Genetic Counseling Graduate Program

Arizona Telemedicine Program

Southwest Telehealth Resource Center TRC

Improving Access to Quality Medical Care Webinar Series

Presented by

The UArizona Health Sciences Genetic Counseling Graduate Program, the Southwest Telehealth Resource Center, & Arizona Telemedicine Program

Welcome

- SWTRC region - AZ, UT, CO, NM & NV
- Fellow HRSA Grantees
- All other participants from the US & abroad

The **University of Arizona Health Sciences Genetic Counseling Graduate Program, the Arizona Telemedicine Program,** and **Southwest Telehealth Resource Center** welcome you to this free webinar series!

The series will discuss the the use of telegenetics to improve access to genetics services for patients & providers.

Telemedicine can help you achieve these goals!

Webinar Tips & Notes

- When you joined the webinar your phone &/or computer microphone was muted
- Time is reserved at the end for Q&A
- Please use the **Chat function** to ask questions
- Please complete the post-webinar survey
- Webinar is being recorded
- Recordings will be posted on the ATP website
    - http://telemedicine.arizona.edu/webinars/previous

Incorporating Genetics in the Clinic

Valerie Schaibley, PhD

Associate Director, University of Arizona Genetic Counseling Graduate Program
Administrator, Center for Applied Genetics and Genomic Medicine
Assistant Professor, Educator Scholar Track, Department of Cellular and Molecular Medicine

Incorporating Genetics in the Clinic

Valerie Schaibley, PhD

February 13, 2020

Arizona Telemedicine Program Webinar Series on Telegenetics

Conflict of Interest

- None to disclose.

Check out our past webinars!

- Defining and Discussing Telegenetics, Chris Stallman, MS, LCGC
  - https://swtrc.wistia.com/medias/8vzwgjr2on

- Applications of Telegenetic Counseling in Research and the Clinic, Valerie Schaibley, PhD
  - https://swtrc.wistia.com/medias/6vbu3t9tpq

Learning Objectives

1. Apply common indications for genetic testing.
2. Describe the growing trend of incorporating genetics in clinical care, especially using telemedicine.
3. Access resources for genetic conditions for providers and patients.

Webinar Outline

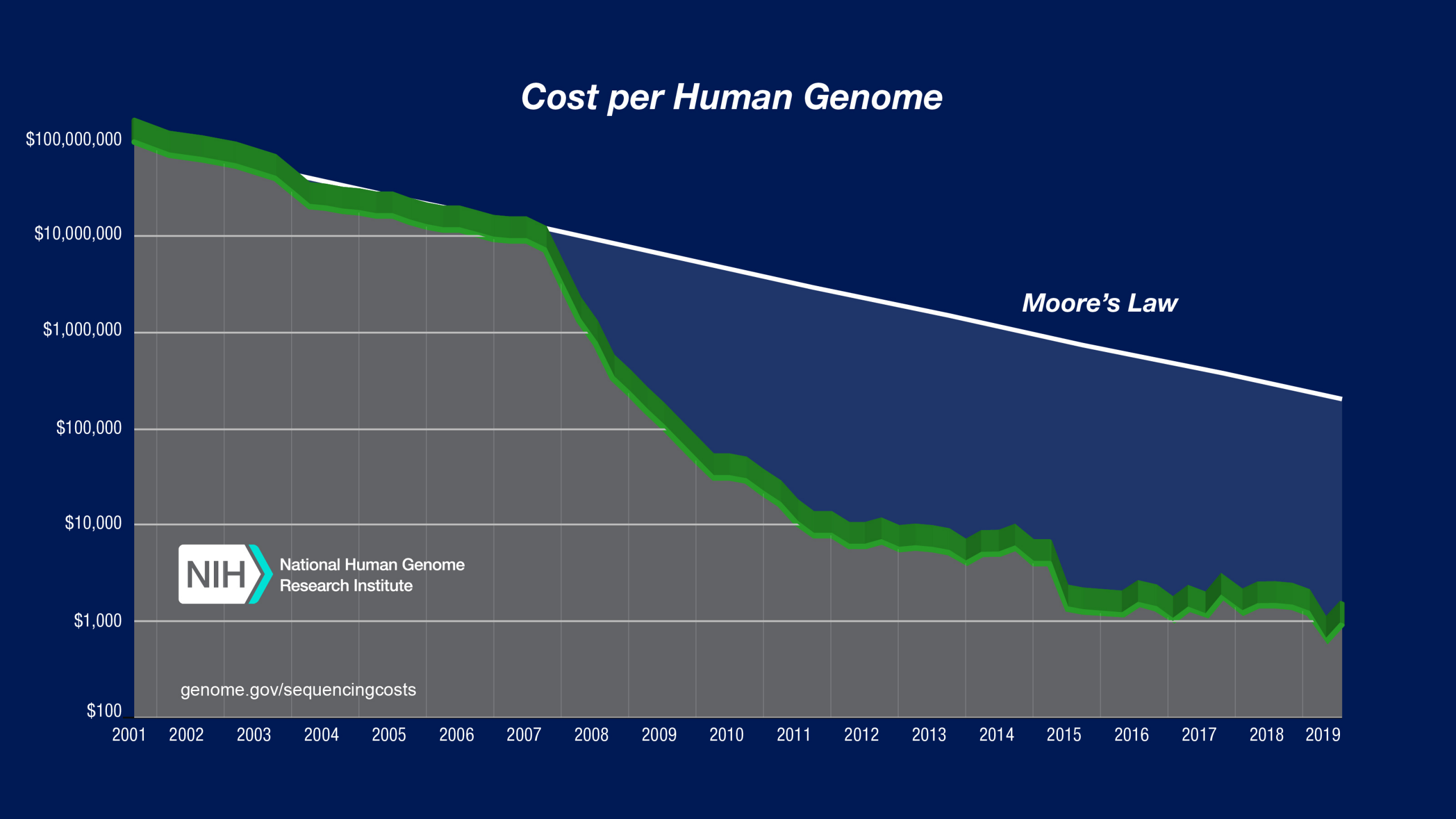
- Incorporating genetics into the clinic – where did we come from and where are we now
- Examples from common genetic traits
  - Hereditary breast and ovarian cancer
  - Colorectal cancer
- How can genetic counselors help access genetics services?
- Resources for providers and patients

Incorporating genetics into the clinic – where did we come from and where are we now?

The Human Genome Project

- International collaborative effort to sequence and map all of the genes in humans, collectively called *the genome*
- Created the first reference sequence of the human genome
- Project started in 1990 and completed in 2003
- Finished early and under budget
- Developed significant new technologies and methods to generate and analyze human DNA sequence
- At the end of the Human Genome Project, the cost to sequence a human genome was about $50 million

