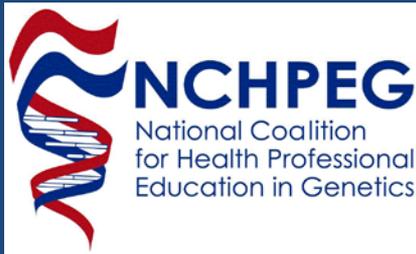


# IMPACT OF WHOLE GENOME SEQUENCING ON THE PRACTICE OF HEALTH CARE



Emily Edelman, MS, CGC  
National Coalition for Health Professional  
Education in Genetics

SACGHS Meeting  
Washington, D.C.

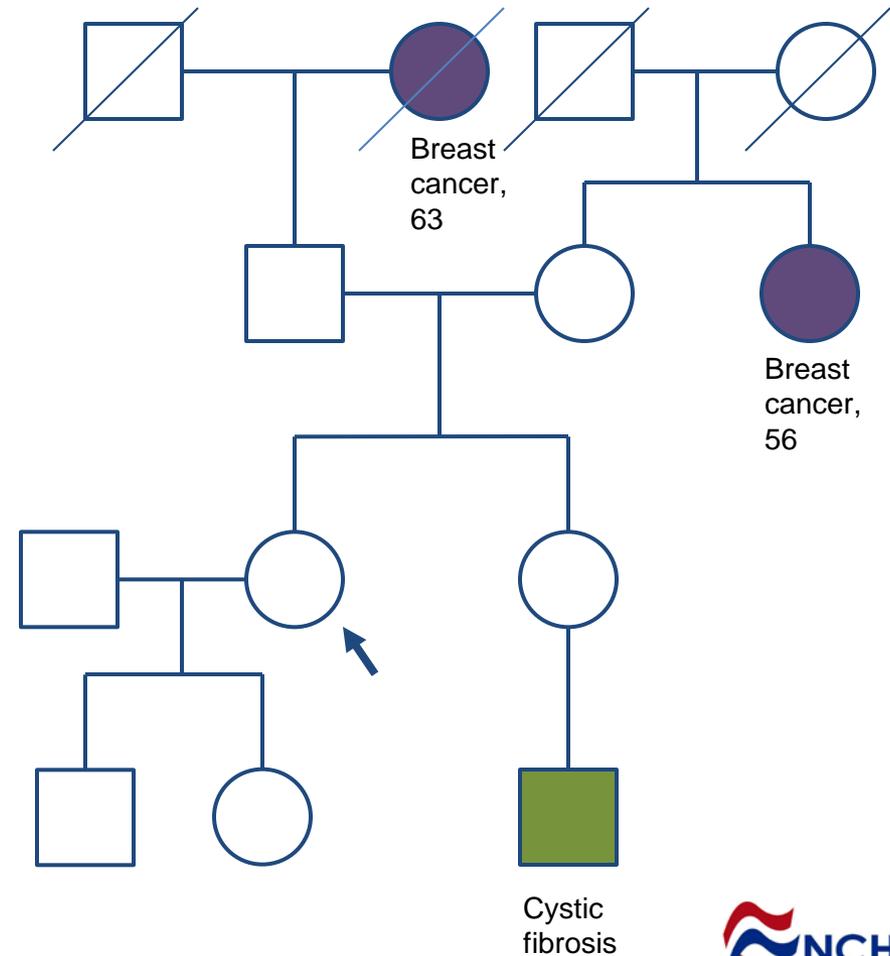
15 June 2010

# Introduction

- How is WGS being applied to clinical practice?
- How will WGS change the practice of medicine?
  - Informed Consent
  - Results Communication
  - Approach to variants of uncertain significant
  - Clinician Education
  - Patient Education

# Primary Care + \$1,000 genome

- Sara, 32 years
- Presents to OBGYN for breast cancer screening and family planning
- OB orders genome



# Primary Care + \$1,000 genome

- Sara and husband return for follow-up
- OB logs into Sara's genome portal or EHR
- Queries "breast cancer" and "prenatal panel"
  - ▣ Breast cancer: heterozygous *CHEK2* and *FGFR2* mutations
  - ▣ Cystic fibrosis: heterozygous mutation
- Clinical decision support

