

Genetics and Your Health

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SEPTEMBER 24, 2020

Overview

- Genetics 101
- What is Genetic Testing?
- At Home (Direct to Consumer) Testing
- Clinical Genetic Testing
- Questions and Discussion

Genetics 101





Why pursue genetic testing?

Ancestry

Explanation of
health concerns

Individuals with
developmental
delay or
differences at birth

Family history of
disease

Pregnancy
planning

For fun

What can genetic testing do for me?

- Guide and improve screening or management
- Provide an explanation for unexplained symptoms
- Gauge potential risks to family members
- Non-clinical benefit such as insight into ancestry or traits

What can genetic testing do for my family?

- Determine specific risks for family
- Provide intervention options
- Reassurance

Suzy Q is 30 and diagnosed with breast cancer



Pre-test genetic counseling



Clinical genetic testing



BRCA1 mutation



Impacts surgery and future screening



Relatives at risk informed

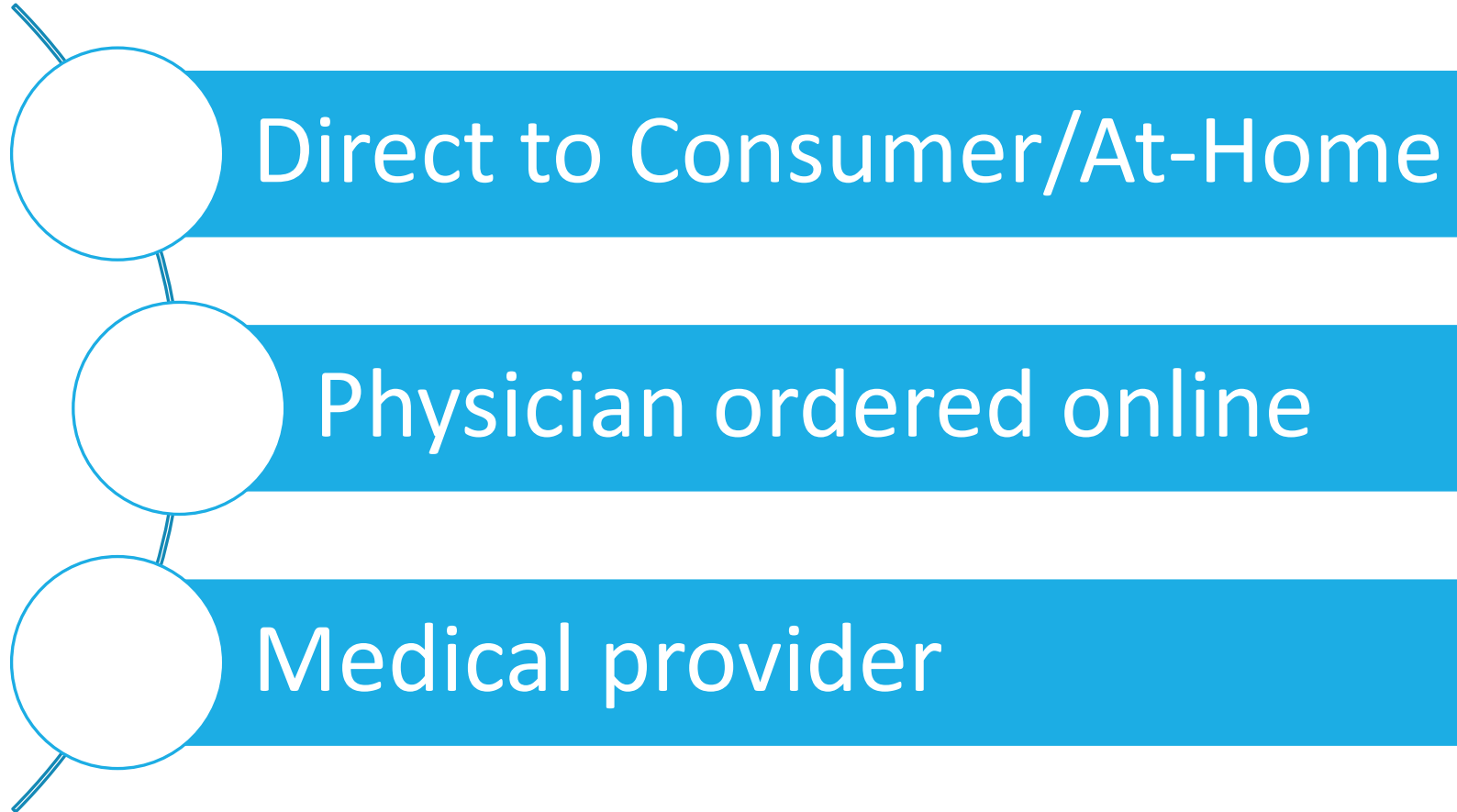


Sister Beth tests negative



Sister Anna tests positive

Ways to pursue genetic testing

- 
- Direct to Consumer/At-Home
 - Physician ordered online
 - Medical provider

Direct to Consumer At-Home Testing

What makes DTC unique?