https://www.genome.gov/about-genomics/fact-sheets/Sequencing-Human-Genome-cost


Cost per Human Genome
$100,000,000
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Moore's Law
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National Human Genome
Research Institute
genome.gov/sequencingcosts
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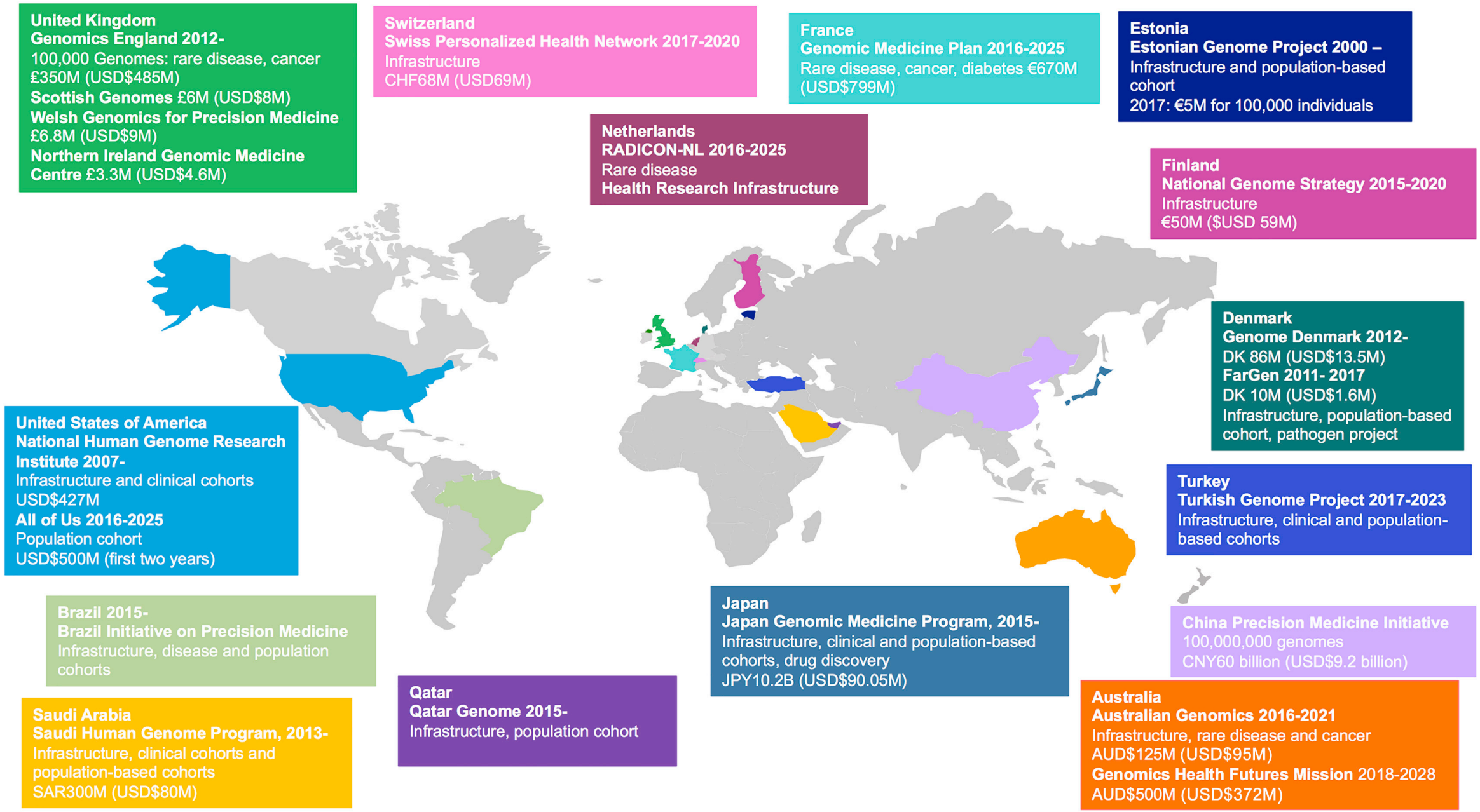
Integration of Genetics into Clinical Care

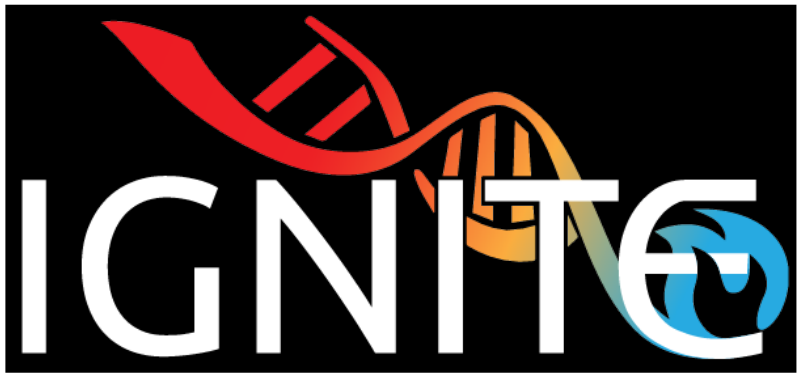
- Diagnostics for pediatric genetics
- Newborn screening
- Prenatal genetic testing and screening
- Screening for carriers of genetic conditions
- Identifying high-risk genetic variants that significantly increase risk of disease
- Tumor profiling to identify most effective treatments
- Pharmacogenomics
- Polygenic risk scores
- Whole exome or whole genome sequencing for unknown disease diagnosis

What are some of the considerations with integrating genetics into clinical care?

- Cost effectiveness
- Education of patients and healthcare providers
- Insurance reimbursement
- Limited evidence and/or recommendations for some conditions
- Limited access to genetics services, especially in underrepresented populations
- Lack of EMR integration of genetic testing results



Stark et al., 2019; PMID: 30609404



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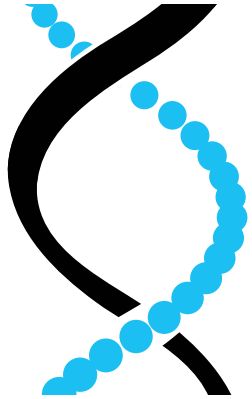

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Clinical Sequencing Evidence-Generating Research

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ELECTRONIC MEDICAL RECORDS & GENOMICS

