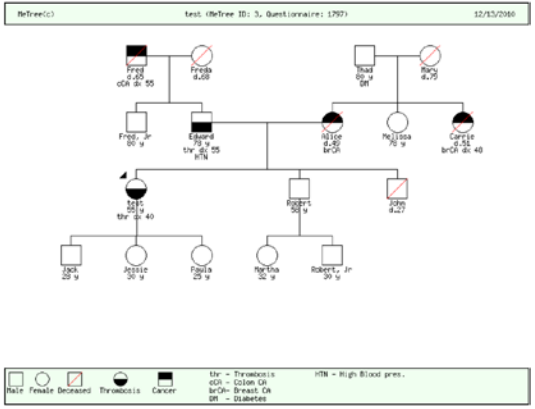


Implementation of Family History in Primary Care

Geoffrey S Ginsburg MD, PhD

Director, Centers for Genomic and Personalized Medicine
Duke University

Genomic Medicine Institutes Colloquium

December 5 2011

DUKE INSTITUTE FOR GENOME SCIENCES & POLICY

DukeMedicine

Family History

"It's the best kept secret in health care."

– Charis Eng

- Captures genetic risk
- Readily available and easy (?) to collect
- Clinical providers are familiar with it and why it is important
- Evidence-based guidelines for screening and treatment exist

… but not effectively used

Use of family history implementation as the basis for developing the clinical genomic model -- "The Genomedical Connection"

Funding: Department of Defense

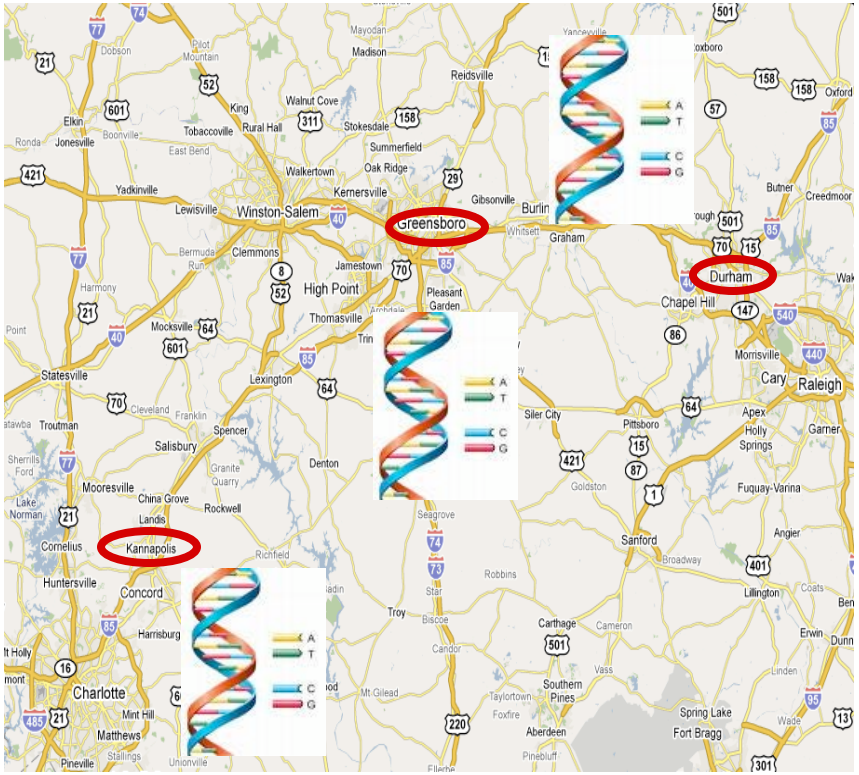
DukeMedicine

DUKE INSTITUTE FOR GENOME SCIENCES & POLICY

The I-85 Corridor for Genomic and Personalized Medicine

Models for Personalized Medicine

- Duke University/Durham
  - T2DM and PGx in Primary Care
  - Personal Genomics
- Moses Cone/UNCG/Duk
  - Family Hx
- Kannapolis
  - The MURDOCK Study



DukeMedicine

DUKE INSTITUTE FOR GENOME SCIENCES & POLICY

Guilford County Family History Project

- Recruitment: All adults scheduled for future 'well visits' at 2 community based primary care practices in Cone Health System
  - 10 & 4 physician group with 31,000 patients
  - Concurrent control primary care practice
  - Enrollment Goal: 1500 patients
- Collaboration between Cone Health, Duke University, University of North Carolina-Greensboro
- Focus on
  - Education
  - Integration into practice
  - Outcomes measurement

