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

Roles of Genetics Providers in Clinical Care Settings

Valerie Schaibley, PhD

Associate Director, University of Arizona Genetic Counseling Graduate Program
Administrator, Center for Applied Genetics and Genomic Medicine
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Roles of Genetics Providers in Clinical Care Settings

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Arizona Telemedicine Program Webinar Series on Telegenetics

May 14, 2020

Conflict of Interest

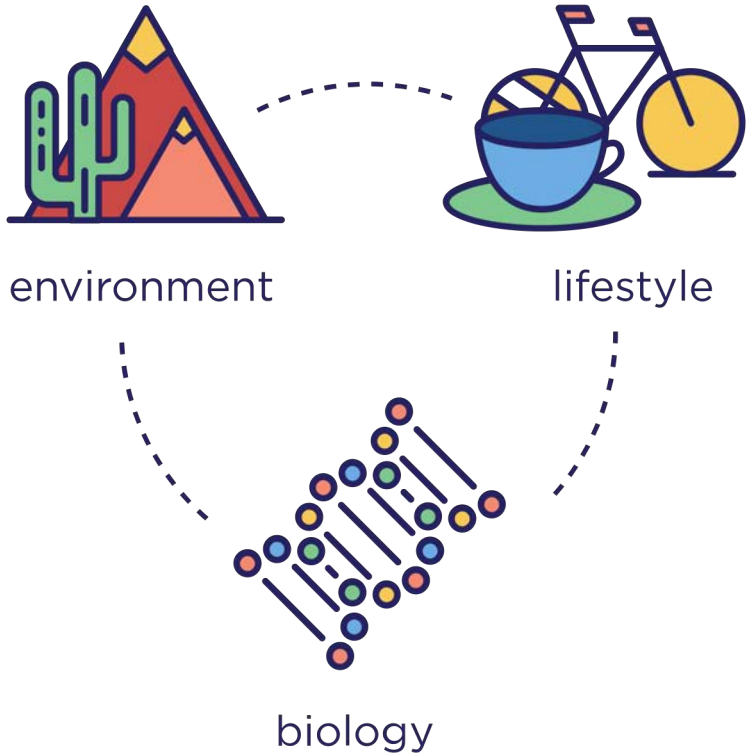
- None to disclose.

Webinar Outline

- Precision medicine is here – are healthcare providers ready?
- Who are specialty genetics providers and how can they help?
- How can telegenetics expand care in genetics?

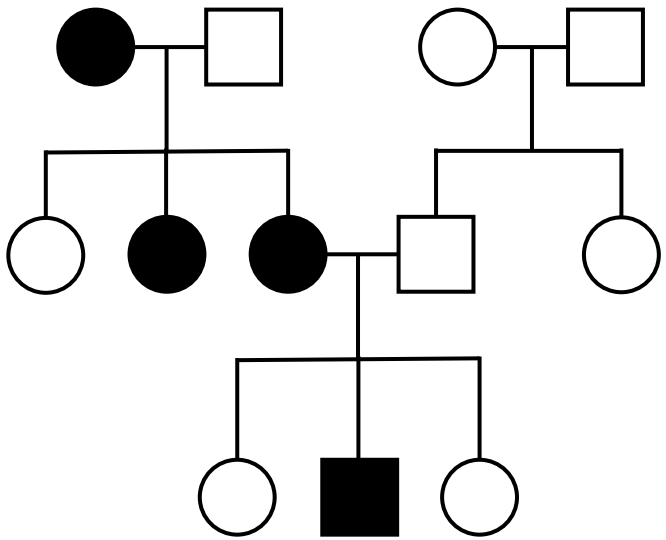
What is precision medicine?

- Precision Medicine Initiative: “An emerging approach for disease treatment and prevention that takes into account individual variability in genes, environment, and lifestyle for each person.”
- Tailoring surveillance and management for an individual
- Incorporating genetic testing into routine clinical care

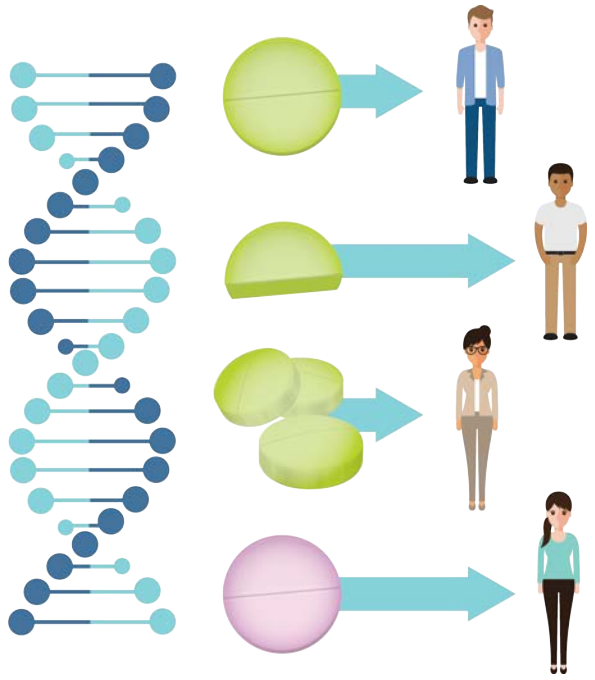


How is genetic data used in the clinic?

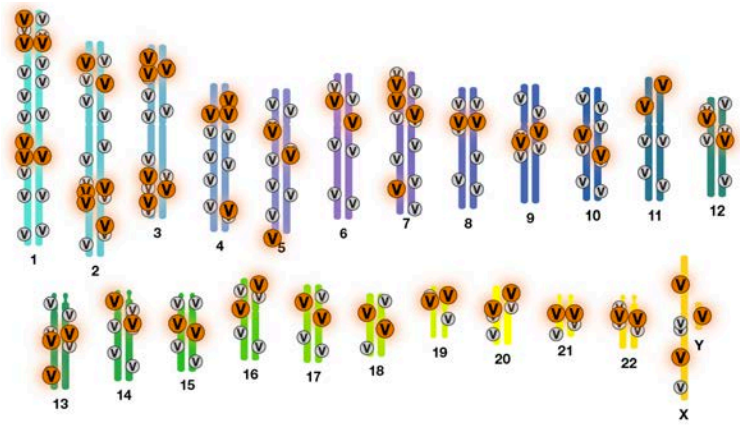
Genetic Diagnostics



Pharmacogenomics/ Precision Therapeutics

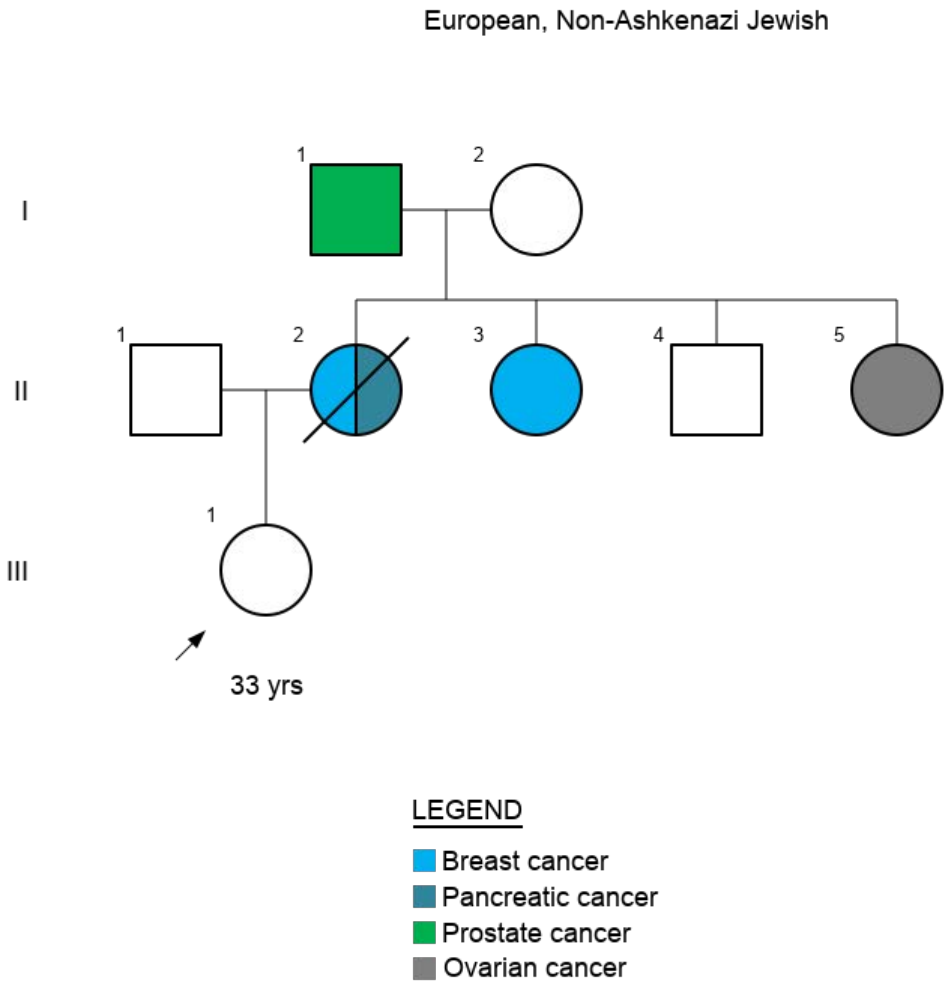


Polygenic Risk Scores



Example: Family History of Cancer

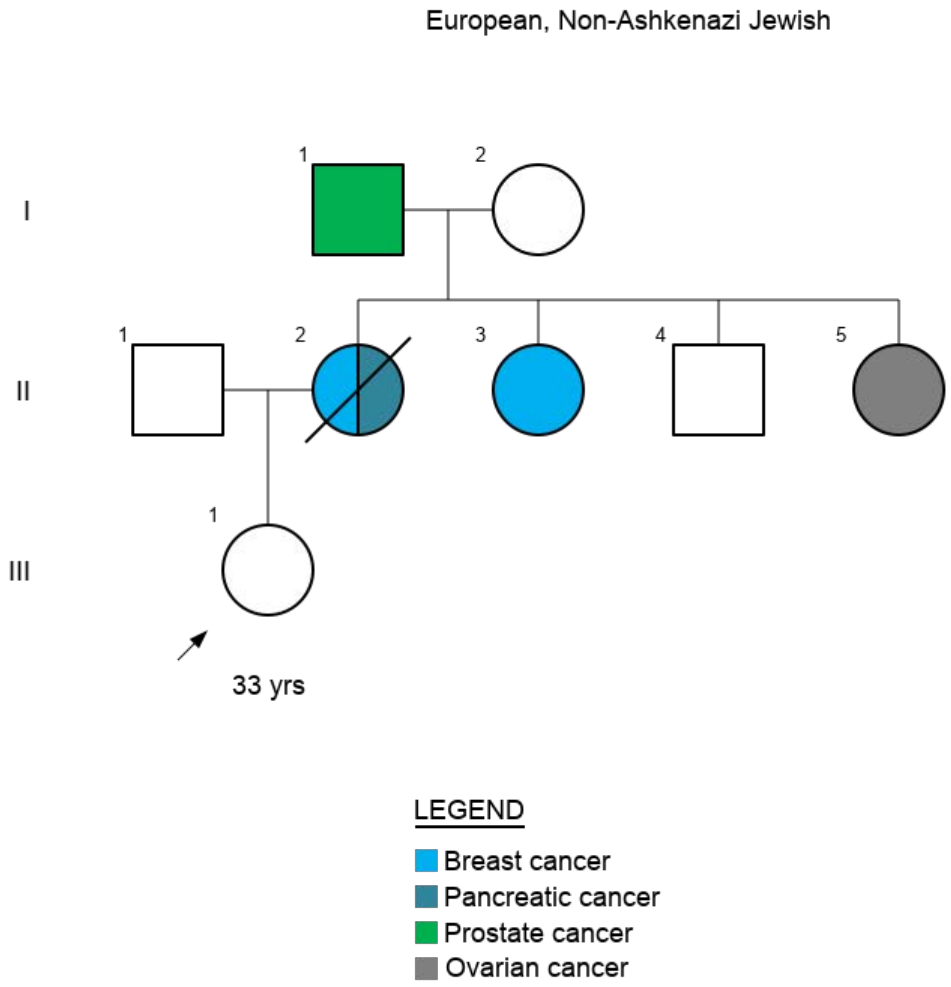
A 33-year-old woman mentions her family history of cancer during a routine gynecological exam. Her mother was diagnosed with **breast cancer at age 45** and later died from **pancreatic cancer at age 55**. One of her maternal aunts was diagnosed with **ovarian cancer at age 58**, another with **breast cancer at age 39**, and her maternal grandfather was diagnosed with **prostate cancer at age 55**.