All of Us

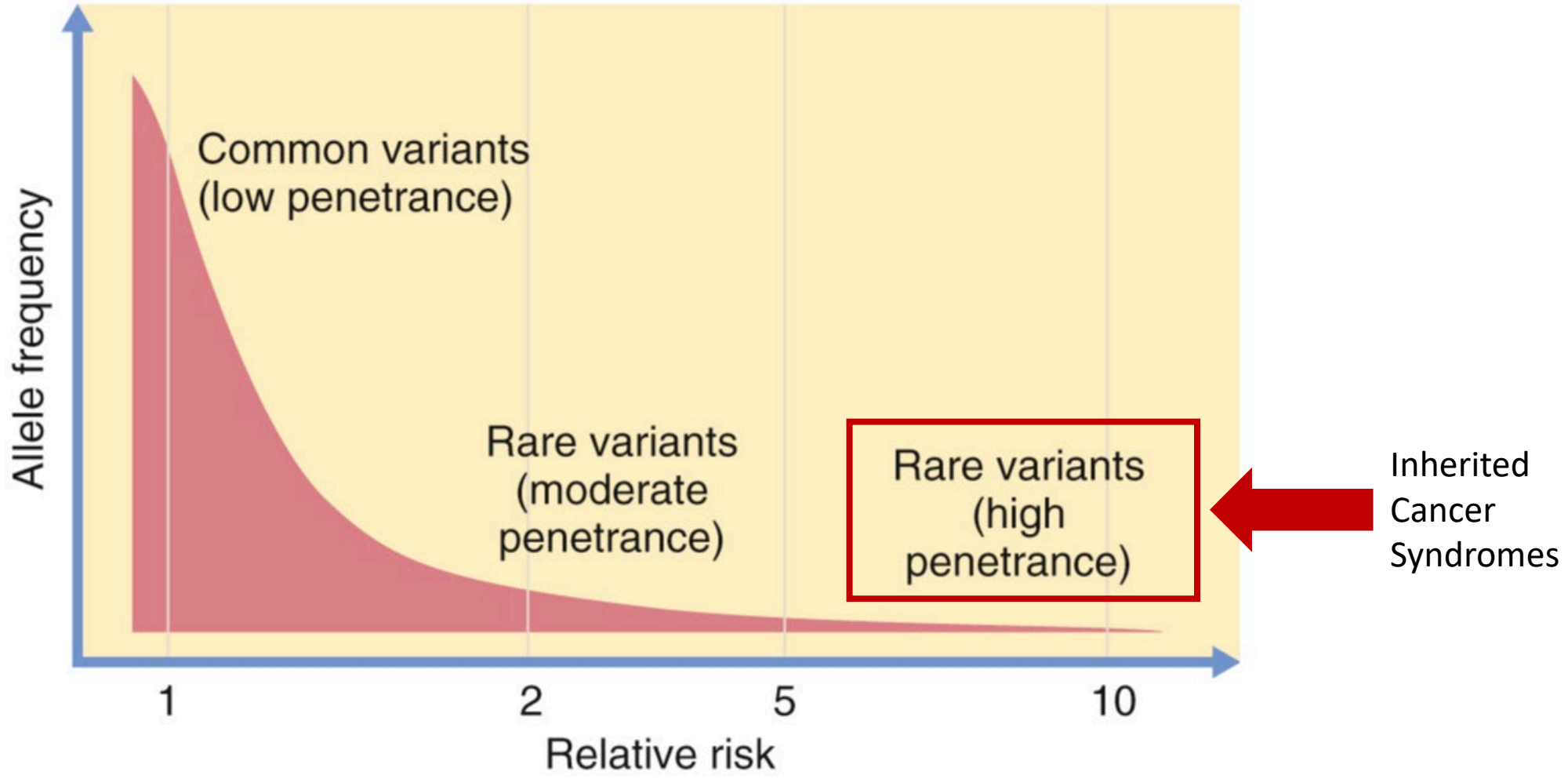
RESEARCH PROGRAM

SANFORD imagenetics

Webinar Outline

- Incorporating genetics into the clinic – where did we come from and where are we now
- Examples from common genetic traits
  - Hereditary breast and ovarian cancer
  - Colorectal cancer
- How can genetic counselors help access genetics services?
- Resources for providers and patients

Rare and Common Genetic Variants Affect Cancer Risk



Inherited Cancer Syndromes

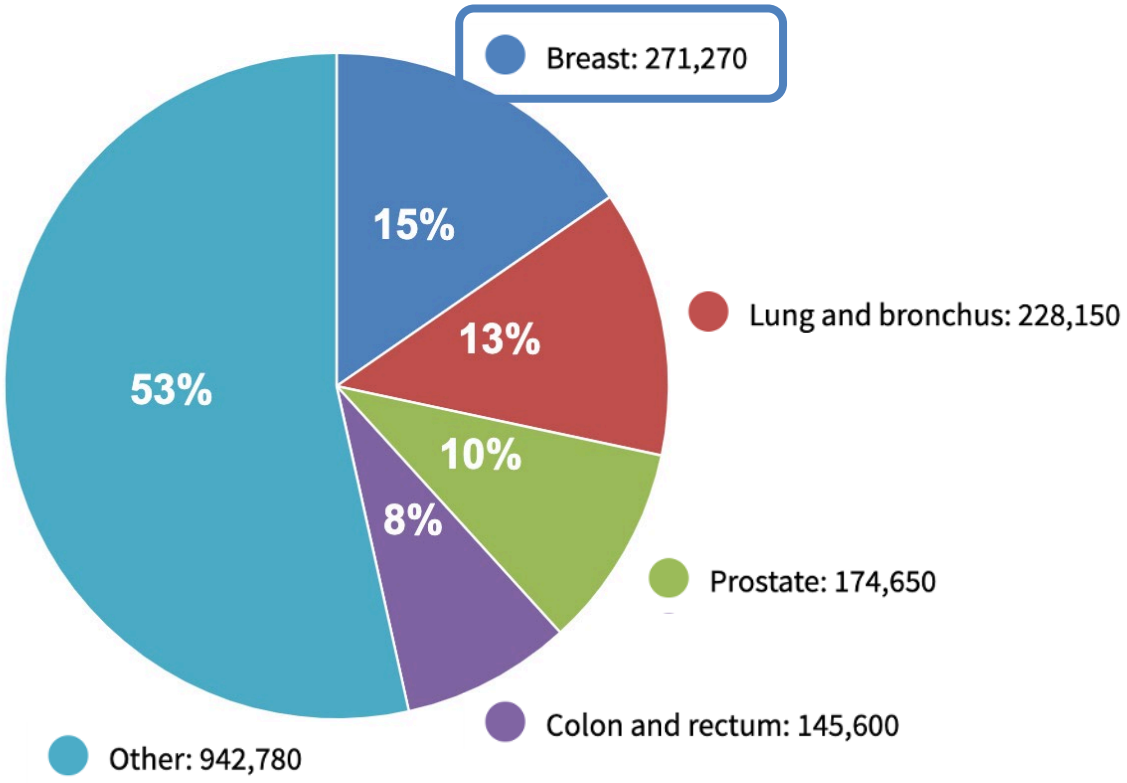
- About 5-10% of cancers are due to an inherited predisposition
- Inherited predispositions to cancer are due to germline genetic variants in a cancer predisposition gene
- Inheriting a germline variant in a cancer predisposition gene ***does not* mean that a person will develop cancer**
- Rather, it **increases the risk** of cancer developing in an individual
  - Environment and lifestyle can still play a role in overall risk
- Inherited cancer syndromes predispose to specific types of cancer

Features of Inherited Cancer Syndromes

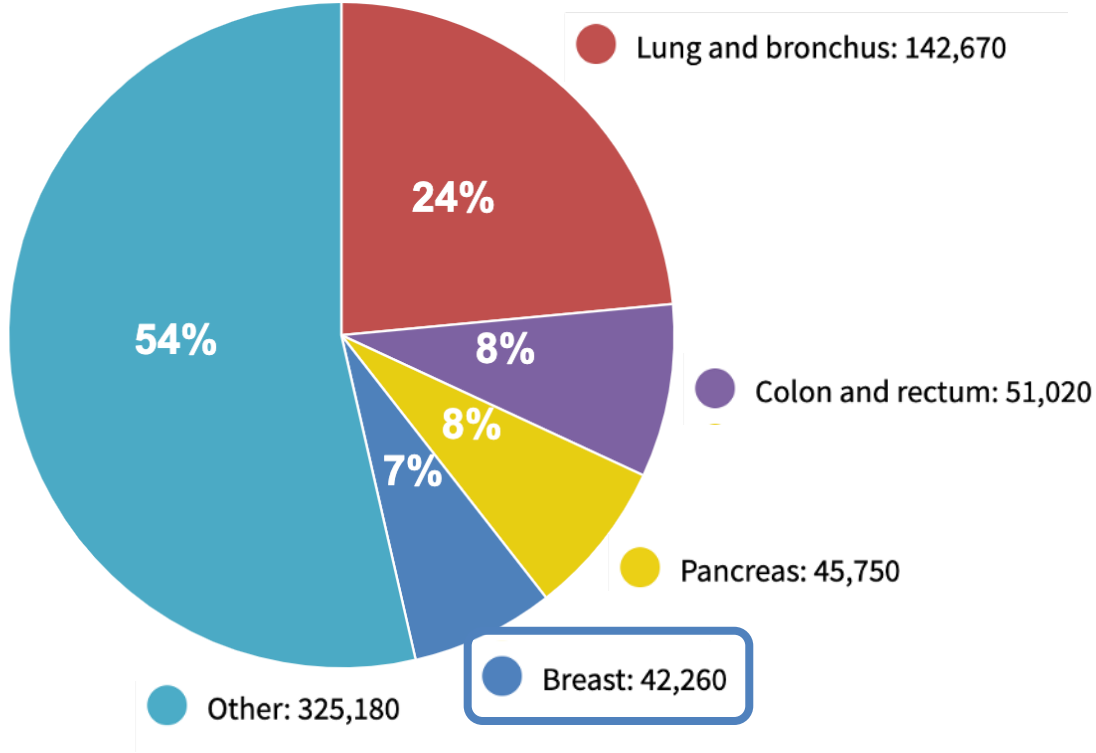
1. Several relatives with the same or related cancers
2. Younger age of onset than is typical
3. Autosomal dominant pattern of inheritance
4. Presence of rare cancers
5. Excess of multifocal or bilateral cancers
6. Excess of multiple primary cancers
7. Presence of other nonmalignant features
8. Absence of environmental risk factors

Breast cancer is the most common type of cancer and 4th most deadly type of cancer in the US.

