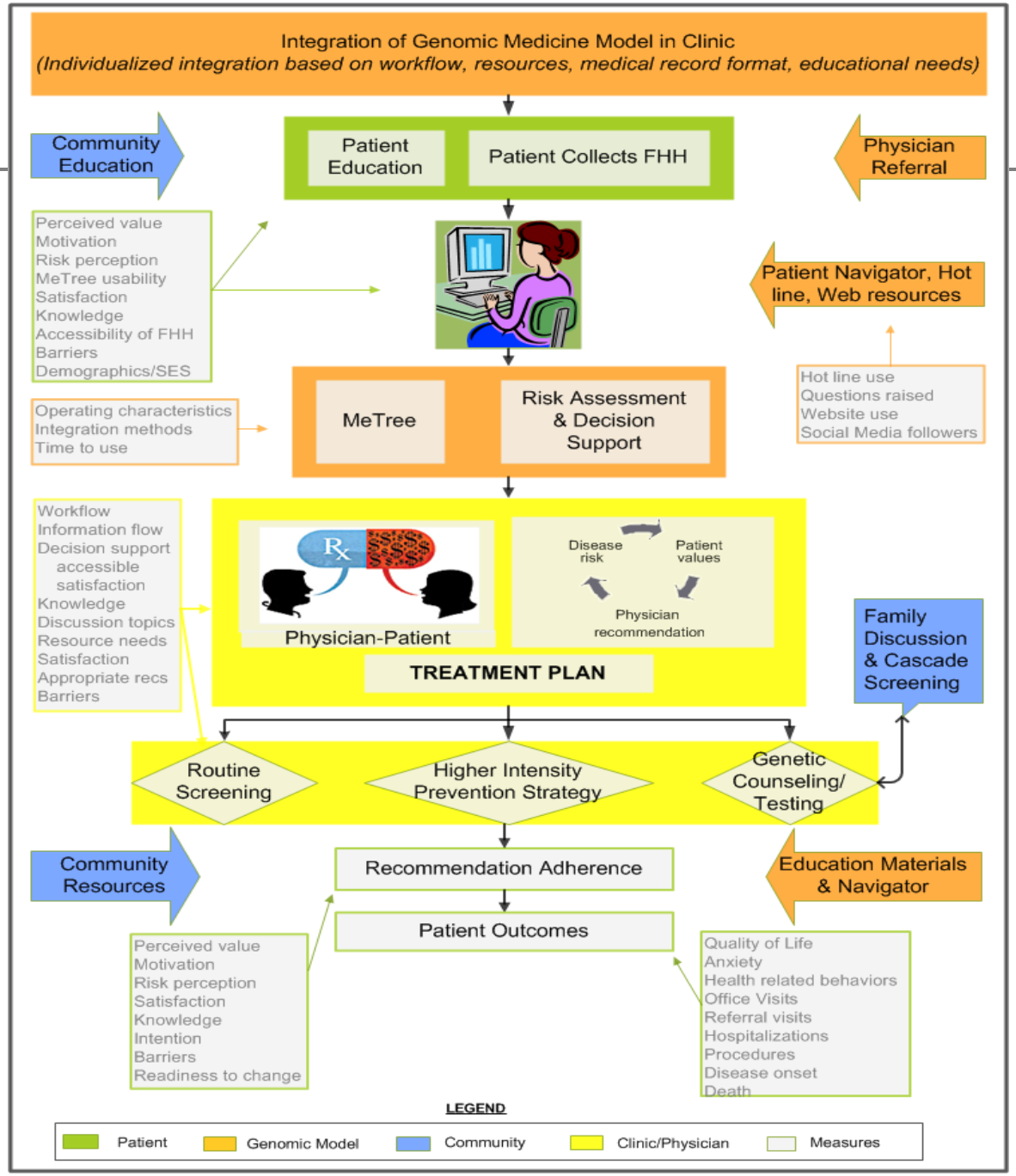
Summary:

- Pragmatic cluster randomized trial
- 748 adult patients
- No previously diagnosed CVD
- 24 primary care practices
- FHH collection using mailed questionnaires
- Identified more high risk patients eligible for targeted intervention than did usual care (4.3% vs 0.3%)
- No long term clinical outcomes

Key Messages:

- Systematic collection of family history data is feasible
- Could improve cardiovascular risk assessment and target patients at high risk for preventive interventions

Information, Participant and Evaluation Flow



Outcome Measures

	Patient	Provider	System
Emotional	• SF-12 (quality of life) • Patient Activation Measure • Prochaska Stage of Change • Satisfaction • Quality of clinical encounter • Perceived/real barriers to Model use • Unanswered questions arising from Model integration	• Satisfaction • Knowledge • Perceived/real barriers to Model use • Concur with decision support • Quality of clinical encounter • Perceived quality of CDS on overall clinical care • Quality/usefulness of CDS output	• Staff satisfaction • Organizational readiness to change (ORCA) • Implementation climate
Behavioral	• Morisky (medication adherence) • Stanford Brief Activity Survey • The Rapid Food Screener • Self-reported Tobacco use • Implemented provider prevention recommendation (uptake)	• Discussion of prevention • Discussion of risk • Screening appropriately • Implemented CDS prevention recommendations • % time CDS output used (uptake)	• Work flow and processes • Implementation policies and practices • Implementation climate • Intervention values and task fit • % up to date on screening • % at ideal weight (BMI) • % exercising 3 days/week • % smoking • % adherence to CDS recommendation by provider • % adherence to provider recommendation by patient
Biological	• Demographics • FHH	• FHH documentation & counseling	• FHH tool completion rate • FHH time to complete • FHH accuracy
Clinical	• Laboratory Data (i.e. LDL) • Screening tests performed (i.e. colonoscopy, breast MRI, mammogram) • Vital Signs • Weight and BMI • Number of medications	• Diagnoses made • Disease control goals met • Referrals made • Medication adverse events	• % high risk patients • % with complications • % at goal for disease control • Visit length • Wait times • Missed appointments
Financial	• Socio-economic status • Medication costs	• Medication classes ordered • Screening test utilization	• Utilization (# office visits, ER visits, hospitalizations, and length of stay) • Resources need for Model sustainability

Next Steps

- More discussion tonight
- Develop subgroups and work plans for 3 ideas (or more)
- Respond to demonstration project RFA
- Link to sequencing WG
  - Mendelian traits
  - Complex traits
- Seek advice from distinguished colleagues and invited guests