Example: Family History of Cancer

BRCA2 Screening and Management Guidelines

- Breast: SBE, CBE, mammograms, MRIs, chemoprevention, BL mastectomies
- Ovaries: Bilateral salpingo-oophorectomy (BSO), TV sonograms, CA-125 blood testing
- Prostate: PSA and rectal exam
- Pancreas: endoscopic ultrasound, CA19-9 blood testing
- Male Breast: SBE, CBE
- Melanoma: full body skin exams, eye exams



How are genetic tests integrated into patient care?

Providers refer patients to genetics specialists to order tests and educate patients

Providers directly order tests and educate patients on risks/benefits

Patients self-order tests through consumer-initiated testing and bring results to the provider's office

Data returned to patients as part of participation in a research study

Are healthcare providers ready for precision medicine?

- Objective knowledge to effectively integrate genetic testing into the clinic
- Self-perceived knowledge and confidence in using and interpreting genetic tests
- Time to discuss genetic testing options, limitations and nuances with patients

PCPs Factual Genetics Knowledge

Question	% Correctly Answered
The DNA sequences of two randomly selected healthy individuals of the same sex are 90-95% identical. (FALSE)	31.5%
Most common diseases, such as diabetes and heart disease, are caused by a single gene variant. (FALSE)	96.2%
All the genetic variation in an individual can be attributed to either spontaneous (*i.e. do novo*) or inherited changes in the human genome. (TRUE)	63.6%
Individual genetic variants are usually highly predictive of the manifestation of common disease. (FALSE)	77.7%
Prevalence of many Mendelian diseases differs by racial groups. (TRUE)	86.0%
A patient who is found to be at increased genetic risk can reduce or modify their overall disease risk with changes to their health management, treatment or lifestyle. (TRUE)	97.0%

Adapted from Haga et al., 2019

PCPs Perceived Knowledge of Genetics

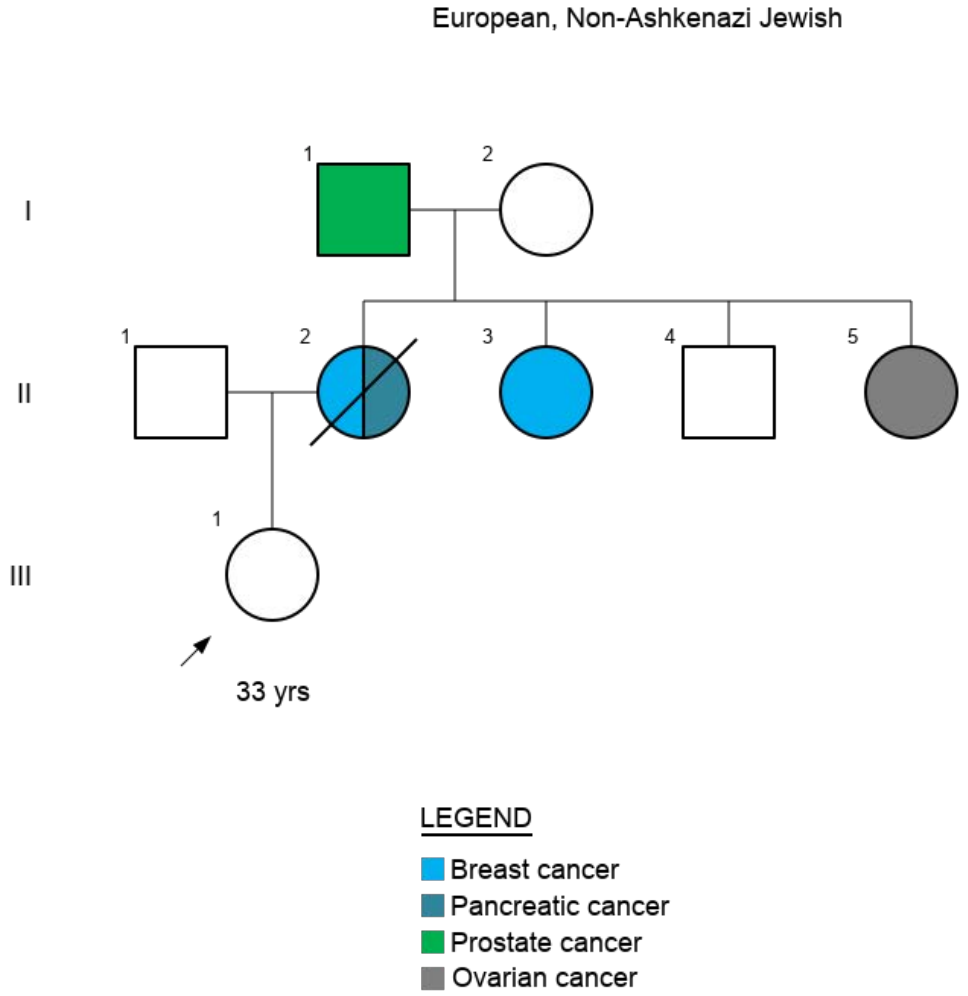
	Pharmacogenomics	Genetics of Complex Disease	Genome-Wide Association Studies	Basic Genetics Principles	When and How to Incorporate Genomic Information into Practice
No knowledge	7.7%	6.9%	52.3%	0.8%	10.0%
Minimal	53.8%	48.5%	40.0%	9.2%	51.5%
Moderate	30.8%	39.2%	6.9%	58.5%	33.8%
Above Average	7.7%	5.4%	0.8%	29.2%	4.6%
Expert	0	0	0	2.3%	0

Adapted from Haga et al., 2019

Example: Family History of Cancer

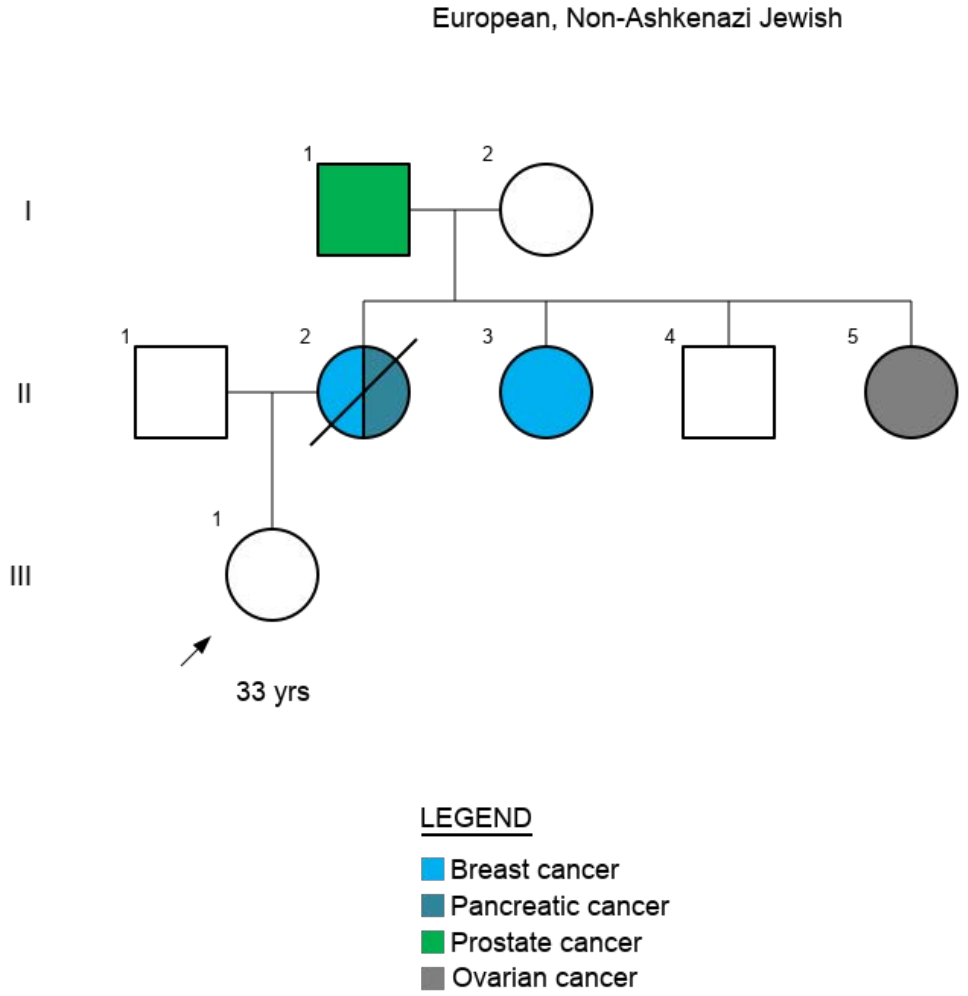
A 33-year-old woman mentions her family history of cancer during a routine gynecological exam. Her mother was diagnosed with **breast cancer at age 45** and later died from **pancreatic cancer at age 55**. One of her maternal aunts was diagnosed with **ovarian cancer at age 58**, another with **breast cancer at age 39**, and her maternal grandfather was diagnosed with **prostate cancer at age 55**.

She tells her provider that she is a 23andMe customer, and her test came back **negative for the breast cancer genes**. When she downloaded the **raw data** and ran it through a 3rd party-interpretation service, it found an **increased risk for breast cancer**.