New Cancer Cases, 2019



Cancer Deaths, 2019



SEER Cancer Statistics

Risk Factors for Breast Cancer

Modifiable

- Alcohol use
- Weight
- Physical Activity
- Radiation (Ionizing)
- Occupational Exposures
- Environment

Non-Modifiable

- Sex
  - Women have a higher risk of breast cancer
- Age
- Ethnicity/Race
- Hormones (age at menarche/menopause, parity, breastfeeding)
- Certain breast conditions (LCIS, atypical hyperplasia)
- Family history

Hereditary Breast and Ovarian Cancer (HBOC)

- Inherited cancer syndrome caused by pathogenic genetic variants in DNA damage repair genes:
  - *BRCA1*
  - *BRCA2*
- Inheritance Pattern: autosomal dominant
- In the general population, about 1/400 – 1/800 people carry a variant in *BRCA1* or *BRCA2*
- Estimated 5-10% of breast and ovarian cancers are due to variants in *BRCA1* or *BRCA2*

https://www.cancer.gov/types/breast/hp/breast-ovarian-genetics-pdq

Cancer Risks Associated with *BRCA1/2* Variants

Cancer Type	General Population Risk	*BRCA1* Risk	*BRCA2* Risk
Breast	12%	46-87%	38-84%
Second primary breast	2% within 5 years	21.1% within 10 years	10.8% within 10 years
Ovarian	1-2%	39-63%	16.5-27%
Male breast	0.1%	1.2%	Up to 8.9%
Prostate	6% through age 69	8.6% by age 65	15% by age 65
Pancreatic	1.6%	1-3%	2-7%
Melanoma	2.3%	-	Elevated

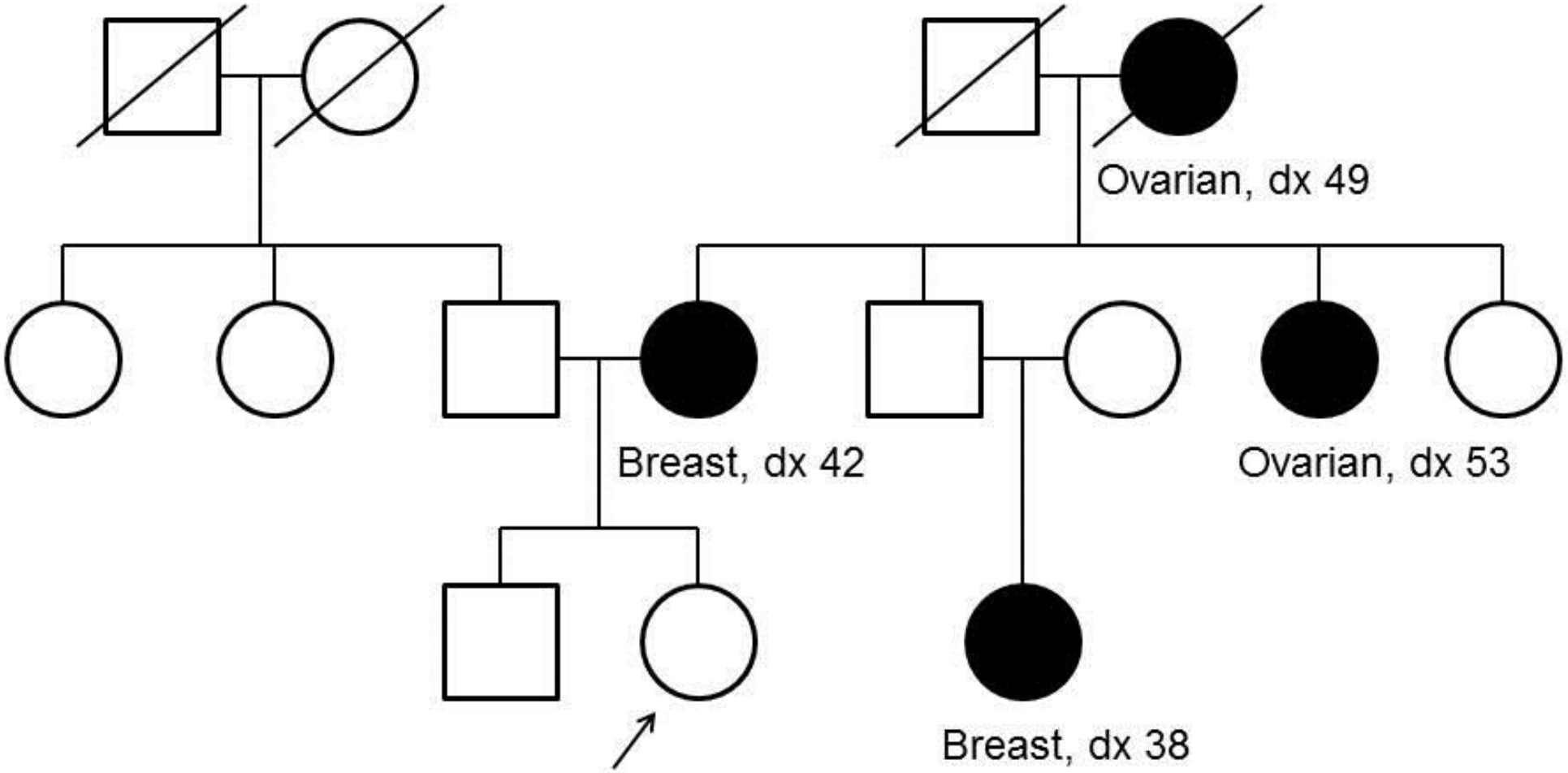
https://www.ncbi.nlm.nih.gov/books/NBK1247/

Family History Suggestive of HBOC

- Breast cancer diagnosed at or before age 50
- Ovarian cancer
- Multiple primary breast cancers in one individual, either unilateral or bilateral
- Triple-negative breast cancer
- Pancreatic and/or prostate cancer with ovarian and/or breast cancer
- Ashkenazi Jewish ancestry
- ≥2 relatives with breast cancer, with one under age 50
- ≥3 relatives with breast cancer at any age
- Previously identified genetic variant in *BRCA1* or *BRCA2*