DukeMedicine

DUKE INSTITUTE FOR GENOME SCIENCES & POLICY

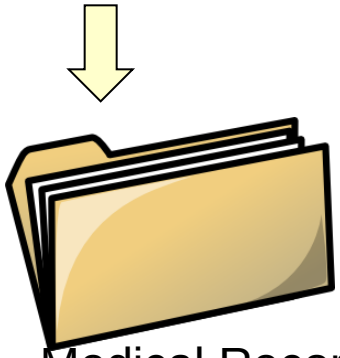
Flow of Family History Information: Current

Patient

Physician

What diseases?
What's the utility?
Cancer syndromes??
What time is it?

Data Processing



Medical Record

Healthcare Plan

DukeMedicine

Data from:
AHRQ Evidence Report (2009). Family History and Improving Health
http://www.ahrq.gov/clinic/tp/famhimptp.htm.

DUKE INSTITUTE FOR GENOME SCIENCES & POLICY

Genomedical Connection Platform

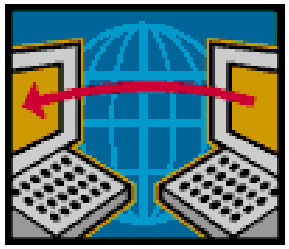
Prepare with worksheet to talk with relatives

Contact available for questions or problems

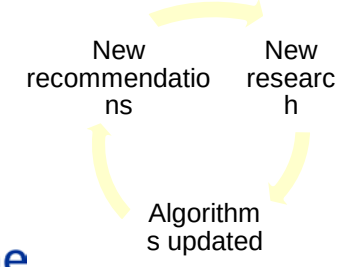
Patient entry from home or clinic *

Update input if incorrect

Family history expertise for questions about recs



Data sent to medical record, processed and report generated



Appointment

Access to genetic counselor, testing, other expertise

Patient-Physician

*

Healthcare Plan

DukeMedicine

DUKE INSTITUTE FOR GENOME SCIENCES & POLICY

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





THE GENOMEDICAL CONNECTION | TALK TO YOUR FAMILY. TALK TO YOUR DOCTOR.

MeTree Development

- Guiding principles
  - Easy for patients to use
  - Risk stratification based on published guidelines
  - Patient & physician reports at appropriate level
  - Reports encourage patient-physician discussion
  - Program constructs & prints family history (pedigree)
  - Give physicians summary report for quick reference
- Development team
  - Genetic counselors, medical geneticists, cardiologist, oncologist, health behaviorist, IT experts

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

DukeMedicine

DUKE INSTITUTE FOR GENOME SCIENCES & POLICY

MeTree Algorithm Sources

Condition	Source(s)
Thrombosis	American College of Chest Physicians[1]
Breast/ ovarian cancer	US Preventive Services Task Force[2] Published expert opinion[3] NSABP & STAR trials[5,6] National Society of Genetic Counselors[7] American Cancer Society[4,11] National Comprehensive Cancer Network[10]
Colorectal cancer	American Cancer Society, US Multi-Society Task Force on Colorectal Cancer, American College of Radiology[8] International Collaborative Group on HNPCC[9] National Comprehensive Cancer Network[10]

1Buller HR et al., Chest 2004; 2U.S. Preventive Services Task Force. Ann Intern Med. 2005; 3Hampel H et al. J Med Genet. 2004; 4Smith RA et al. CA Cancer J Clin. 2008; 5Fisher B et al. J Natl Cancer Inst 1998; 6Vogel VG et al. JAMA. 2006; 7Berliner JL et al. J Genet Counsel. 2007; 8Levin B et al. CA Cancer J Clin. 2008; 9Vasen HF et al. Gastroenterology. 1999; 10National Comprehensive Cancer Network. 2010. http://www.nccn.org/professionals/physician_gls/; 11Saslow D et al., CA Cancer J Clin. 2007

Sample Physician Report

12/13/2010

MeTree© Personalized Risk Profile

MeTree ID: 3
Questionnaire ID: 1797
user, test
DOB: 12/13/1955 Age: 55
BMI: 25.01

RECOMMENDED ACTION(S)

Recommended Actions

- **Refer to genetic counseling** for comprehensive cancer risk assessment & management[2,3,7-9]
- **Coordinate routine colorectal cancer screening**[8]

INDICATION(S)

Indications

Family History

- At least 2 relatives with breast cancer.
- At least 1 relative was diagnosed with breast cancer ≤ age 50.