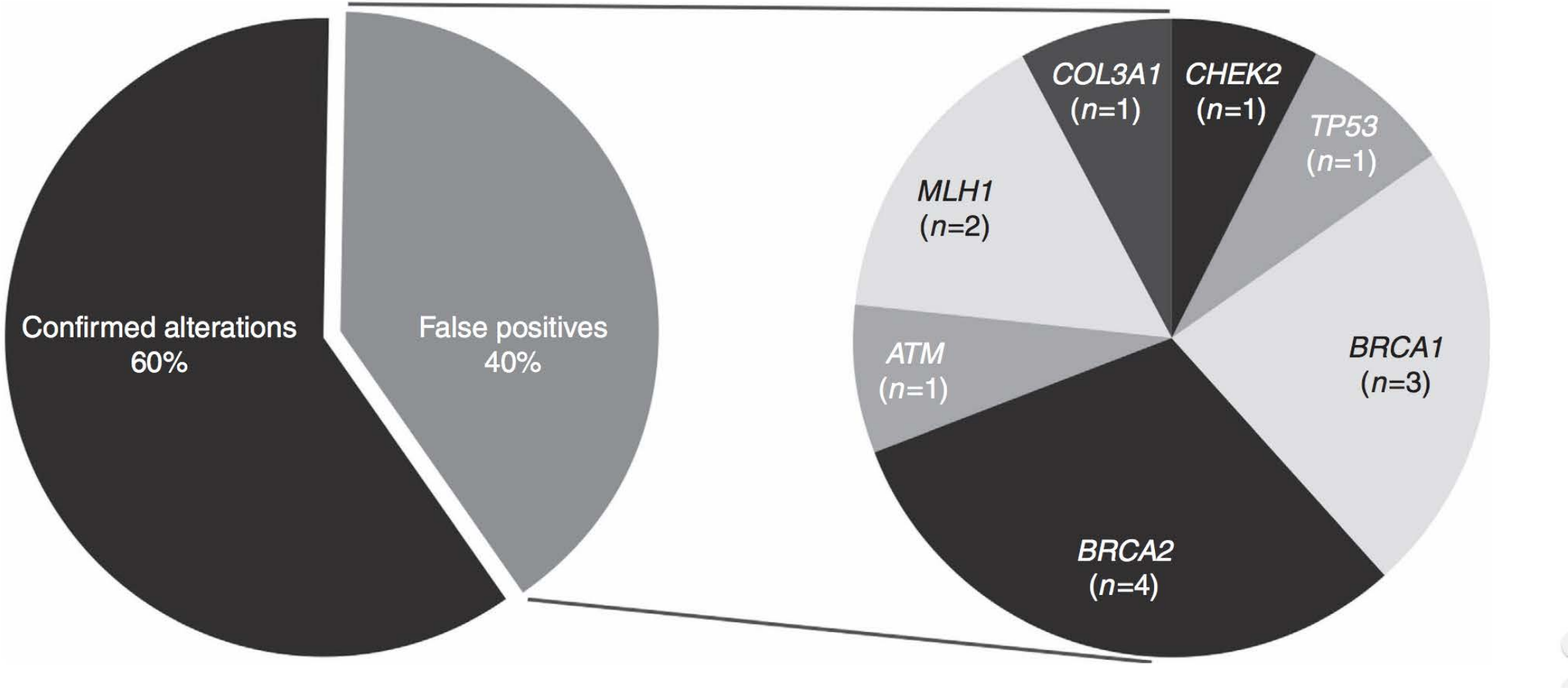


BRCA1/2 in 23andMe

- Tests for three common Ashkenazi Jewish founder mutations
  - *BRCA1* c.68_69delAG (185delAG) – 1% frequency
  - *BRCA1* c.5266dupC (5382insC) – 0.1%-0.15% frequency
  - *BRCA2* c.5946delT (6174delT) – 1.52% frequency
- Combined, they have a frequency of about 1:40 in the Ashkenazi Jewish population
- Much less common in non-Ashkenazi populations



Accuracy of Array-Based Consumer-Initiated Genetic Testing



Tandy-Connor et al. False -positive test results released by direct-to-consumer genetic tests highlight the importance of clinical confirmation testing for appropriate patient care. 2018

How accurate are 3rd party interpretation services?

Gene	Variant	DTC/third party[a]	Ambry[b]	ClinVar[c]	ESP[d]	1000 Genomes[e]	dbSNP[f]
ATM	p.M1040V (c.3118A > G)	Increased risk	Benign	Benign	1.36%	0.95%	1.48%
BRCA1	p.Q356R (c.1067A > G)	Increased risk	Benign	Benign	4.59%	2.81%	3.97%
BRCA2	p.N372H (c.1114A > C)	Increased risk	Benign	Benign	23.32%	24.26%	24.44%
COL3A1	p.A698T (c.2092G > A)	Increased risk	Benign	Benign	21.39%	21.16%	19.16%
COL5A1	c.655-8689C > T	Increased risk	Deep intronic—benign	N/A	N/A	N/A	N/A
COL5A1	c.654+2749A > G	Increased risk	Deep intronic—benign	N/A	N/A	N/A	N/A
COL5A1	c.1827+399C > T	Increased risk	Deep intronic—VUS	N/A	N/A	N/A	N/A
COL5A1	c.1827+1142T > C	Increased risk	Deep intronic—benign	N/A	N/A	N/A	N/A

DTC, direct to consumer; N/A, not available; VUS, variant of unknown significance.
aVariant classification provided by the DTC company or a third-party interpretation service. bVariant classification provided by Ambry. cVariant classification provided in ClinVar (clinical laboratory submissions only). dExome Sequencing Project population frequency database. e1000 Genomes population frequency database. fdbSNP population frequency database.

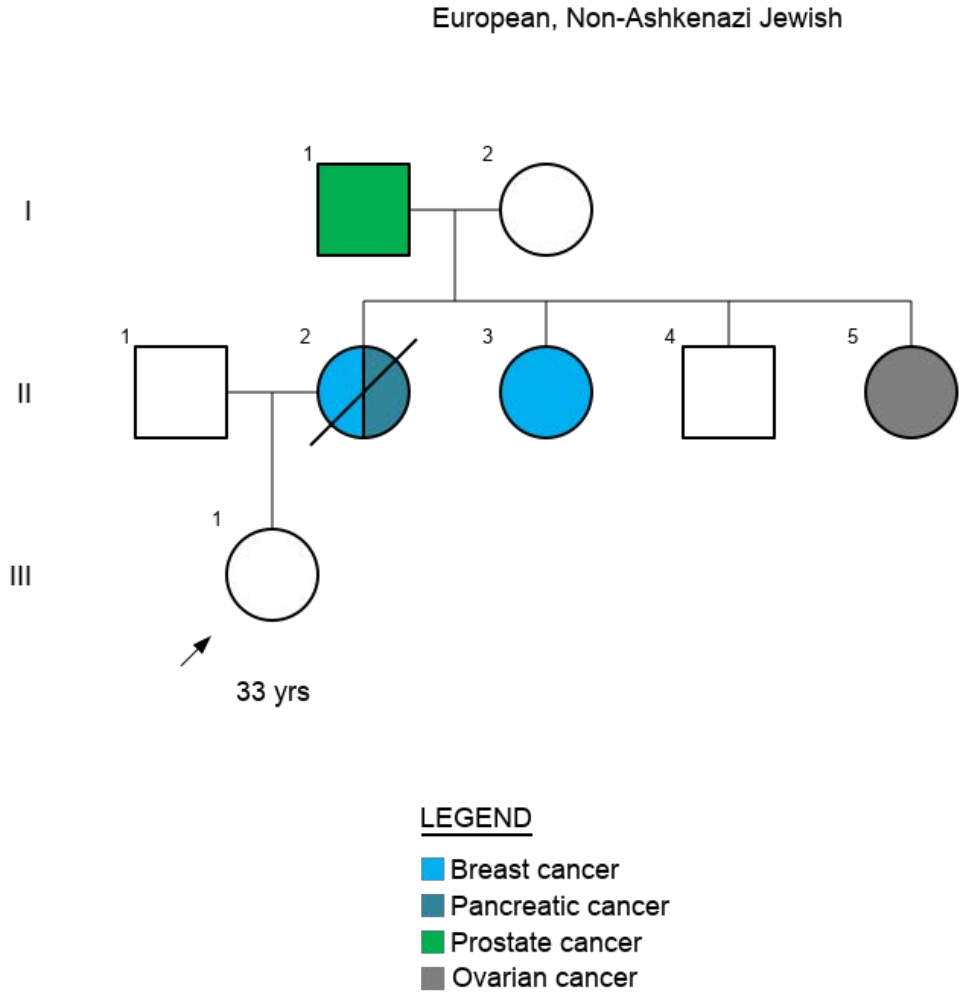
Tandy-Connor et al. False -positive test results released by direct-to-consumer genetic tests highlight the importance of clinical confirmation testing for appropriate patient care. 2018

Example: Family History of Cancer

This patient has a **high risk** for an inherited cancer predisposition syndrome.

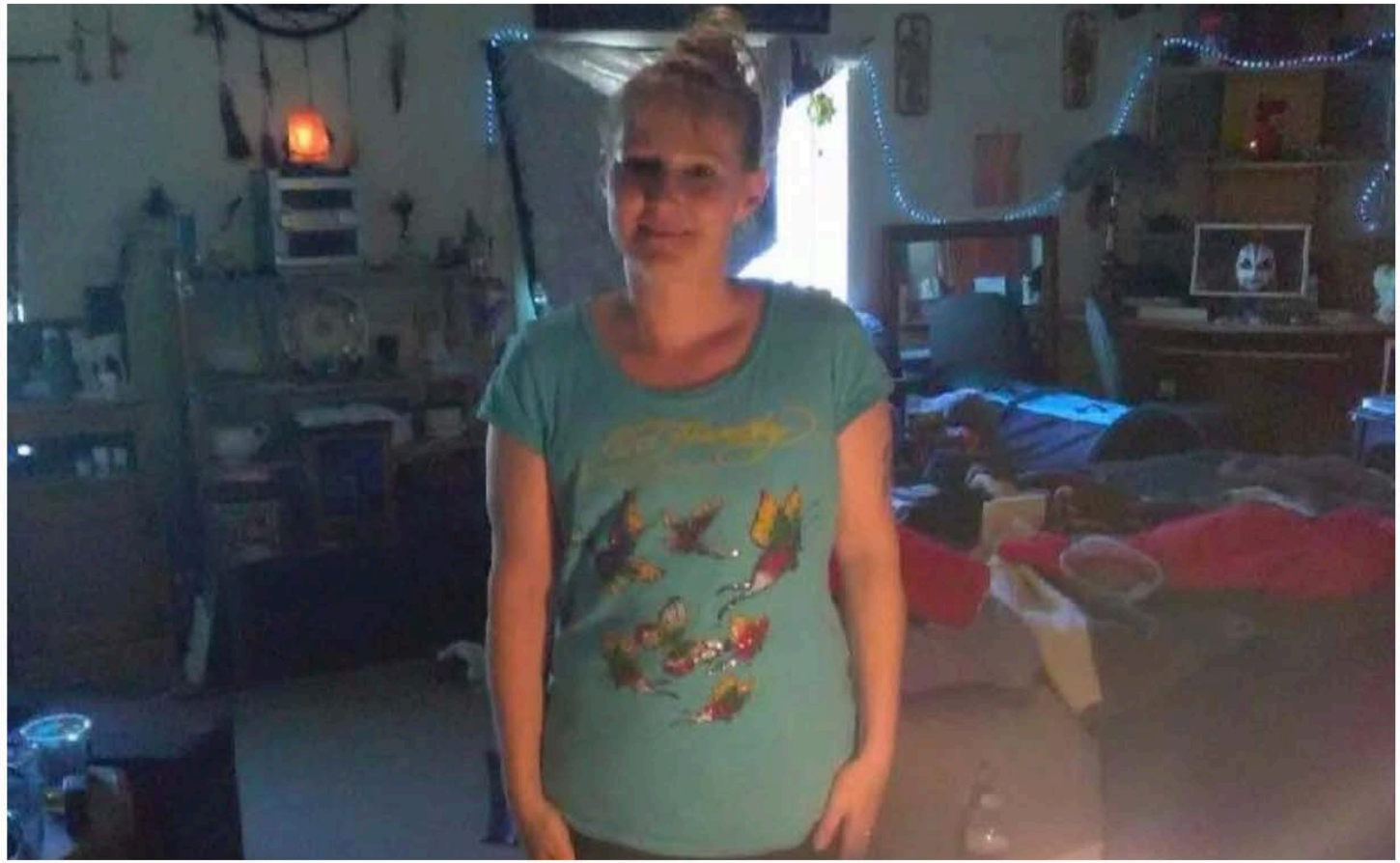
She should be offered **confirmatory diagnostic testing** and should be properly counseled on the **risks and benefits** associated with genetic testing.

If she is positive for a pathogenic genetic variant, **cascade testing** should be offered to her family members.