https://www.ncbi.nlm.nih.gov/books/NBK1247/

Family with a *BRCA1* Variant



https://www.cancer.gov/types/breast/hp/breast-ovarian-genetics-pdq

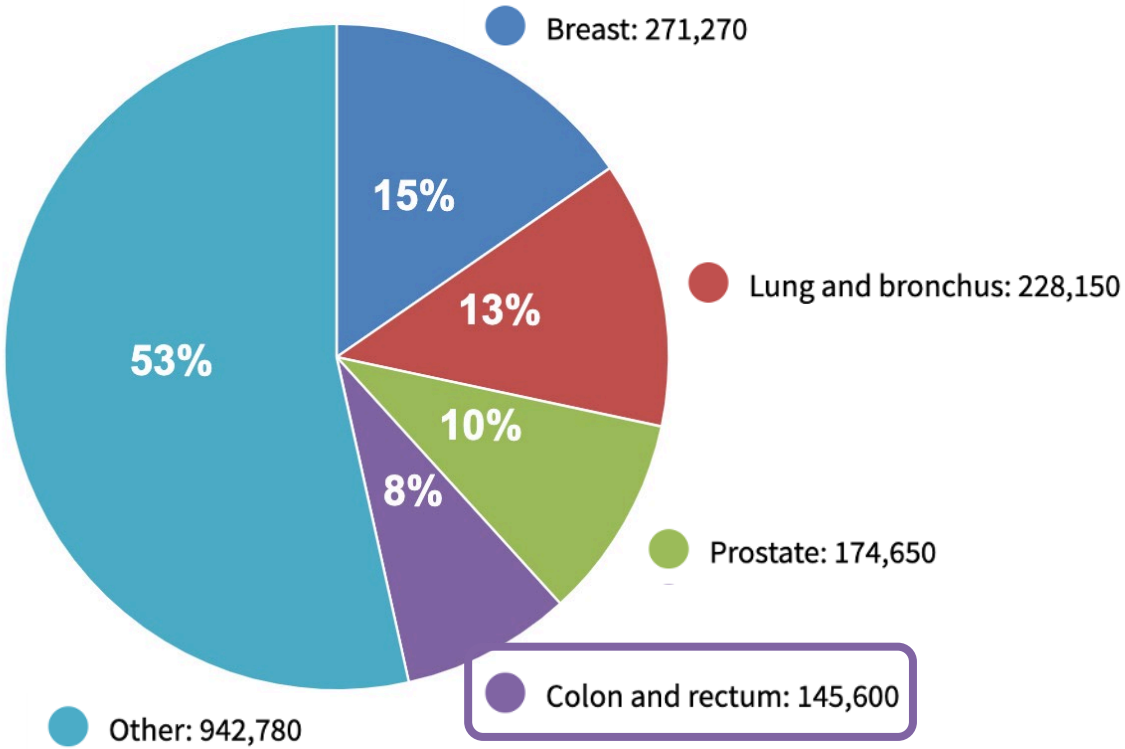
Management of Individuals with *BRCA1/2* Variants

- Female breast cancer
  - Annual breast MRI for women ages 25 – 29 years
  - Annual mammogram for women > age 30 years, consider tomosynthesis and breast MRI
  - Consider risk-reducing mastectomy
- Ovarian cancer
  - Consider transvaginal ultrasound and CA-125 screening starting at age 30 – 35 years
  - Consider risk-reducing salpingo-oophorectomy after childbearing is complete

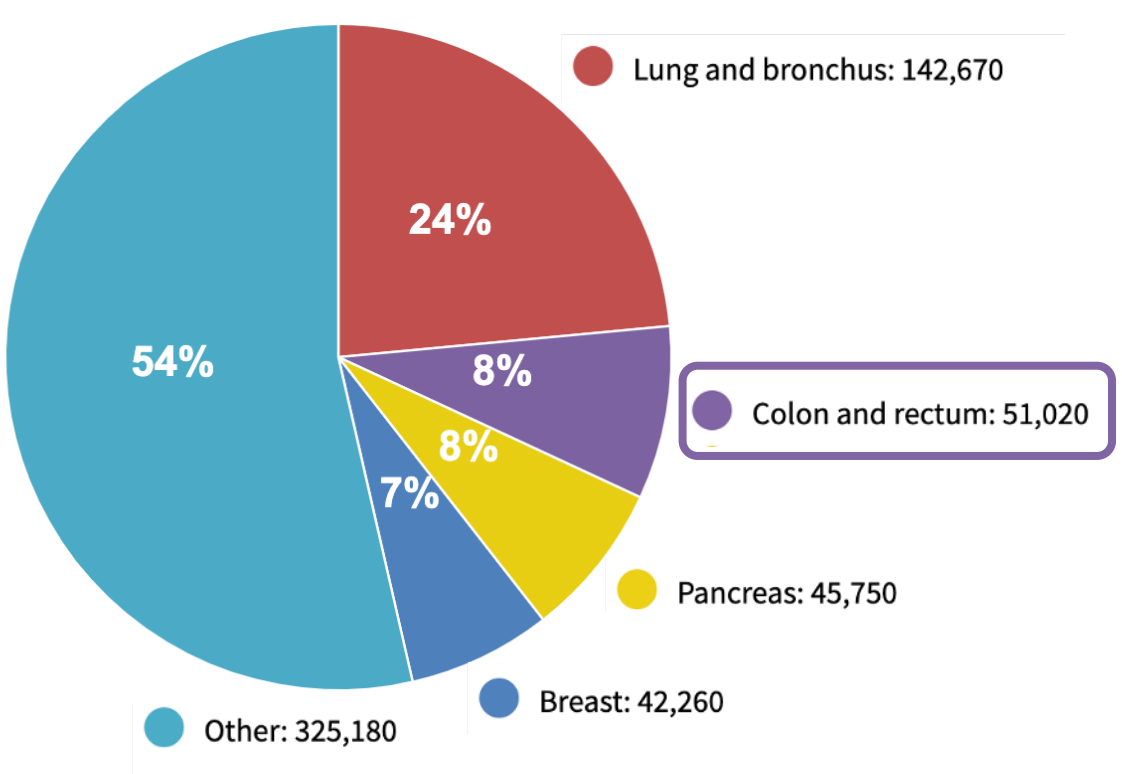
NCCN Guidelines – Genetic/Familial High-Risk Assessment: Breast, Ovarian and Pancreatic, Version 1.2020, 12.4.19

Colorectal cancer is the 4th most common cancer and 2nd most deadly cancer in the US.