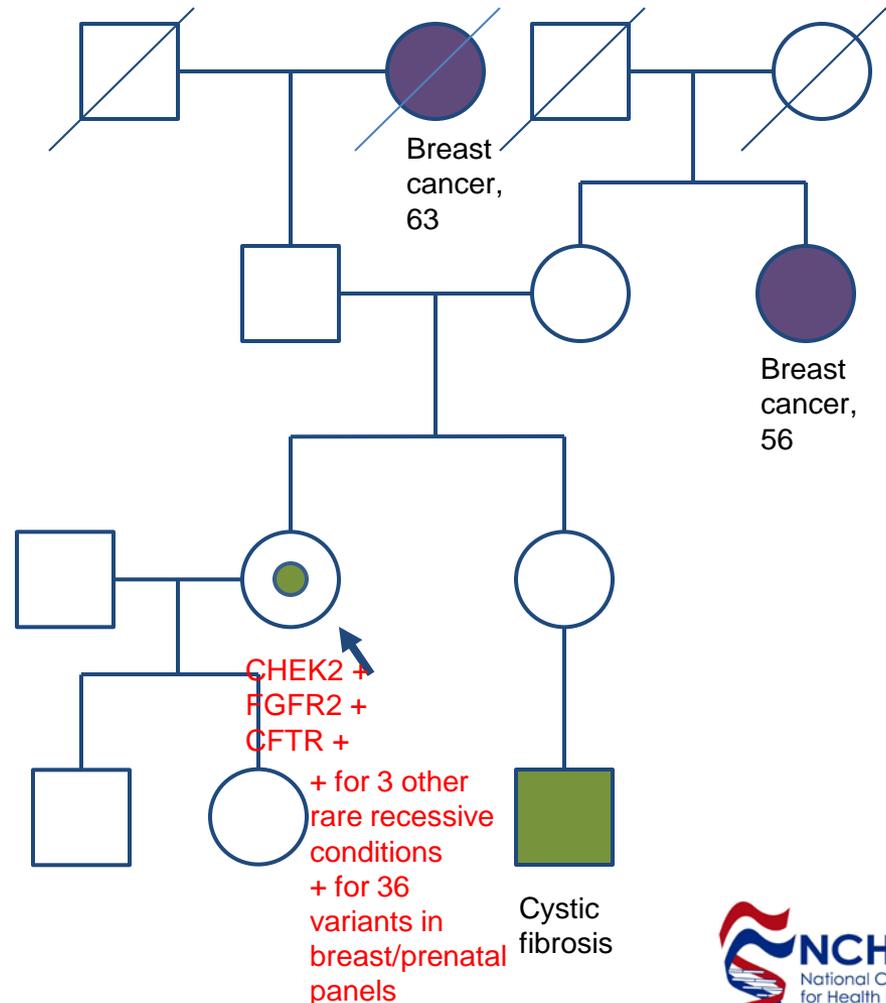


variants in  
breast/prenatal  
panels

Cystic  
fibrosis

# Primary Care + \$1,000 genome

- OB counsels Sara about breast cancer risk
  - ▣ Mammogram at 35
- Discusses uncertainty with variants
- Orders husband's genome
- Refer to genetic counselor
  - ▣ husband's results discussion
  - ▣ CF and other recessive disease counseling



# How to get from here to there?

- ❑ Affordable testing
- ❑ Clinical validity and utility data for specific gene-disease and SNP-disease associations
- ❑ Data management and decision-support systems
- ❑ Payor recognition
- ❑ Recommendations from professional society
- ❑ Clinician education
- ❑ Patient and consumer education

What will be completed first?

# Clinician & Consumer Motivations

- What is the goal of ordering or requesting WGS?
  - What does the clinician or consumer hope to learn?
  - Will WGS results change patient management qualitatively?
- 
1. Risk assessment, diagnosis, or treatment for specific disease, targeted care
  2. Preventative health, primary care
  3. Direct-to-consumer

# Informed Consent & Consumer Education

↑ Complexity of Information

↑ Length of consent form

↓ Patient comprehension  
Likelihood of reading

*IRB: Ethics & Human Research*  
32, no 3 (2010): 7 – 11.

# Informed Consent & Consumer Education

- Depends on clinical model
- Clinician, client motivations
- Supplementary education
  - ▣ Risks, benefits, limitations
  - ▣ Possible results
- Current research WGS consenting: 30 – 60 min, 8 – 9 pgs.
- Current clinical WGS consenting:  $\geq$  45 min appt., 9 pgs.

Biesecker et al. 2009. *Genome Res*  
Personal correspondence with F.  
Facio, J. Sapp, J. O'Daniel

# Is WGS Different from What We Already Do?

1. Knowledge base
2. Ability to manage uncertainty
3. Ability to communicate risks and uncertainty

# Results & Risk Communication: Who?

- Genetics professionals: MDs, counselors, nurses
- MD - PCPs, family medicine, internal medicine
- Nurses, PAs
- Pharmacists
- Health educators
- Lab personnel

# Results & Risk Communication: Who?



Lab support for interpretation of results



Requirement for clinician's level of genetics expertise in the interpretation of results

# Results & Risk Communication: How?

- What information needs to be conveyed?
- In what context?
- How to address variants?

# A Genetic Counselor's Approach

Cleveland Clinic, 2009

1. Pre-test group education opportunity
2. DTC personal-genome screening
3. Pre-appointment family history, results review, and phone contracting
4. Build discussion of genomic results into genetic counseling appointment
  - ▣ Family history risk assessment & recommendations
  - ▣ Share information with managing provider

# A Genetic Counselor's Approach

## Challenges

1. Time
2. DTC testing company
3. Results confirmation
4. Mix of types of results
5. Limited clinical validity and utility
6. Limited IT tools and resources
7. Need for education among other health care providers

# Health Professional Education

- Challenges to education for single-gene highly penetrant disorders
  - ▣ Clear management recommendations
  - ▣ Standard of care
- Low penetrant variants, variants of uncertain significance
  - ▣ Variable validity
  - ▣ Shifting risk estimates

# Closing the Gap

- Genetics professional guidance
  - ▣ Mentors
  - ▣ Consultants
  - ▣ Educators
  - ▣ Lab employees
- Pre-clinical + continuing education
  - ▣ Integrate “genomic thinking” into all aspects of medical education
  - ▣ Teachable moments
  - ▣ Point of care
  - ▣ Clinical decision support
- Lab and 3<sup>rd</sup> party support, Professional Societies

# Conclusions

- Affordable WGS is coming soon to a clinic near you
- It will be affordable before we understand the clinical importance of all of the data
- Genetics professionals will be heavily involved initially, with care spreading to other providers over time
- It will be extremely challenging to educate clinicians and consumers in this time of rapidly changing information
- Risk communication principles are similar to those for traditional genetic medicine, but will be applied in a different context

# Acknowledgements

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Flavia Facio, MS, CGC

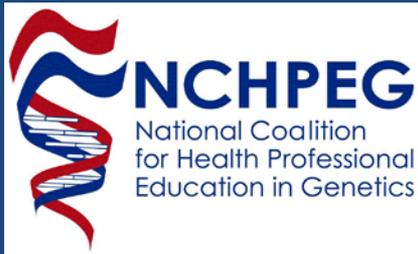
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# THANK YOU!



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