Health

‘Damaged for the rest of my life’: Woman says surgeons mistakenly removed her breasts and uterus

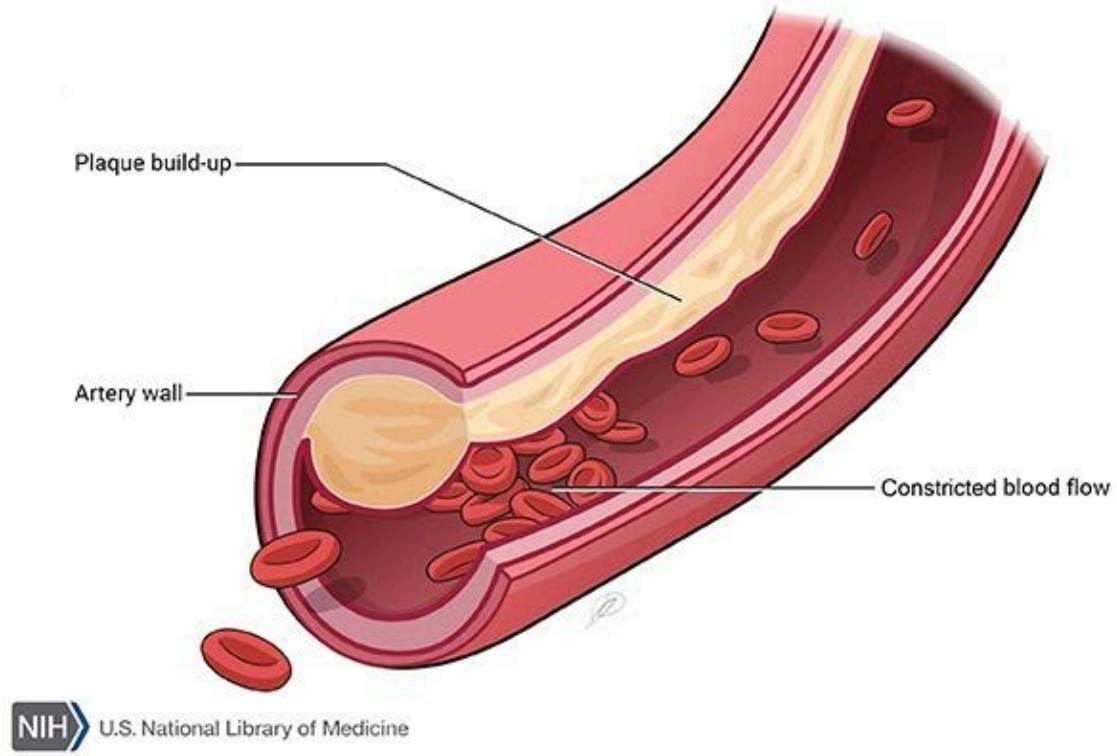


Elisha Cooke-Moore (Elisha Cooke-Moore)

By **Lindsey Bever**

Example: Familial Hypercholesterolemia (FH)

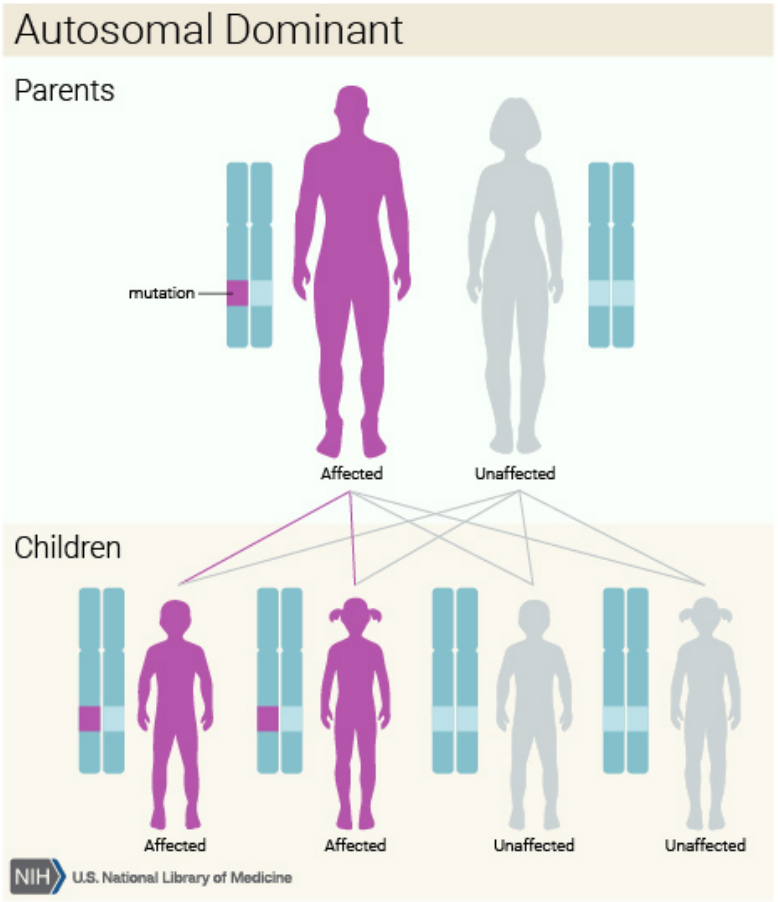
- Characterized by severely elevated LDL cholesterol levels
  - Untreated FH adults: LDL-C > 190 mg/dL
  - Untreated FH children/adolescents: LCL-C > 160 mg/dL
- Untreated men – 50% risk for coronary event (angina/myocardial infarction) by age 50
- Untreated women – 30% risk for coronary event (angina/myocardial infarction) by age 60
- Treated with cholesterol-lowering medications and aggressive management of lifestyle risk factors
- FH affects about 1 in 250 people
- Most common inherited heart condition



https://ghr.nlm.nih.gov/condition/familial-hypercholesterolemia
https://www.ncbi.nlm.nih.gov/books/NBK174884/

Genetics of FH

- Caused by genetic variants in 3 genes that are important for the normal function of lipoprotein receptors which regulate cholesterol levels in the bloodstream
  - *LDLR (*most common gene involved in FH)
  - *APOB*
  - *PCSK9*
- Autosomal dominant inheritance
  - A pathogenic variant in one allele is sufficient to cause the condition
  - Children of an affected parent have a 50% chance of inheriting FH
  - Siblings of an affected person have a 50% of having FH



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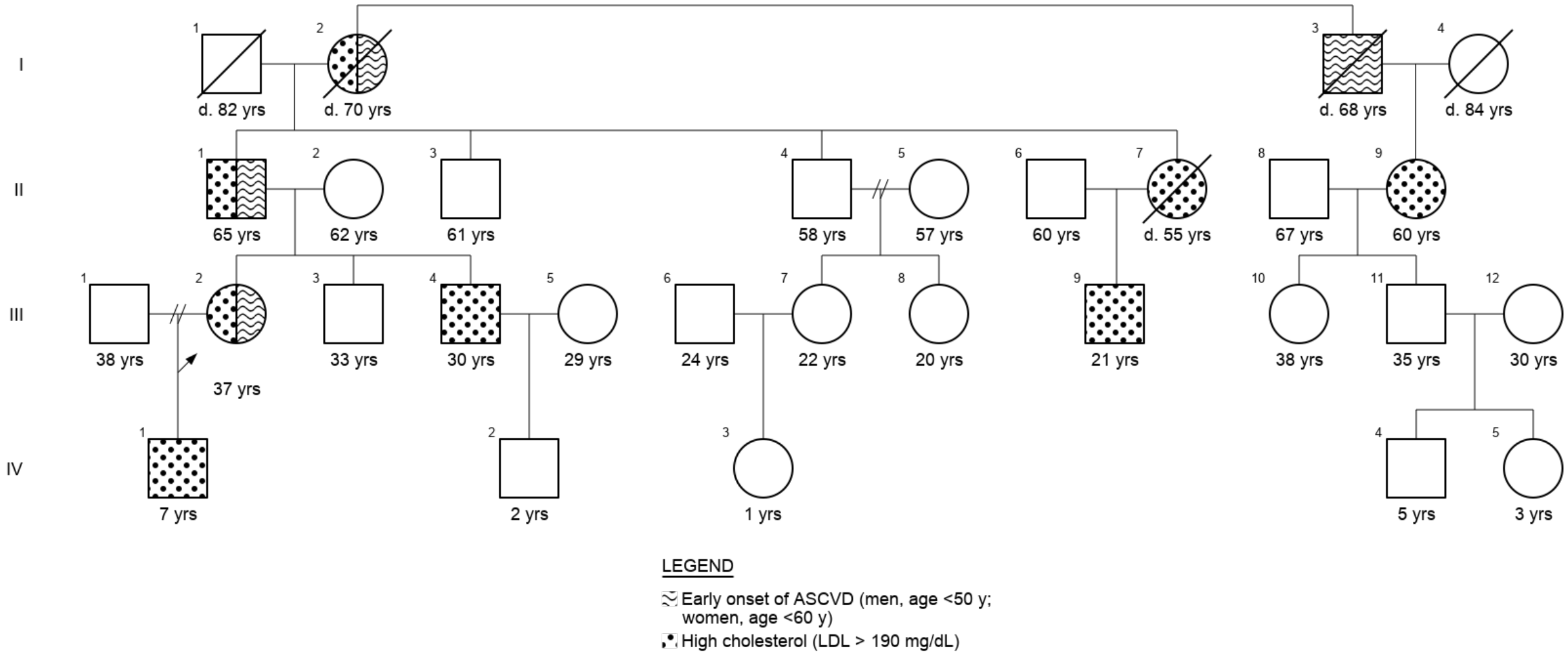
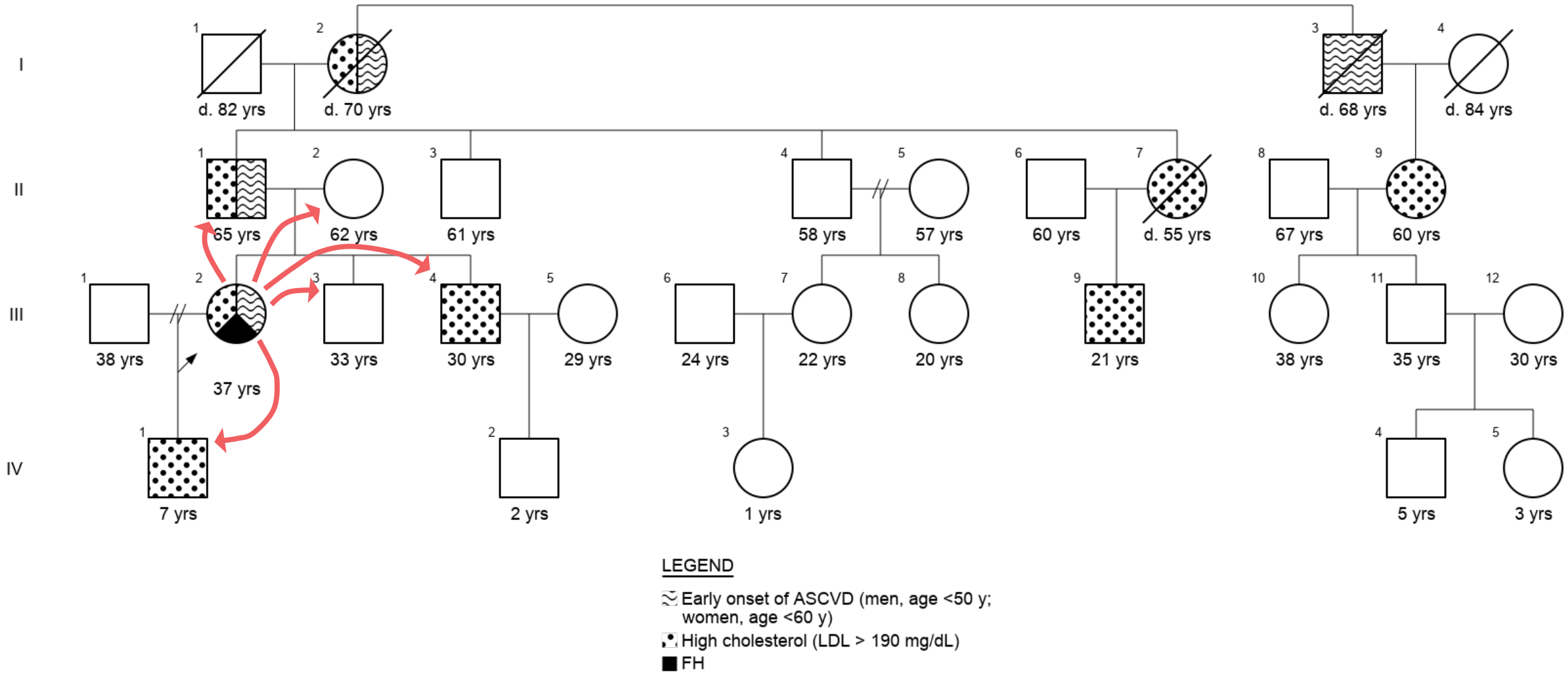