New Cancer Cases, 2019



Cancer Deaths, 2019



SEER Cancer Statistics

Risk Factors for Colorectal Cancer

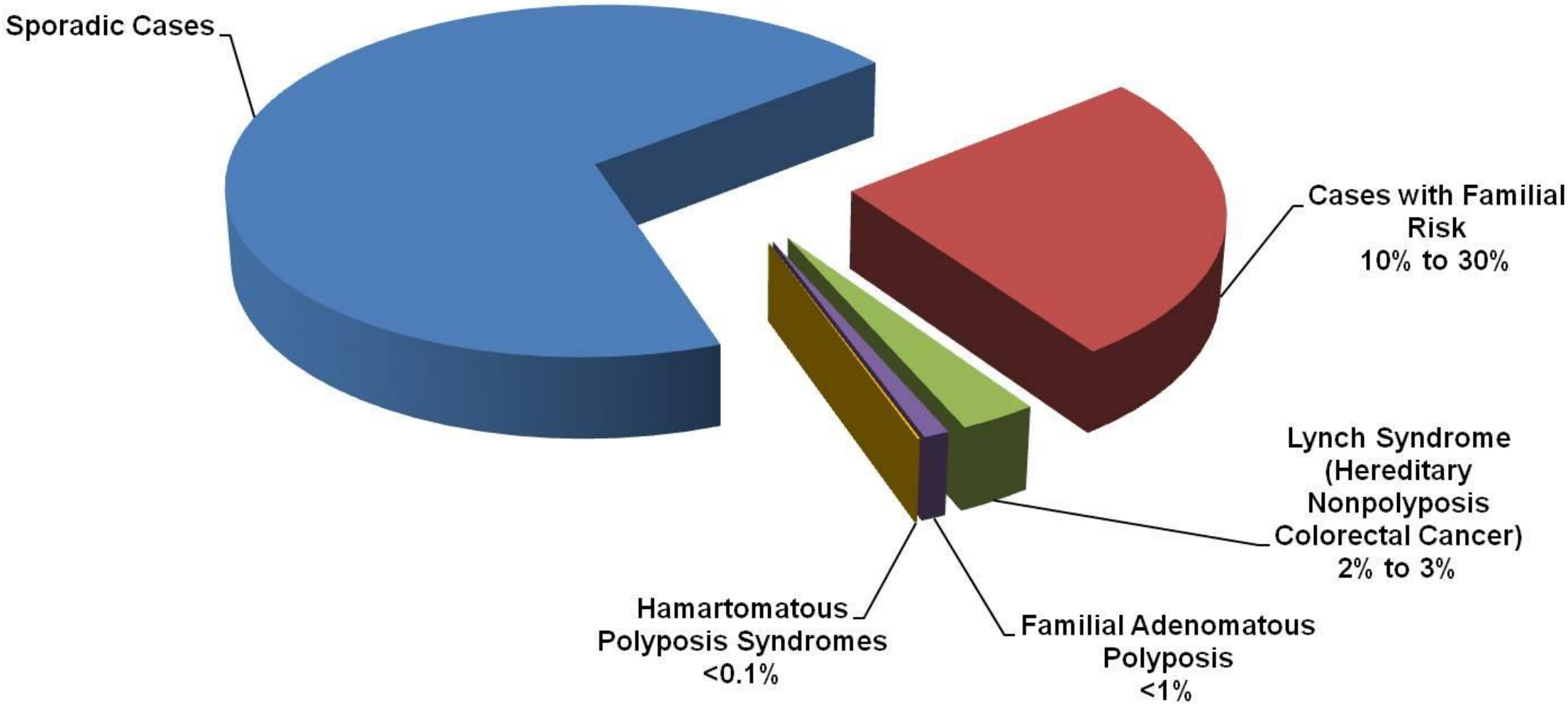
Modifiable

- Overweight or obese, especially in men
- Physical inactivity
- Diet
  - Diets high in red meat, processed meat and fat increase risk
  - Diets high in fruit, vegetables and fiber decrease risk
- Tobacco use
- Alcohol use
  - Limit to 2 drinks/day for men and 1 drink/day for women

Nonmodifiable

- Age
- Personal history of polyps or CRC
- Inflammatory bowel disease
- Diabetes
- Family history of polyps or CRC
- Race/ethnicity
  - African Americans have a higher incidence of CRC
- Sex
  - Men have a higher incidence of CRC

Colon Cancer Cases Arising in Various Family Risk Settings



https://www.cancer.gov/types/colorectal/hp/colorectal-genetics-pdq

Lynch Syndrome

- Inherited cancer syndrome caused by pathogenic genetic variants in DNA mismatch repair genes:
  - *MLH1*
  - *MSH2*
  - *MSH6*
  - *PMS2*
  - *EPCAM*
- Accounts for about 3% of all colorectal cancer diagnoses
- Inheritance pattern: autosomal dominant
- Also known as hereditary nonpolyposis colorectal cancer (HNPCC)

Cancer Risks Associated with Lynch Syndrome

Cancer Type	General Population Risk	*MLH1* and *MSH2*
Colorectal	5.5%	Male: 27-74% Female: 22-53%
Endometrial	2.7%	14-54%
Gastric	<1%	0.2-13%
Ovarian	1.6%	4-20%
Small bowel	<1%	4-12%
Hepatobiliary tract	<1%	0.2-4%
Urinary tract	<1%	0.2-25%
Brain	<1%	1-4%
Sebaceous neoplasms	<1%	1-9%
Pancreas	1.5%	0.4-4%
Prostate	16.2%	9-30%
Breast	12.4%	5-18%

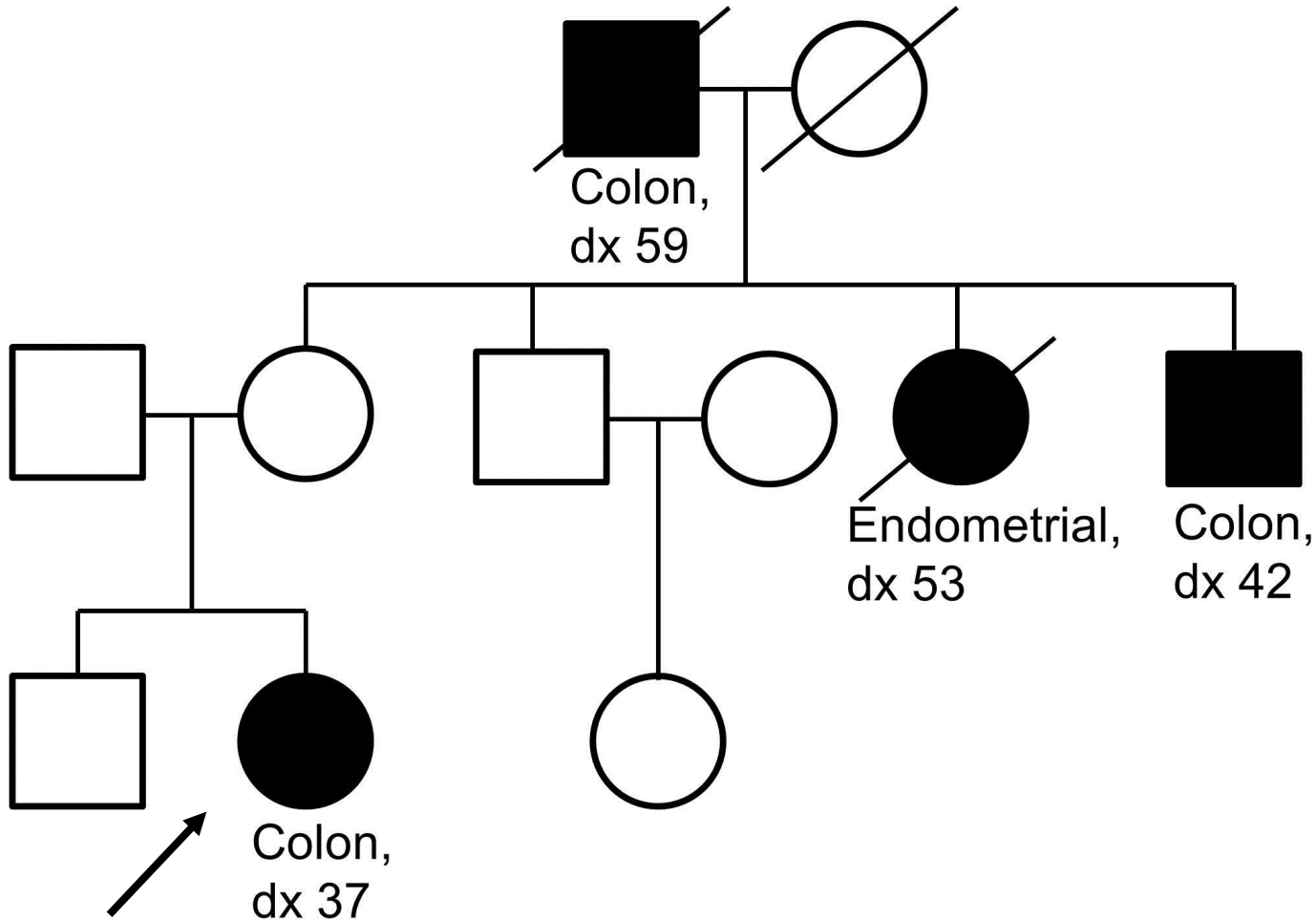
https://www.ncbi.nlm.nih.gov/books/NBK1211/