POINT(S) TO CONSIDER

Points to Consider

- Genetic testing results and genetic counselor consultation may help guide risk management decisions, including whether to recommend following NCCN guidelines for Hereditary Breast and Ovarian Cancer syndrome[10].
- Pt age ≥ 50 yrs.

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.

[1]Chest Guidelines Chest 126; 3 September 2004 Supplement 401S
[2]U.S. Preventive Services Task Force. Ann Intern Med. 2005;143:355-61.
[3]Hampel H et al. J Med Genet. 2004;41:81-91.
[4]Smith RA et al. CA Cancer J Clin. 2008;58:161-79.
[5]Fisher B et al. J Natl Cancer Inst 1998;90:1371-88.
[6]Vogel VG et al. JAMA. 2006;295:E1-E15
[7]Berliner JL et al. J Genet Counsel. 2007;16:241-60.
[8]Levin B et al. CA Cancer J Clin. 2008;58:130-60.
[9]Vasen HF et al. Gastroenterology. 1999;116:1453-6.
[10]National Comprehensive Cancer Network. 2010. http://www.nccn.org/professionals/physician_gls/
[11]Saslow D et al., CA Cancer J Clin. 2007;57:75-89.
[12]Berry DA, et al., J Clin Oncol. 2002;20:2701-2712.
[13]GeneReviews. http://www.ncbi.nlm.nih.gov/sites/GeneTests/review?db=GeneTests