Adapted from Knowles et al., 2017



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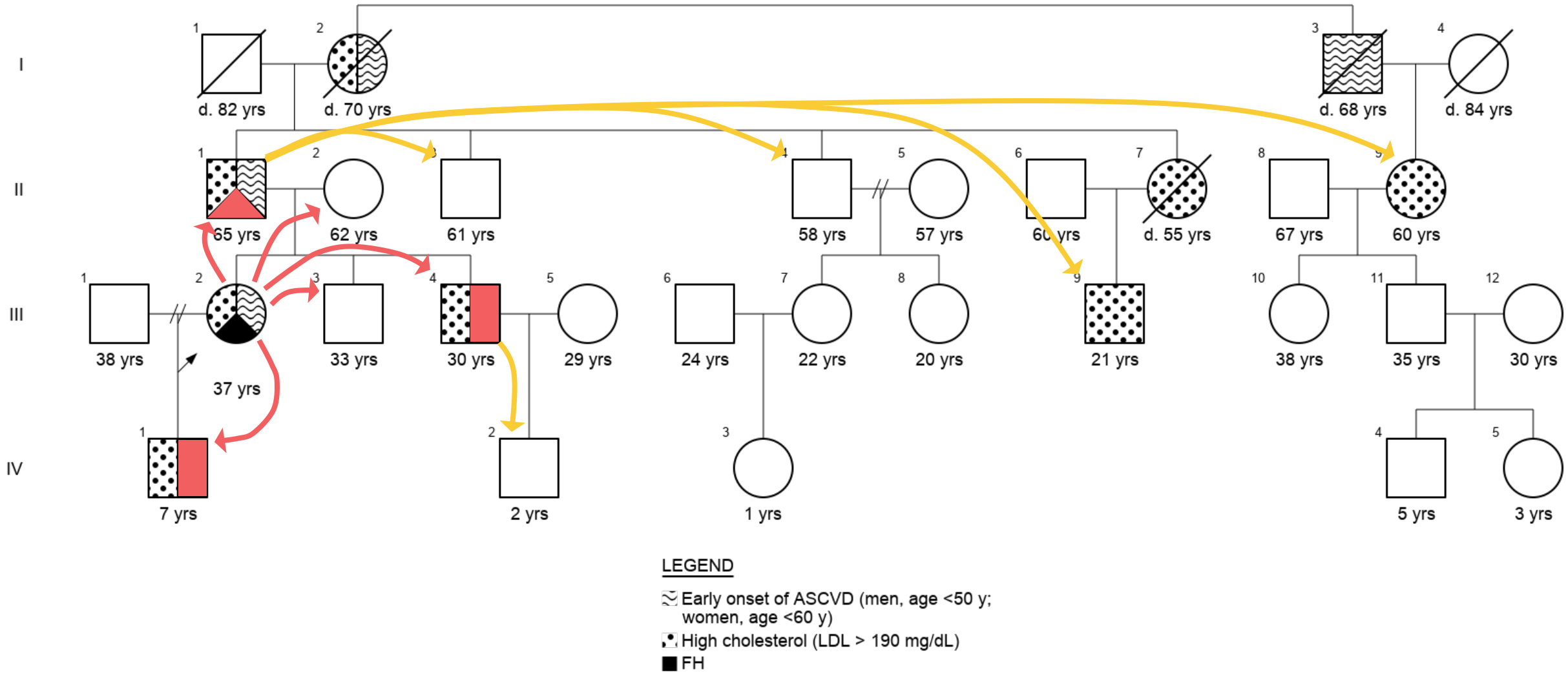

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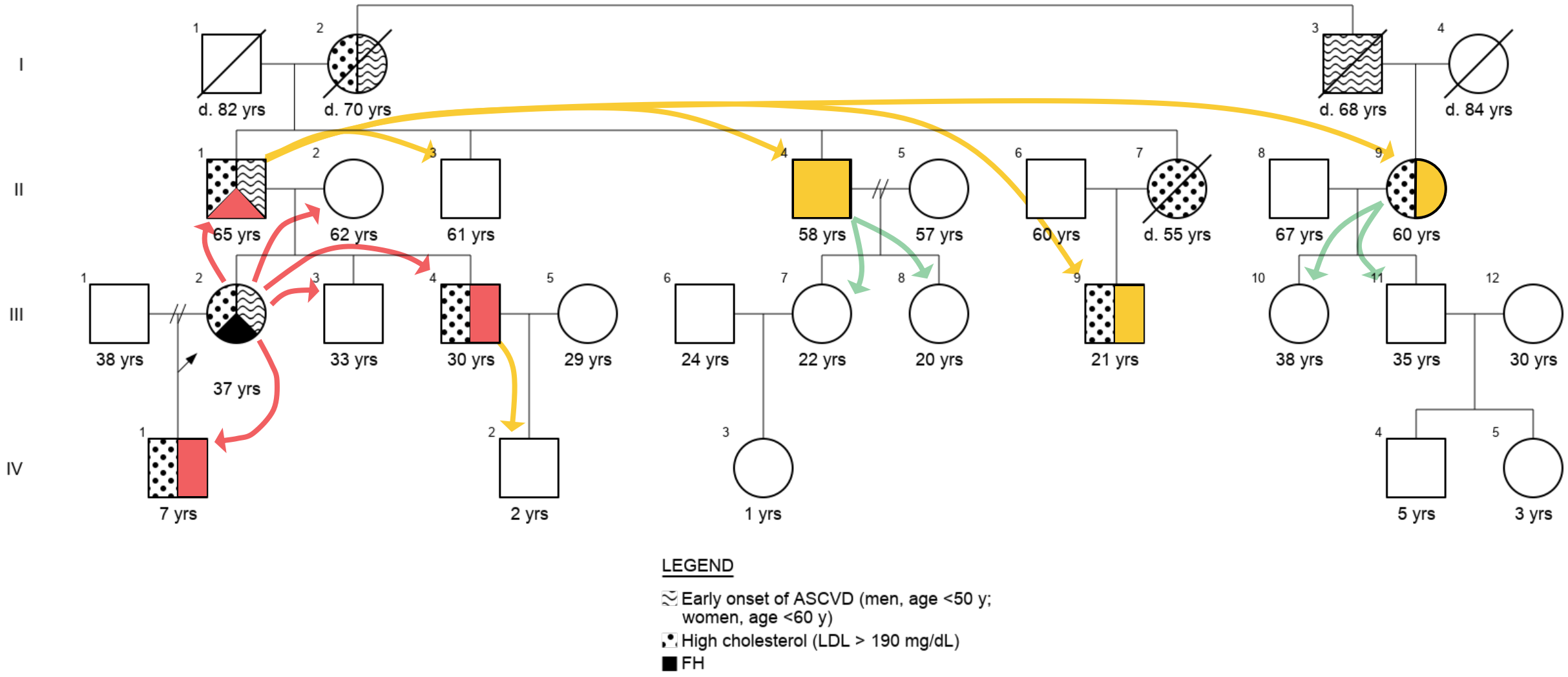

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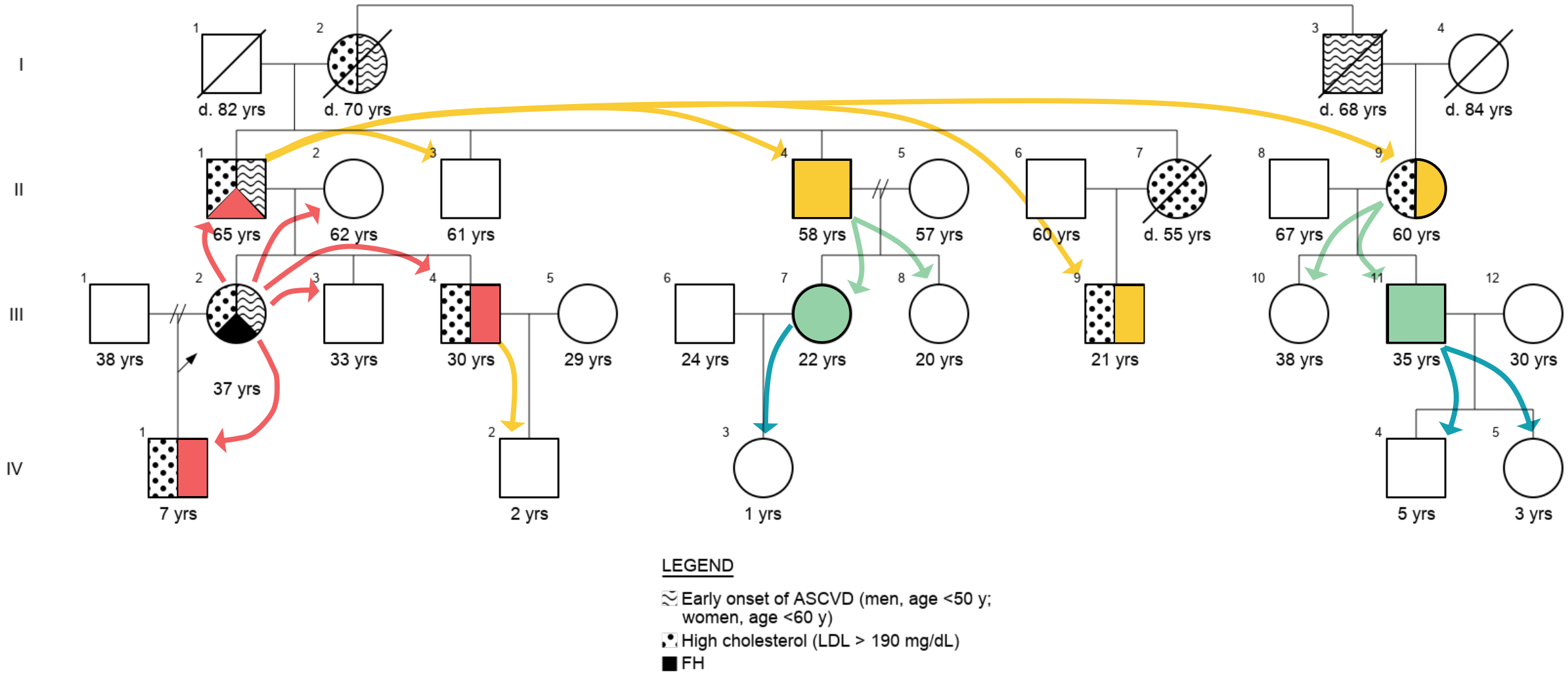