- Convenient
- A physician is not reviewing and signing off on the test
- It may not be comprehensive
- Varying levels of guidance for what your results mean for you and your family
- Generally should not be used for making medical management decisions

Trait testing

Wellness

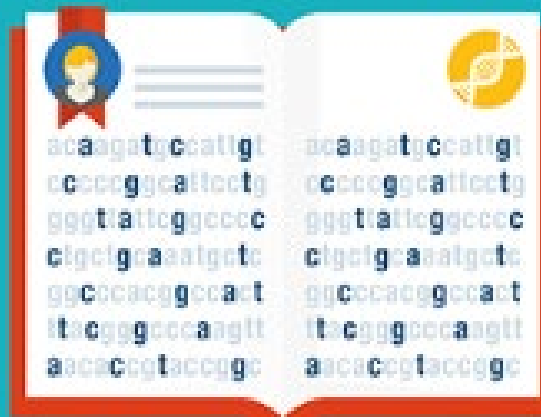
Ancestry

Carrier status

Paternity testing

Technology

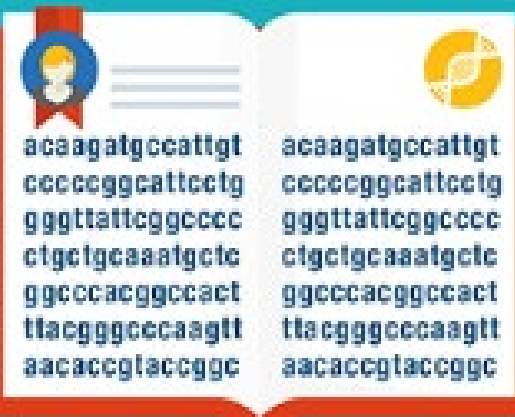
Genotyping



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ccccggcattctg cccccggcattctg
gggttattggcccc gggttattggcccc
ctgctgcaaatgctc ctgctgcaaatgctc
ggcccacggccact ggcccacggccact
ttcgggccaagtt ttcgggccaagtt
aacaccgtaccggc aacaccgtaccggc

OR

Sequencing



acaagatgccattgt acaagatgccattgt
ccccggcattctg cccccggcattctg
gggttattggcccc gggttattggcccc
ctgctgcaaatgctc ctgctgcaaatgctc
ggcccacggccact ggcccacggccact
ttcgggccaagtt ttcgggccaagtt
aacaccgtaccggc aacaccgtaccggc



Ancestry
Composition



Family Tree



DNA Relative Finder



Traits reports



Haplogroups



Neanderthal
Ancestry

Reports include

- Hair: Color, Curliness, Male Bald Spot
- Taste & Smell: Sweet vs. Salty, Bitter
- Facial Features: Cheek Dimples, Unibrow, Freckles

Heart and Blood Health

Cardiomyopathy

Familial Hypercholesterolemia

Hereditary Hemochromatosis

Hereditary Thrombophilia

BRCA1/BRCA2 (Selected Variants)

[Learn more](#)

Genetic risk based on a limited set of variants for breast, ovarian and other cancers

3 variants in the BRCA1 and BRCA2 genes; relevant for Ashkenazi Jewish descent

Raw Data – What is it?

- Downloading “raw data” report and uploading into a third party website
 - Not recommended! - Typically not accurate
 - Could tell you information you weren't looking for
 - May lead to false positives
 - Data may not come from reliable sources

Considerations when ordering at-home DNA tests

- What are your goals?
- Is the company trustworthy?
- What will you find out?
- What is the follow up for your results?
- Will your information be protected?
- What are possible unanticipated results?

What are your goals?

- Fun
- Interested in ancestry or your lineage
- Health

Is the company trustworthy?

- Is lab reliable and certified?
 - Clinical Laboratory Improvement Amendments (CLIA)
 - College of American Pathologists (CAP)
- Are employees and board members experts?
 - Certified genetic counselors, pathologists, geneticists, biologists, or molecular pathologists

What will you find out and how?

- Health conditions, carrier status, ancestry, traits, or a combination?
- How will you receive the results?
- What will you do with your results?
- What will you do with unexpected information?
- Will you share these results with your family?
- Will the lab contact you with clinically significant health updates that could impact you?