DUKE INSTITUTE FOR GENOME SCIENCES & POLICY

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

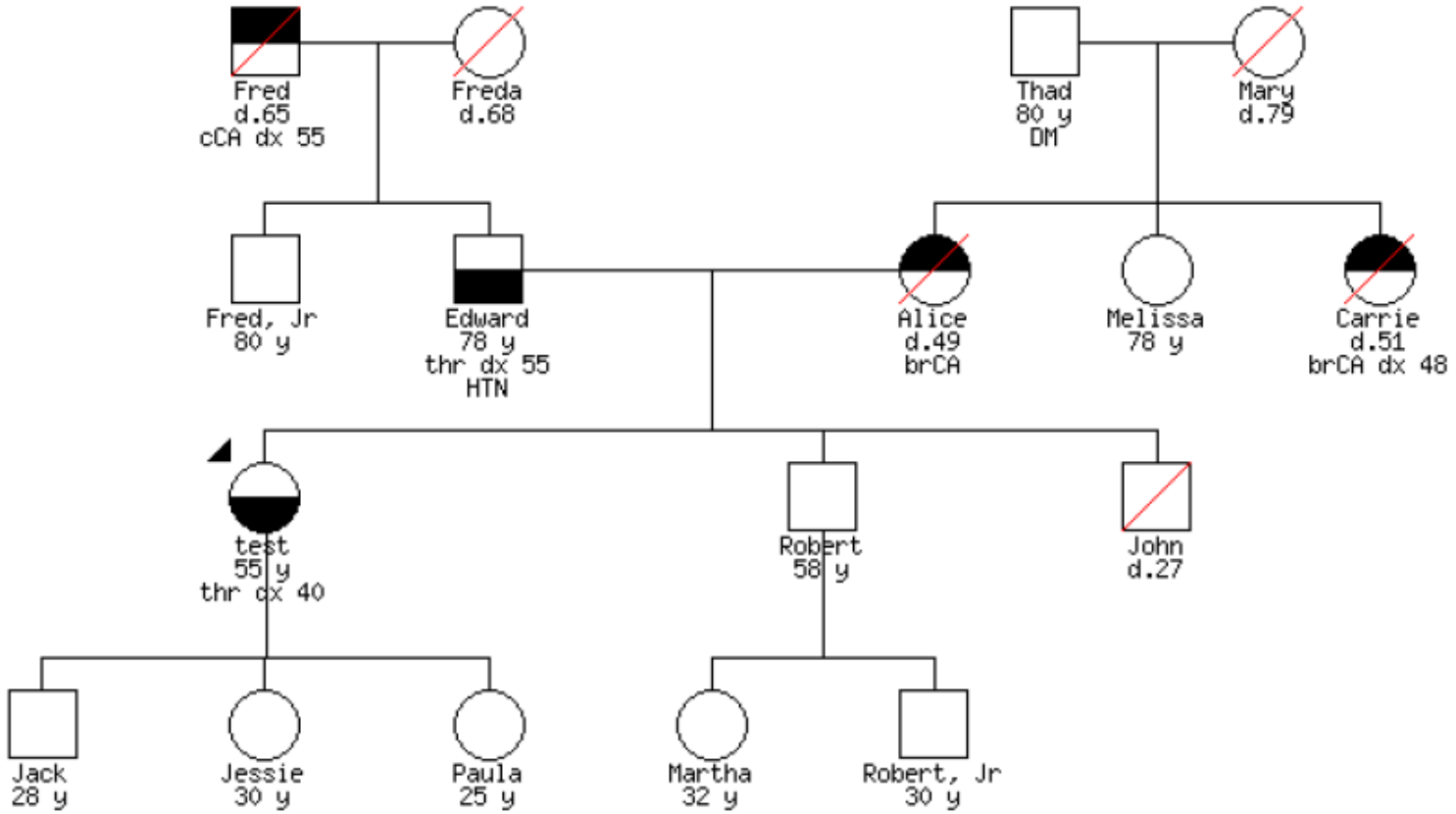
Family History Collection

Tools	Patient Entry	Relatives named	Includes hereditary cancer	Decision support	Integrated in Clinic workflow
GGMI (MeTree)	Yes	Yes	Yes	Yes	Yes
My Family Portrait	Yes	No	No	No	No
VA	No	No	Yes	GC only	Yes
***My Family Healthware**	Yes	No	No	Yes	No
#Organizations (AMA)	Yes	No	No	No	No

*available for research only
#paper based only

DukeMedicine

DUKE INSTITUTE FOR GENOME SCIENCES & POLICY



Male Female Deceased Thrombosis Cancer

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

HTN - High Blood pres.

MeTree© Summary Report

MeTree ID: 3
Questionnaire ID: 1797
Name: test user
Survey date: 12/13/2010
Printed: 12/13/2010

Relation	Firstname	Age	Cause of death	Breast/Ovarian	CRC	Other cancers	Thrombosis related	Chemo prev	General health
self	test	55					blood clots age 40 / blood clots around time of injury /	blood clots /	
brother	John	d27	accident /						
father	Edward	78					blood clots age 55 /		high bp /
paternal Grandfather	Fred	d65	unk /		colon cancer age 55 /				
paternal Grandmother	Freda	d68	unk /						
mother	Alice	d49	cancer /	breast cancer /					
maternal Aunt	Carrie	d51	cancer /	breast cancer age 48 /					
maternal Grandfather	Thad	80							diabetes /
maternal Grandmother	Mary	d79	natural /						

MeTree Pilot Testing

- MeTree tested with lay public (2007)
  - Cognitive testing to make sure questions understood as intended
  - Usability testing to make sure program easy to navigate
  - Round of program revisions post-testing
- Genetic counselors tested MeTree (2008)
  - Asked to enter sample cases & comment on risk stratification, reports
  - Round of program revisions post-testing

Algorithm Updating

- Clinical expertise & literature review to ensure guidelines stay current
- Solicit feedback from physicians
- Genetic counselor reviews sample of reports for glitches
- Development team (content & IT experts) meets to review & fix glitches

Outcomes: Patient, Provider, System

- Process
  - Pt: satisfaction, ease of use; MD: patient flow, usefulness of reports
- Behavior
  - Pt: diet, smoking, knowledge seeking, MD: screenings, referrals
- Clinical validity
  - Sensitivity, specificity
- Clinical utility
  - Net reclassification, cost utilization
- Health (benefits, harms)
  - Appropriate risk based screening,
  - Anxiety, misclassification