Family History Suggestive of Lynch Syndrome

- Personal history of colorectal or endometrial cancer plus,
  - Colorectal or endometrial cancer diagnosed before age 50
  - Synchronous or metachronous Lynch syndrome-related cancer
  - High MSI-histology in tumor biopsy
  - Loss of expression of a mismatch repair gene
  - One or more 1st degree relative with a Lynch-related cancer diagnosed before age 50
  - Two or more 1st degree relatives with a Lynch-related cancer
- Family member who meets the above criteria
- Family member with diagnosis of Lynch syndrome

https://www.ncbi.nlm.nih.gov/books/NBK1211/

Family with Lynch Syndrome



Management Guidelines for Lynch Syndrome Patients

- Colorectal cancer
  - Colonoscopy every 1-2 years, starting at age 20 – 25 years
- Endometrial cancer
  - Consider risk-reducing hysterectomy after child-bearing is complete
- Ovarian cancer
  - Consider risk-reducing bilateral salpingo-oophorectomy after child-bearing is complete
  - Consider transvaginal ultrasound and CA-125 screening
- Consider
  - Upper endoscopy every 3-5 years, starting at age 40 years
  - Annual urinalysis starting at age 30 – 35 years
  - Annual neurological exam starting at age 25 – 30 years

NCCN Guidelines – Genetic/Familial High-Risk Assessment: Colorectal, Version 3.2019, 12.13.19

Webinar Outline

- Incorporating genetics into the clinic – where did we come from and where are we now
- Examples from common genetic traits
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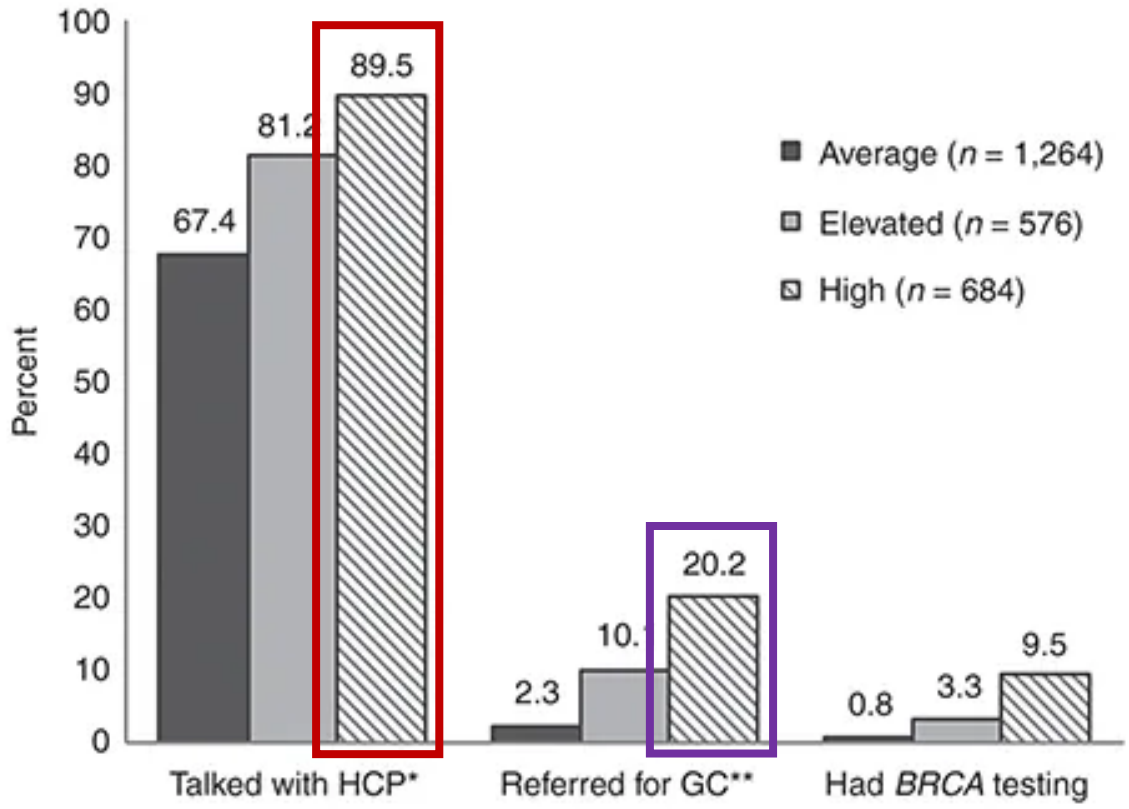
  - Colorectal cancer
- How can genetic counselors help access genetics services?
- Resources for providers and patients

How can genetic counseling help?

- Collect a detailed family history to identify potential genetic conditions in a family
- Discuss the potential benefits and risks of genetic testing with the patient to help them make an informed decision
- Identify most appropriate genetic test given personal or family history
- Interpret genetic test results for providers, patients and family members
- Coordinate cascade testing of family members at risk to inherit a genetic condition
- Refer patients and providers to appropriate resources

Underutilization of Genetic Counseling

- Study interviewed 2,524 women, stratifying their risk of ovarian cancer based on family history
- 89.5% of women with high risk for ovarian cancer talked to their healthcare provider about their family history
- Only about 20% of women with high risk of ovarian cancer reported being referred to genetic counseling

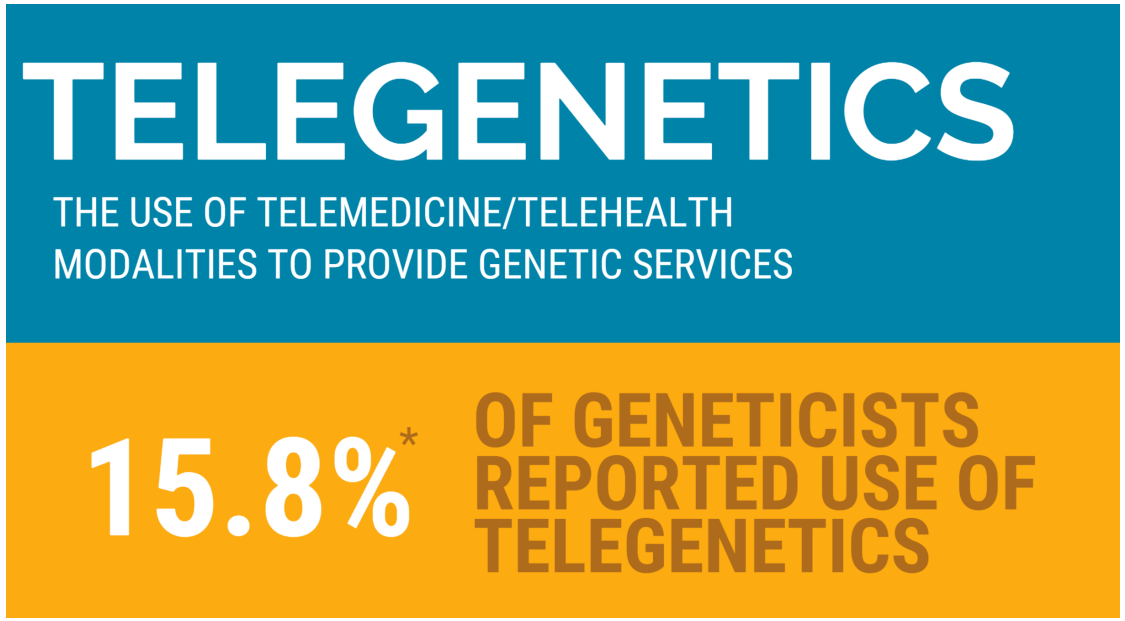


Bellacross et al., 2015

What is telegenetics?

"Telegenetics allows comprehensive genetics services to reach children and families when **travel, distance, and shortage of genetics professionals interfere with access**. By utilizing interactive video and a secure high-speed connection, genetic counselors can "virtually meet" with a patient at a regional clinic or hospital in real time."