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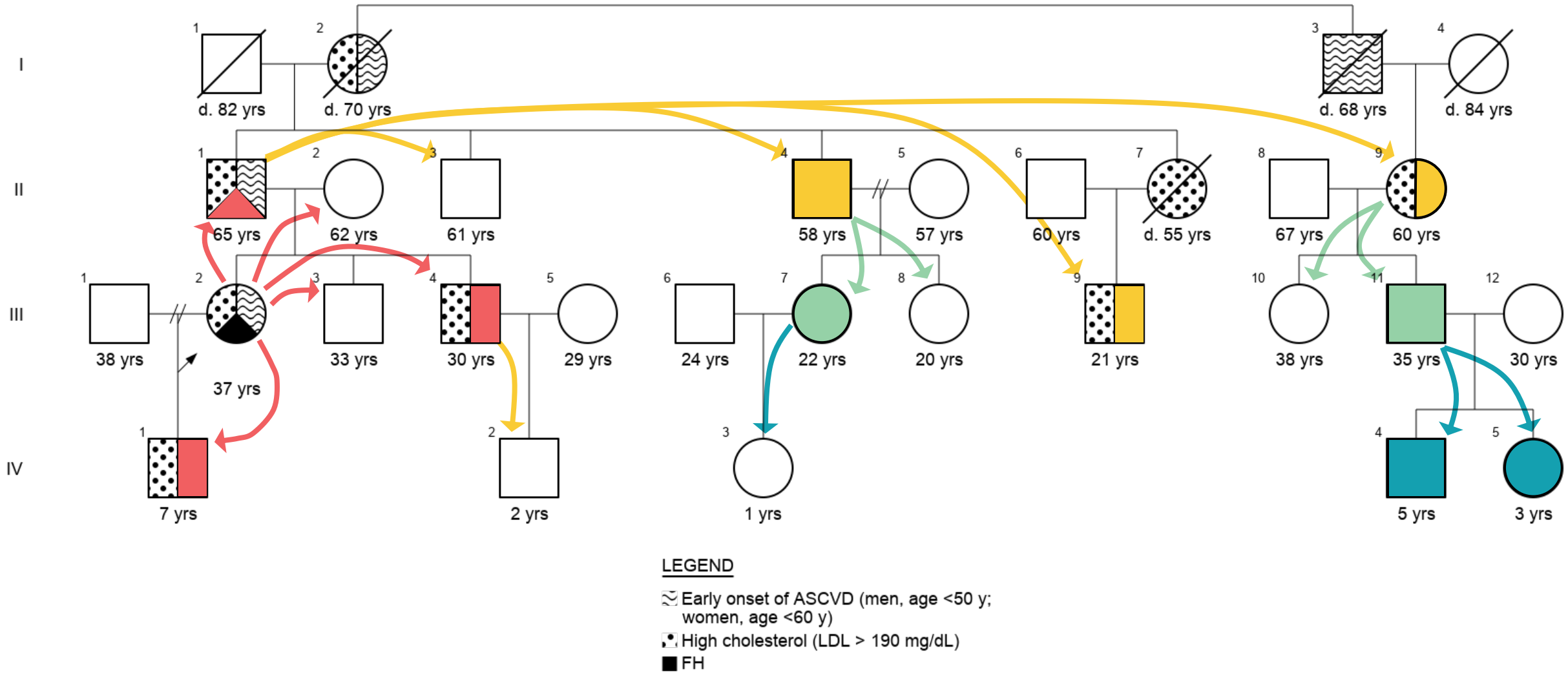

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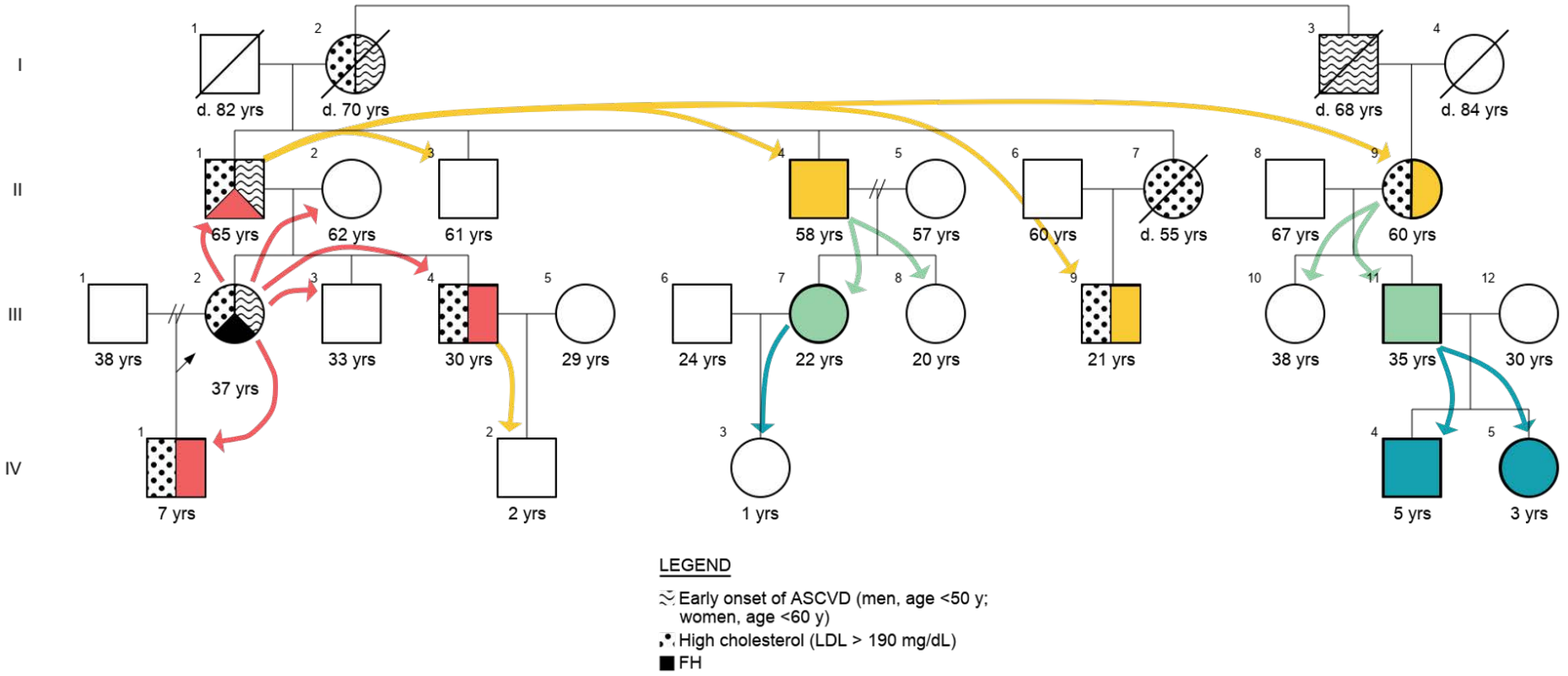
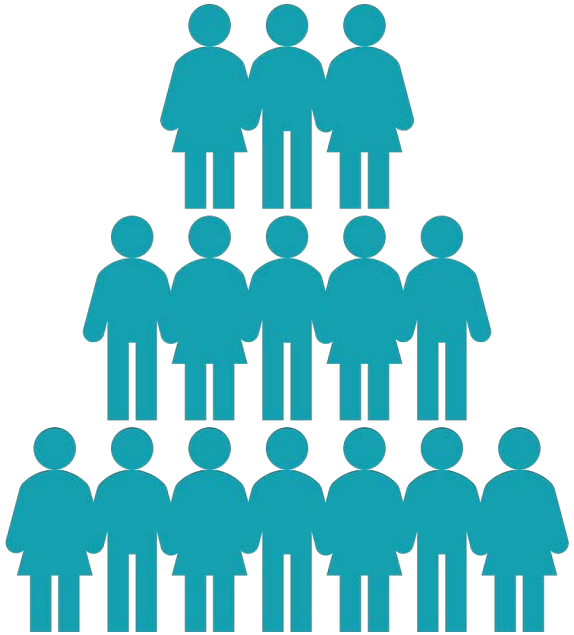


Cascade Cycle	Cumulative Number of Cases
1	3
2	6
3	8
4	10

Adapted from Knowles et al., 2017

Potential Barriers to Cascade Testing

- Family structure and dynamics
- Geographic dispersion
- Health care literacy
- Access to healthcare
- Access to genetics providers
- Privacy concerns



Knowles et al., 2017

How can we get better access to genetics care for patients?

1

Develop educational programs for providers to expand their knowledge and comfort with genetics

2

Better integrate genetics and genetic testing into medical school curricula

3

Work with genetics providers who specialize in genetic medicine

Genetics Health Care Providers

Clinical geneticists

Genetic counselors

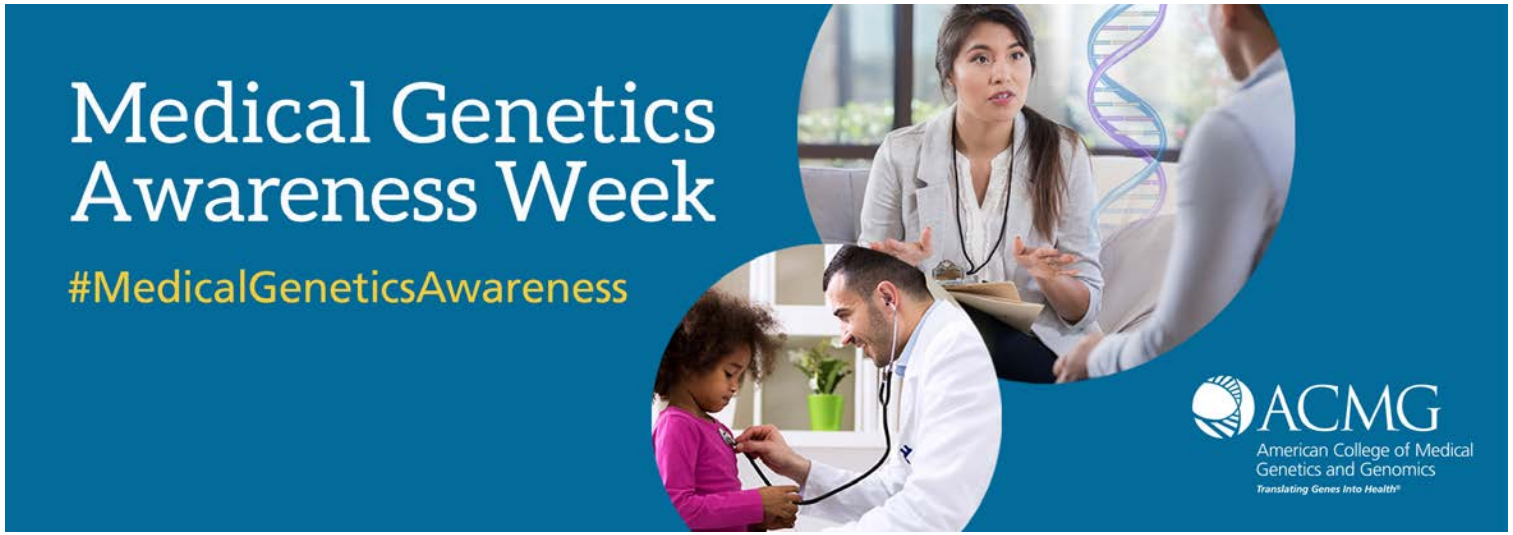
Genetics nurses

Other healthcare providers

Who are clinical geneticists?