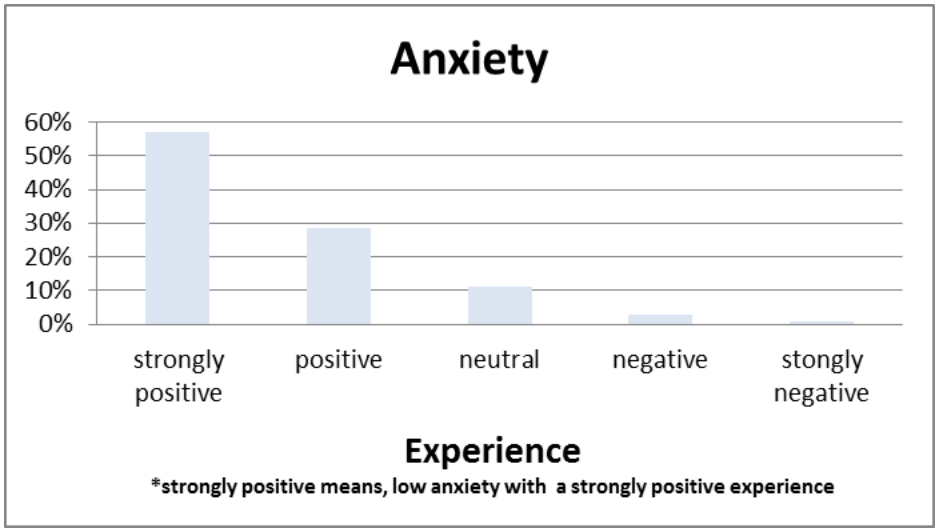
Implementation Outcomes

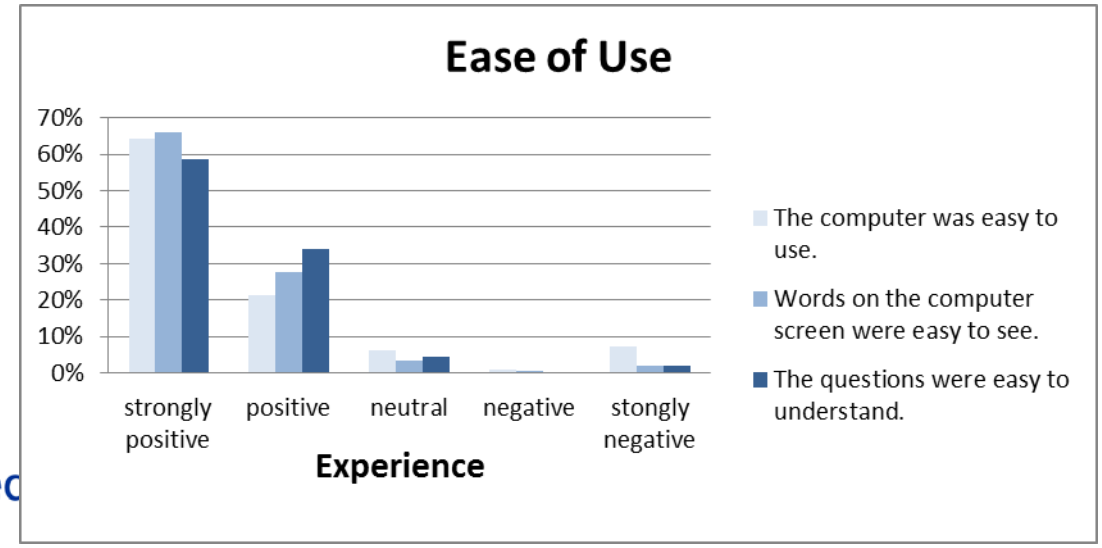
Variable	Measure	Source
Patient experience	Satisfaction Preparedness Ease of use Level of anxiety Time to use Questions\resource needs	Post-MeTree survey MeTree Study Coordinator
Provider experience	Agreement with recommendations Does it change practice Recommend it to peers Quality and usefulness of reports Impact on patient flow Time spent discussing MeTree reports	Provider survey and interviews
Clinic needs	Resource needs to implement Assistance required for patients/providers Training/time required for clinic staff	Study coordinators Interviews