National Society of Genetic Counselors

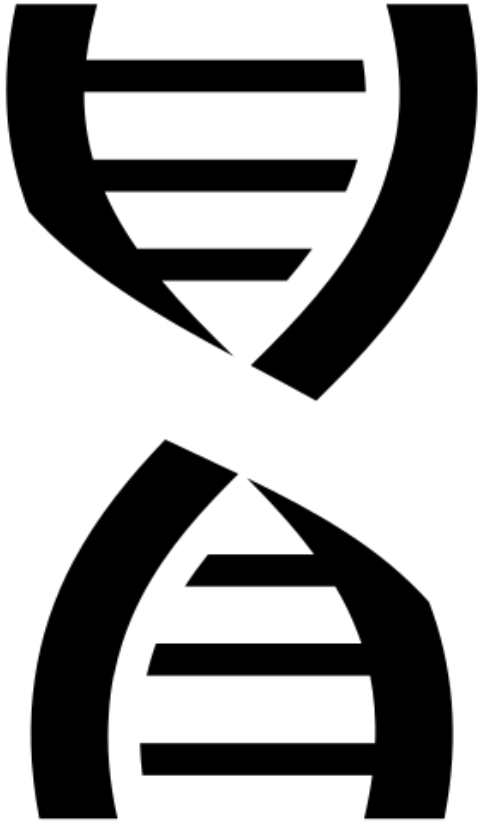


https://nccrcg.org/focus-areas/telegenetics/
https://www.nsgc.org/page/incorporate-genetic-counseling

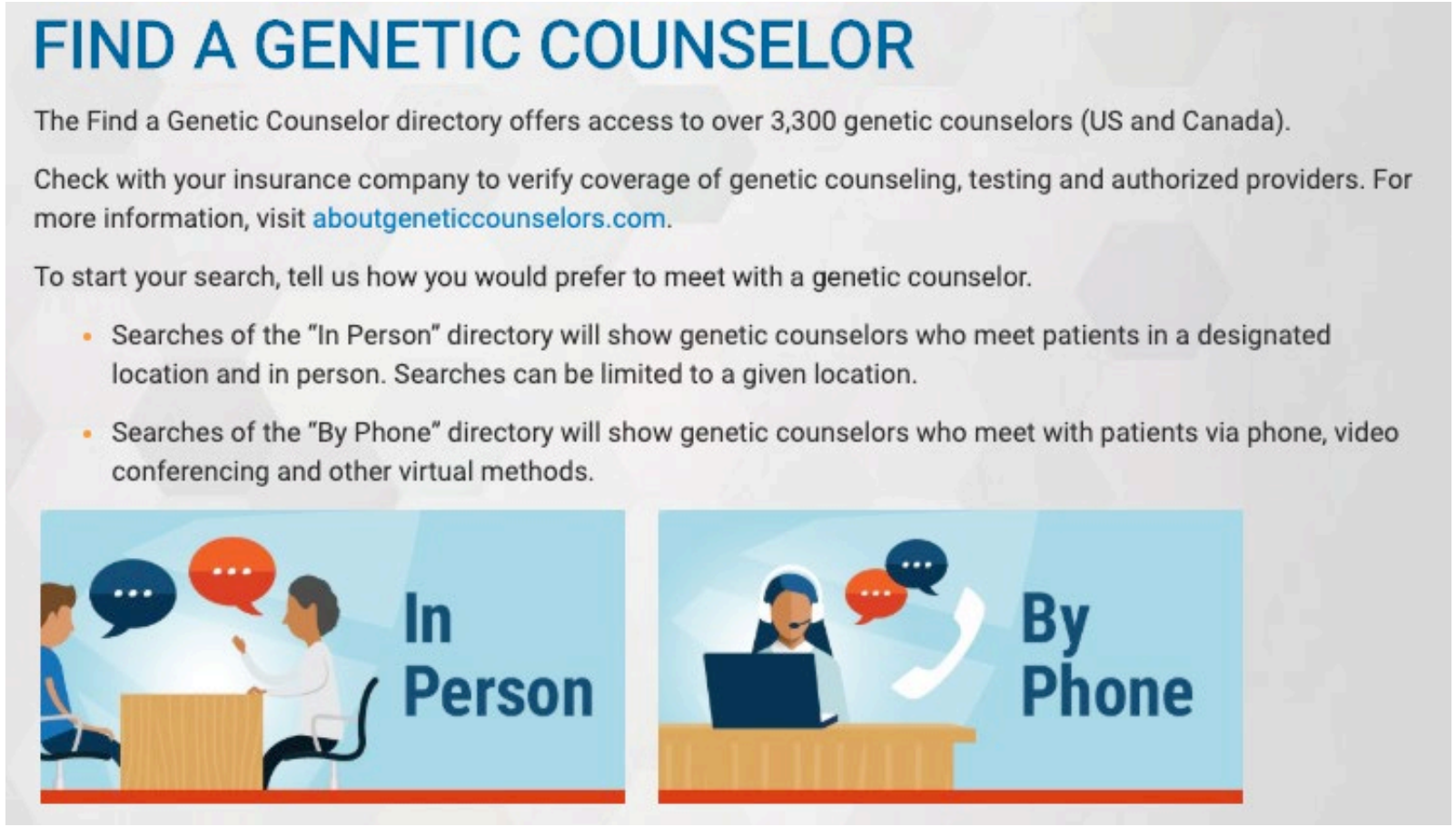
Telegentics Can Expand Genetics Care to Additional Settings

"The use of technology in a healthcare setting *can allow a patient, without local access to geneticist, to receive a genetics assessment* and it can allow *a primary care provider to contact a genetics provider* to become better equipped to provide care to an individual with a complex genetic disorder."

https://nccrcg.org/focus-areas/telegenetics/



NSGC Find a Genetic Counselor



https://www.nsgc.org/page/find-a-genetic-counselor

Resources for Patients

- Genetics Home Reference - https://ghr.nlm.nih.gov/
- National Organization for Rare Disorders (NORD) - https://rarediseases.org/
- American Cancer Society - https://www.cancer.org/
- National Cancer Institute Resources for Patients - https://www.cancer.gov/types

Resources for Providers

- GeneReviews - https://www.ncbi.nlm.nih.gov/books/NBK1116/
- National Cancer Institute Resources for Providers - https://www.cancer.gov/types
- National Society of Genetic Counselors Practice Guidelines – https://www.nsgc.org/page/practiceguidelines
- American College of Medical Genetics and Genomics Practice Resources - https://www.acmg.net/ACMG/Medical-Genetics-Practice-Resources/Practice_Resources/ACMG/Medical-Genetics-Practice-Resources/Medical-Genetics-Practice-Resources.aspx
- National Comprehensive Cancer Network - https://www.nccn.org/

Upcoming Talks in This Series

- Diagnostic Genetic Testing: The Who, What, When and How
  - Shannon Kieran, MS, LCGC, MBA
  - March 12, 2020, 12pm MST

- Direct to Consumer Genetic Testing
  - Shannon Kieran, MS, LCGC, MBA
  - April 9, 2020, 12pm MST

Thank you!

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Improving Access to Quality Medical Care Webinar Series

March 12, 2020: Diagnostic Genetic Testing: The Who, What, When and How

Shannon Kieran, MS, LCGC

April 9, 2020: Direct to Consumer Genetic Testing

Shannon Kieran, MS, LCGC

Please check our websites for upcoming webinars and events

http://www.telemedicine.arizona

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Your opinion is valuable to us.
Please participate in this brief survey:

https://www.surveymonkey.com/r/SWTRCWebinar

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