"Clinical geneticists are **physicians** who care for patients in **clinical settings** and often carry out clinical or translational research related to patient care. They hold **American Board of Medical Genetics and Genomics (ABMGG) certification** in the specialty of **clinical genetics and genomics** and have **broad training** in the evaluation, diagnosis, management and treatment of **inherited conditions in patients across all ages** from birth to adulthood."

American College of Medical Genetics

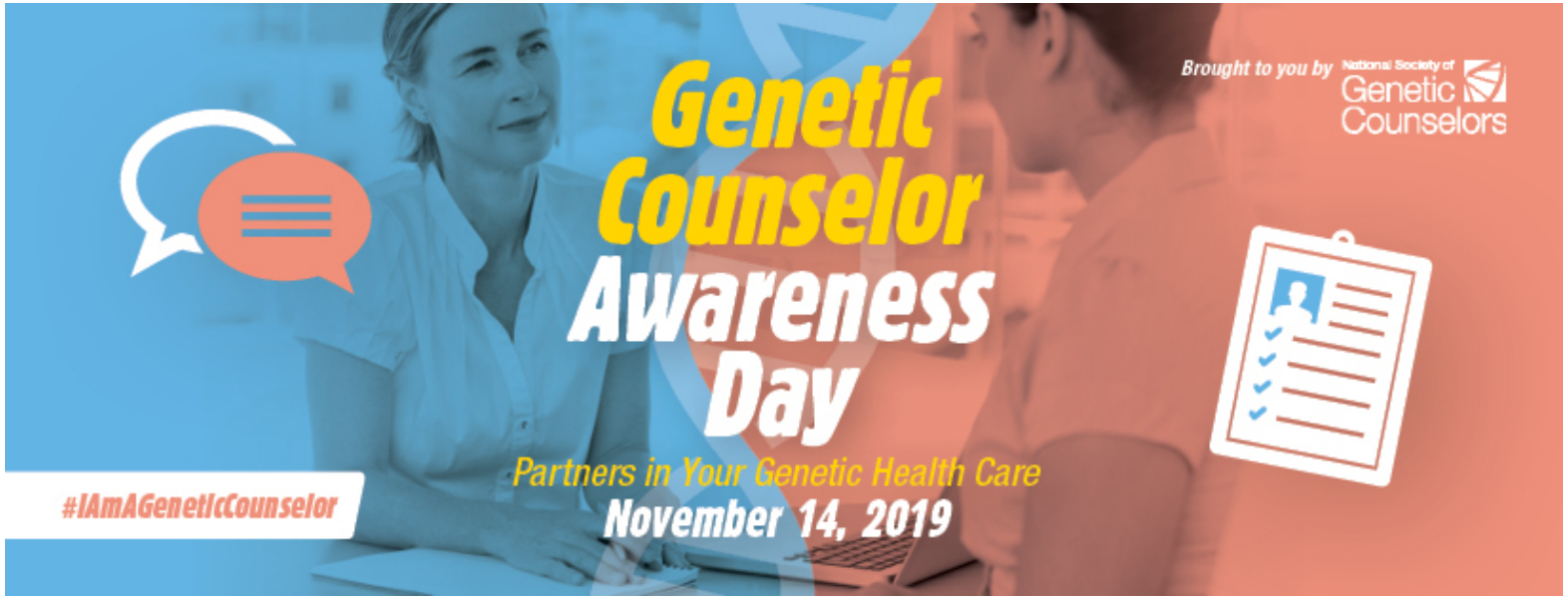


https://www.acmg.net/ACMG/ACMG/Education/Student/Careers_in_Medical_Genetics.aspx

Who are genetic counselors?

"Genetic counselors are professionals who have **specialized education in genetics and counseling** to provide **personalized help** patients may need as they **make decisions about their genetic health**."

National Society of Genetic Counselors

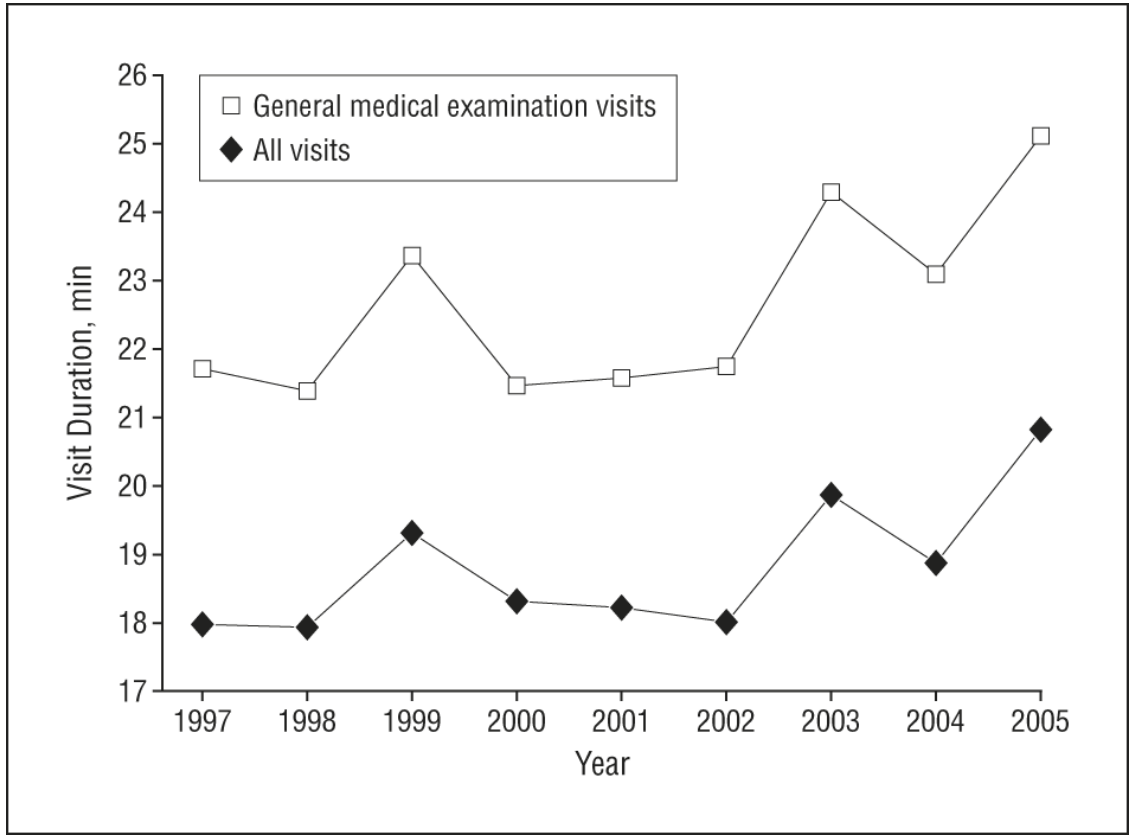


Who are genetic counselors?

- Board certification by the American Board of Genetic Counseling (ABGC)
- Trained medical professionals
- Deliver pre and post test counseling
- Work with the healthcare team to identify appropriate testing
- Work in a variety of specialized areas of medicine (prenatal, pediatrics, adult, cancer, etc)

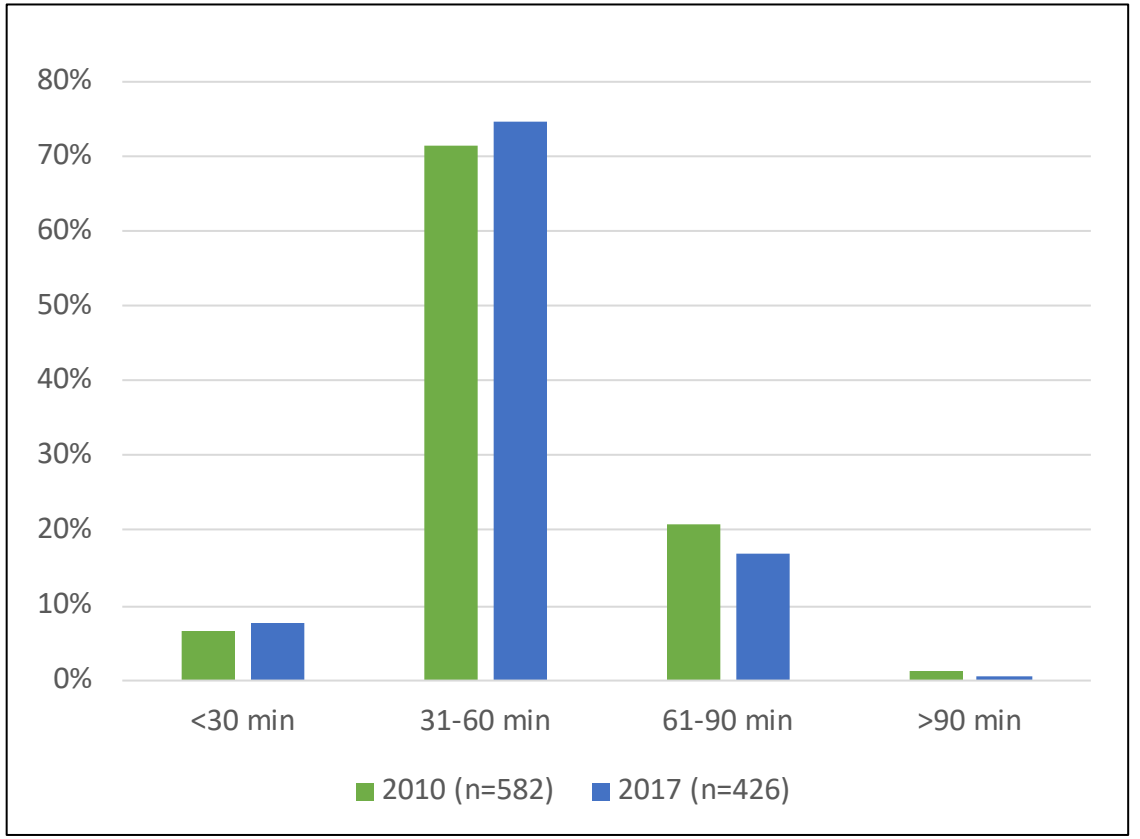
Genetic Counselors Can Spend More Time Counseling Patients on Genetic Testing