DukeMedicine

DUKE INSTITUTE FOR GENOME SCIENCES & POLICY

Assessing Patient Acceptance

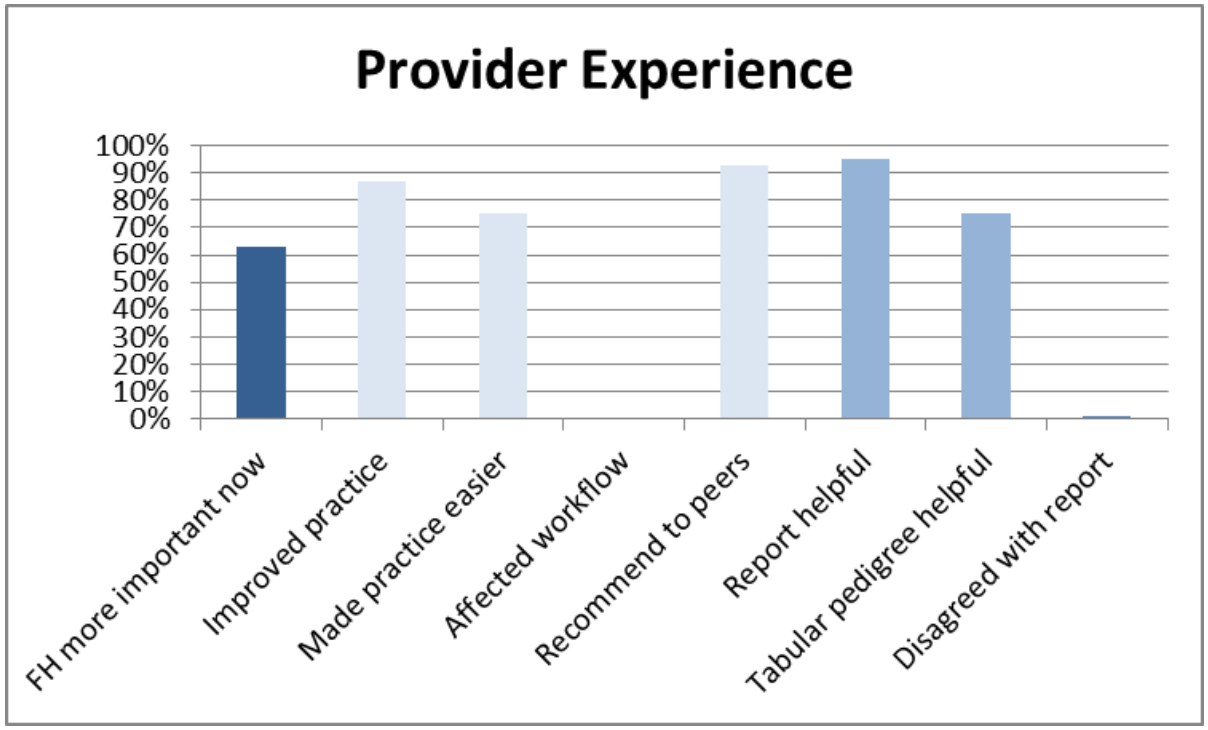




DukeMed

DUKE INSTITUTE FOR GENOME SCIENCES & POLICY

Assessing Provider Acceptance



DukeMedicine

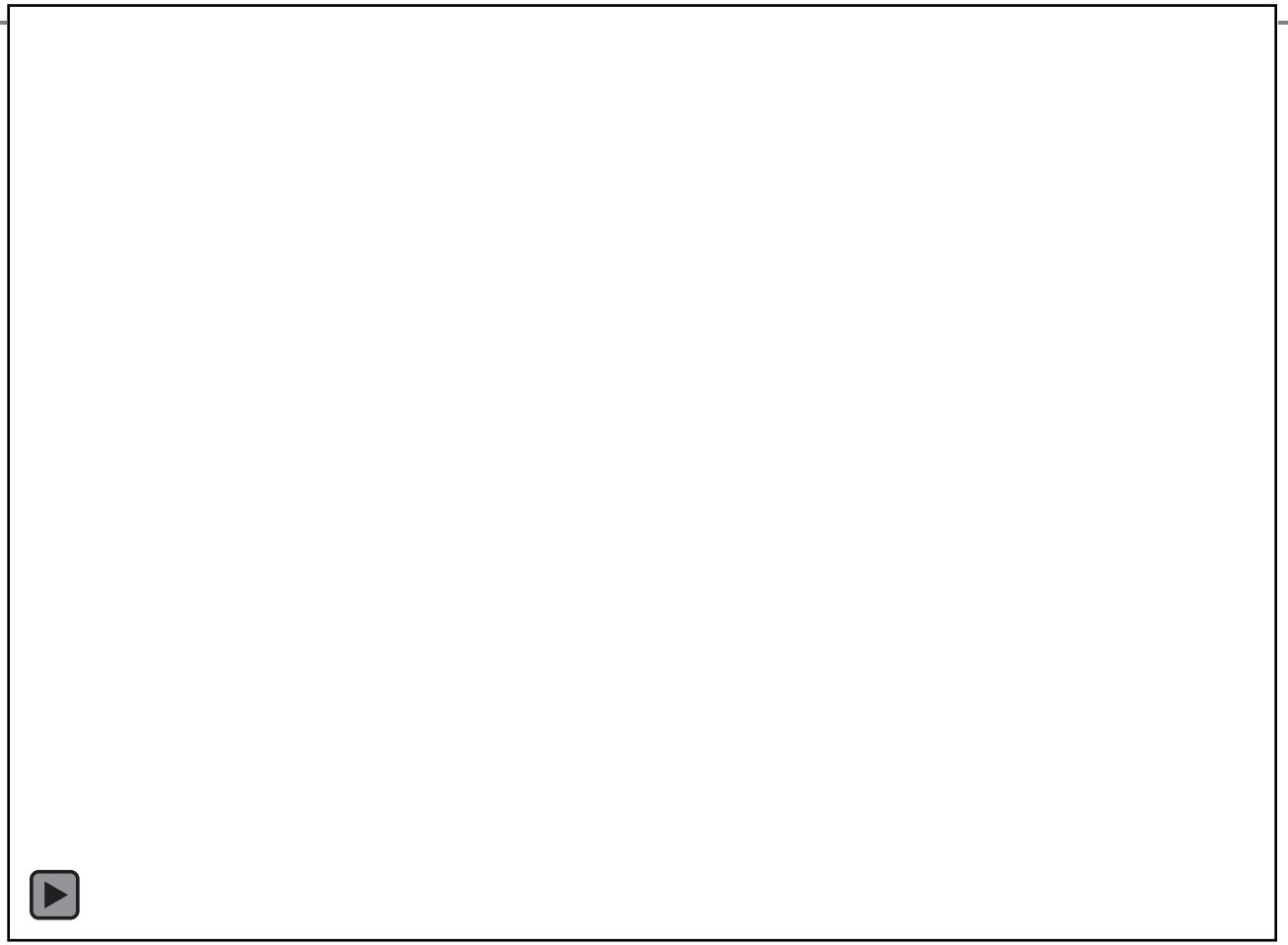
DUKE INSTITUTE FOR GENOME SCIENCES & POLICY

Assessing Clinical Impact

MeTree Provider Recommendation	Male (N=315) Number (%)	Female (N=460) Number (%)	All (N=775) Number (%)
Any genetic counselor	58 (18.4)	161 (35.0)	219 (28.3)
Cancer genetic counselor	57 (18.1)	148 (32.2)	205 (26.4)
Thrombophilia genetic test	7 (2.2)	19 (4.1)	26 (3.3)
Thrombophilia genetic counselor	1 (0.3)	16 (3.5)	17 (2.2)
Breast MRI		7 (1.5)	
Chemoprevention		44 (9.6)	
Gynecologic surveillance		8 (1.7)	
Early CRC screening	30 (9.5)	57 (12.4)	87 (11.2)
Early and more often CRC screening	20 (6.4)	44 (9.6)	64 (8.3)

DukeMedicine

DUKE INSTITUTE FOR GENOME SCIENCES & POLICY



DukeMedicine

DUKE INSTITUTE FOR GENOME SCIENCES & POLICY

Summary

- Family History Demonstration Project
  - 1000 patients enrolled
  - Providers are willing to alter their practice based upon FH decision support
  - FH collection can be a positive experience for patients and providers and can be implemented without disruption to workflow
  - Clinical validity of MeTree vs PCPs benchmarked with independent genetic counselors
  - Clinical utility (in progress)
- Genomic model for clinical practice
  - Built a scalable and transferable (learning) model for delivery of genomic information
  - Established an implementation sciences framework for outcomes research

Moving Forward

- Incorporate additional risk information into platform
- Implement in military clinical practices
- Broaden clinical decision support
- Tablet and iPhone apps to gather family history info
- Text messaging to remind patient to complete MeTree and to follow-up on recommendations (electronic health coaching)
- Expansion to other diseases
- Integration of DNA testing and PGx