Will your information be protected?

- What will the company do with your information now and in the future?
- Do they share or sell your information to database, researchers, or other groups?
- Will they update you if their policies change or if they share your information?

What follow up will they provide?

- Who can you go to with questions?
- Is a genetic counselor or other trained professional available to answer your questions?
- Is this service included in your original cost or is there an extra charge?
- If not, who can you go to review results?

What are possible unanticipated results?

- Genetic risk you were not informed of pre-test
- Non-paternity
- Biological relatives you weren't aware of

Genetic Counseling

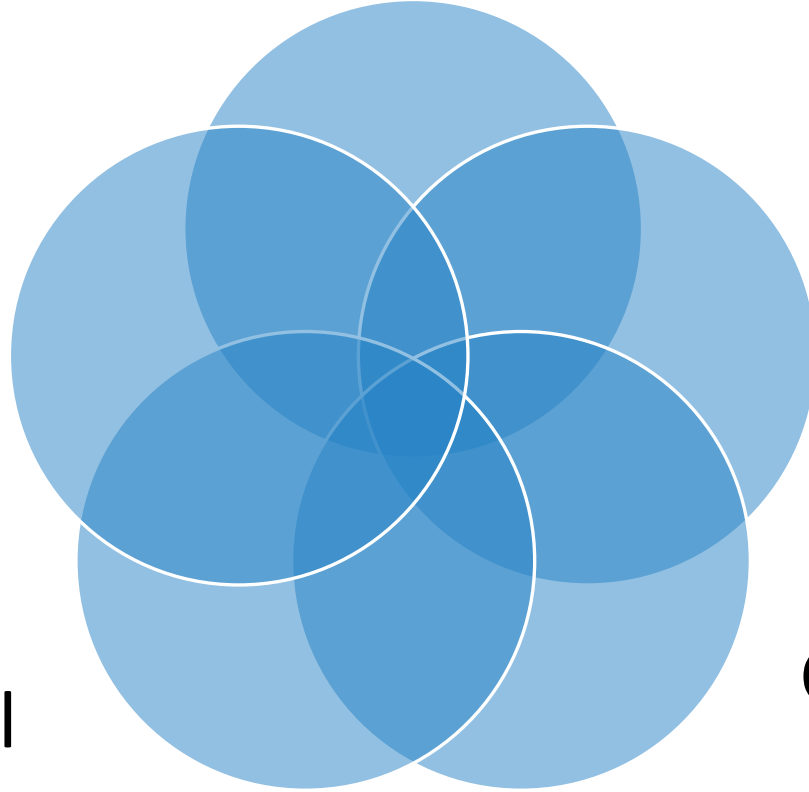
Personal and
Family History

Results
Interpretation

Disease Risk

Psychosocial
Counseling

Coordination
Genetic
Testing





Benefits of Genetic Counseling

- Personalized risk assessment
- Psychological support
- Nondirective guidance
- Up-to-date knowledge of clinical genetic testing
- Interpretation of test results
- Referrals to relevant providers
- Explaining implications of your results to family members
- A point person for all of your genetics questions

Insurance and clinical genetic testing

- When clinically indicated, most insurance companies approve genetic counseling and testing
- Subject co-pays and deductibles
- Talk to a genetic counselor to get personalized insurance information

GINA: the Genetic Information Non-Discrimination Act

- Protects health insurance
 - Deny coverage
 - Adjustment of premiums
 - Otherwise discriminate the basis of genetic information
- Employment
 - Hiring, firing, compensation, or promotion decisions

Exceptions to GINA

- Long-term Care Insurance
- Disability Insurance
- Life Insurance
- Military Service
- Small companies of 15 people or less

Summary

- Genetic information can inform personalized care for you and your family
- Purchase or gift at-home genetic testing as an informed consumer
- Talk to your doctor if you are interested in genetic testing or meeting with a genetics professional
- Go with your gut, if it sounds too good to be true, it probably is

Where can I get more information?

Genetics Home Reference

<http://ghr.nlm.nih.gov>

National Society of Genetic Counselors

www.nsgc.org

www.aboutgeneticcounselors.com

Genetic testing resources:

<http://www.aboutgeneticcounselors.com/Genetic-Testing/Is-Direct-to-Consumer-Genetic-Testing-Right-for-You>

Thank you!

Questions?

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How do I schedule an appointment with a genetic counselor?

- Talk to your doctor to see if they would recommend a referral to medical genetics
- Find a genetic counselor close to you at nsgc.org