Average Primary Care Visit Duration



Chen et al., 2009, JAMA Internal Medicine

Average Genetic Counseling Visit Duration

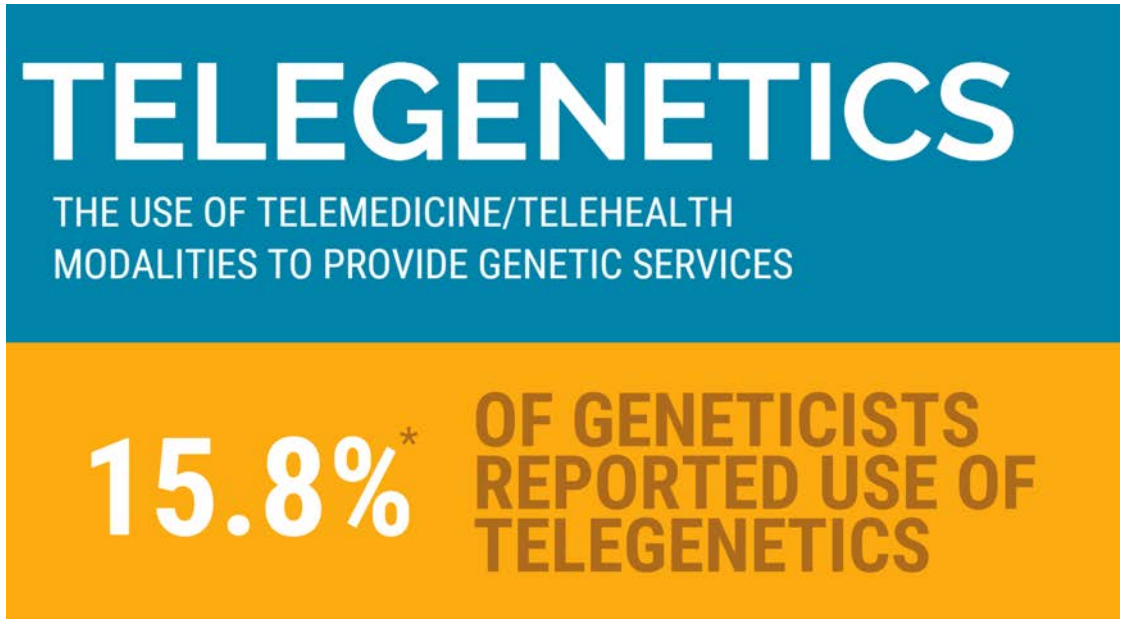


Greenberg et al., 2020, Journal of Genetic Counseling

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

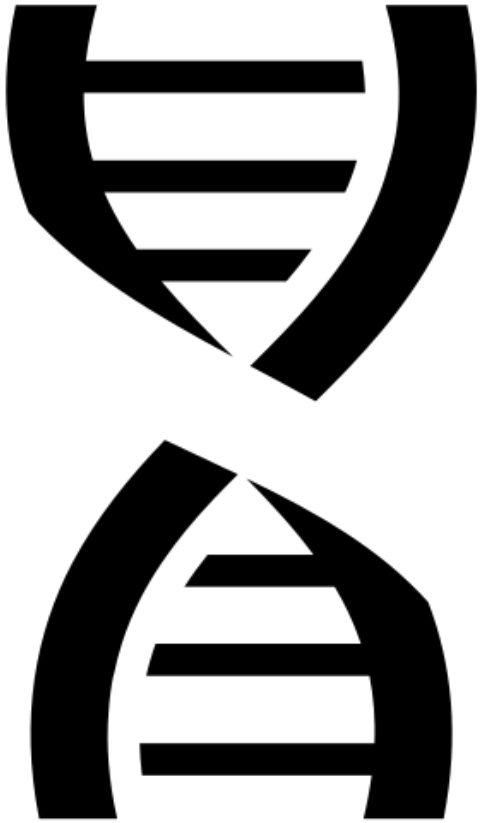


https://nccrcg.org/focus-areas/telegenetics/
https://www.nsgc.org/page/incorporate-genetic-counseling

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/



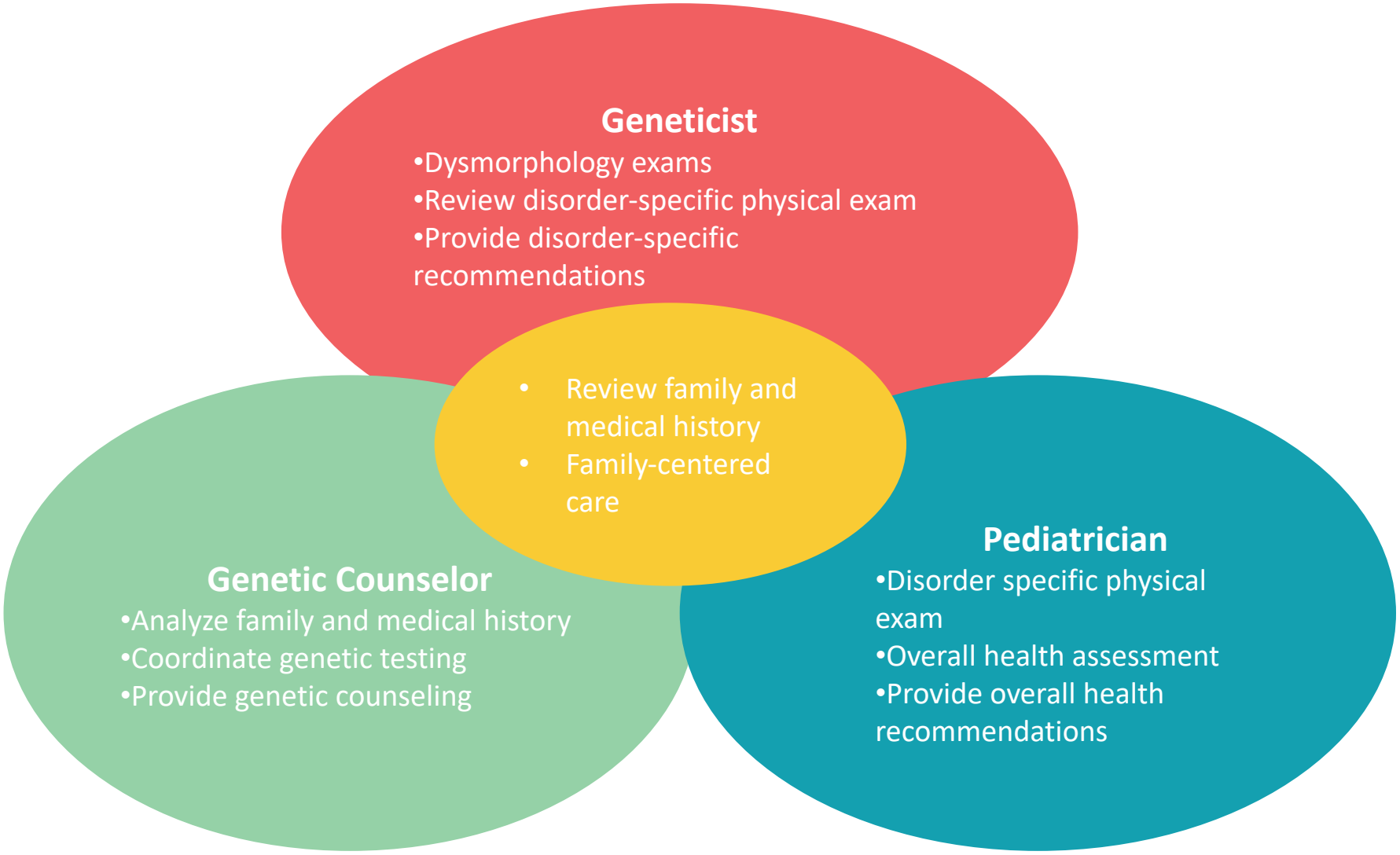
Genetics inMedicine | ORIGINAL RESEARCH ARTICLE Official journal of the American College of Medical Genetics and Genomics

Open

A novel approach in pediatric telegenetic services: geneticist, pediatrician and genetic counselor team

Shobana Kubendran, MBBS, MS, Siddharthan Sivamurthy, MD and Gerald Bradley Schaefer, MD

Pediatric Telegenetics Clinic: An Example from Kansas

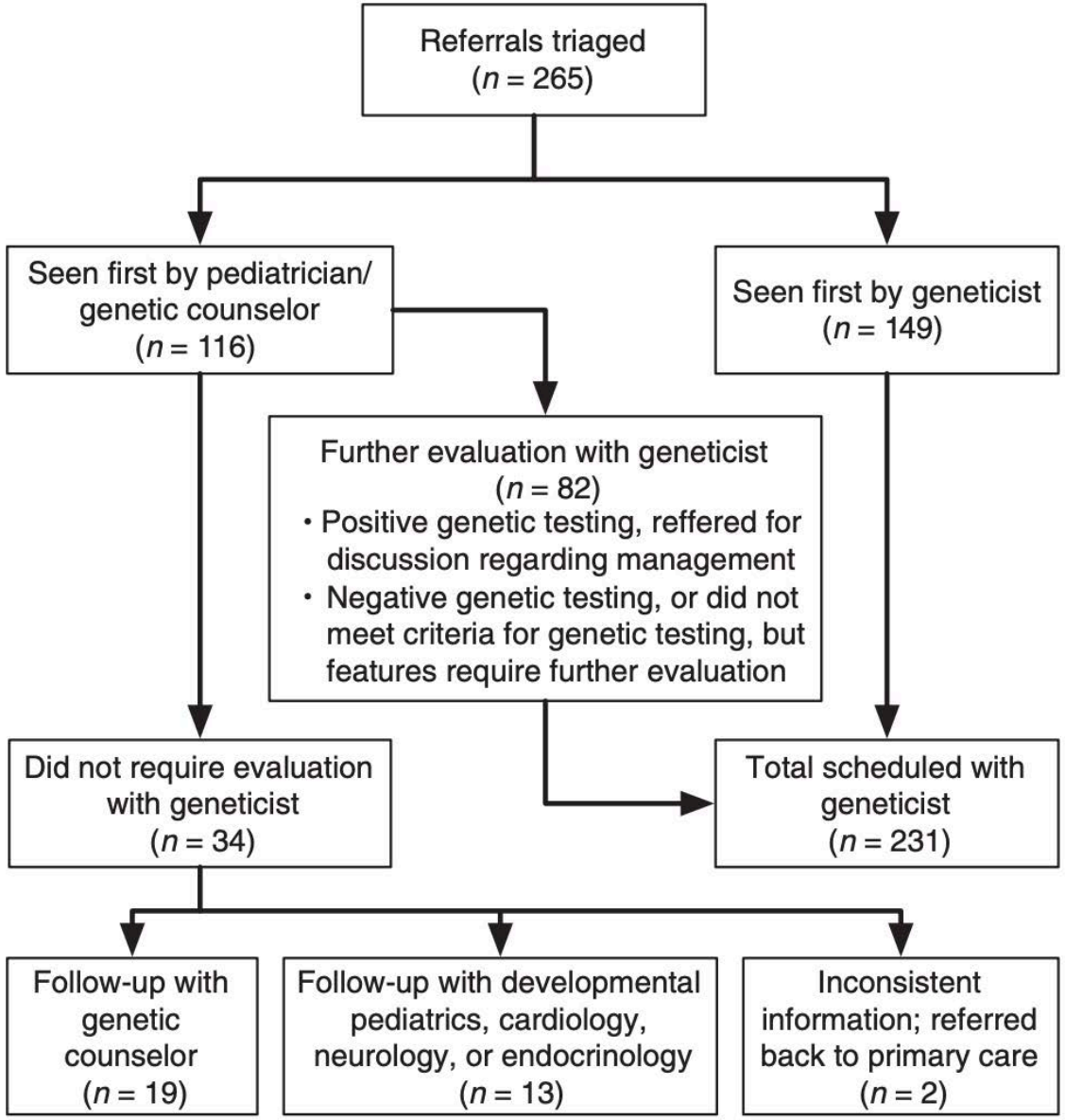


Distant Site: Geneticist

Local Site: Pediatrician and genetic counselor on site with patient

Adapted from Kubendran et al., 2017

Referral and follow-up flow chart



Pediatric Telegenetics Clinic: An Example from Kansas

- Overall, I was satisfied with the visit today
  - 0% Strongly disagree
  - 0% Disagree
  - 24% Agree
  - 79% Strongly agree
- Despite the obstacles to receiving care in person, I would still prefer to travel to see the specialist
  - 25% Strongly disagree
  - 43% Disagree
  - 18% Agree
  - 13% Strongly agree

Kubendran et al., 2017

HEALTH

23andMe study to recruit sickest Covid-19 patients in bid to unravel role of genetics in disease

By REBECCA ROBBINS @rebeccadrobbins / MAY 13, 2020

ADOBE

Thank you!

vschaibley@arizona.edu

THE UNIVERSITY OF ARIZONA
COLLEGE OF MEDICINE TUCSON
Genetic Counseling
Graduate Program

THE UNIVERSITY OF ARIZONA HEALTH SCIENCES
Center for Applied Genetics
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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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