



HOW GENETICS CAN & SHOULD FIT INTO YOUR PRACTICE:

A NON-GENETICS PA'S PERSPECTIVE

Deanna Bridge Najera, MPAS, MS, PA-C

AAPA Annual Conference, May 2022

Lecture Bundle – check out all the talks!

- **IN22126 - Practical Cases in Direct-to-Consumer Genetic Testing**– Nguyen Park, MS, PA-C
- **IN22016 - How Genetics Can & Should Fit Into Your Practice: A Non-Genetics PA's perspective**- Deanna Najera, MPAS, PA-C
- **IN22141 - The Genetics Evaluation: Who to Refer and What to Expect**– Wesley Patterson, PhD(c), MSPA, PA-C

Objectives

The background features a teal and light blue color scheme. A prominent graphic is a DNA double helix structure, with one strand in a lighter teal and the other in a darker teal. Vertical black bars of varying heights are positioned between the strands, resembling a bar chart or a genetic map. The overall aesthetic is clean and scientific.

- Analyze 3-generation pedigrees and learn how to obtain them
- Identify resources which can assist PAs with obtaining a targeted family history for possible genetic causes of disease
- Describe patient motivations and concerns regarding genetic/genomic testing
- Review common presentations in primary care/specialty offices of disorders with genetic underpinnings

Why should I care? Nature vs Nurture



OR



- Study of nearly 45 million people looked at 560 common conditions
 - Nearly 40 percent of the diseases had a genetic component,
 - 25 percent were driven at least in part by factors stemming from sharing the same household, social influences, and the like.
- Purely genetic disorders are individually rare,
 - Account for ~80% of rare disorders,
 - Several thousand rare disorders
 - Means 1 in 17 individuals are affected by them

Why should I care?

Cardiovascular

Early CAD
Sudden death
AAA/TAA

Pulmonary

Asthma/ Atopy
COPD
Cystic fibrosis

Eyes

Macular
degeneration
Colorblindness

GU

Polycystic kidney
disease
Kidney stones

Heme/Onc

Clotting
disorders
Multiple cancers

ENT

Hearing loss/
impairment

GI

Inflammatory
Bowel Disease
Lynch Syndrome

Women's Health

Recurrent
pregnancy loss
GYN cancers
PCOS

Neurology

Huntington's
Migraines
Dementia

Constitutional

Weight
Malignant
Hyperthermia

Endocrinology

DM
Multiple endocrine
neoplasia
Von Hippel-Lindau

Ortho/Rheum.

Connective
tissue disorders
Clubfoot
RA, Lupus

Others?

Genetics and Family History

- We've done genetic assessments before we knew there were genes
- Family history assessment remains inexpensive, extremely accessible tool
- Optimizes health outcomes
- Required for certain situations
- May save a life!



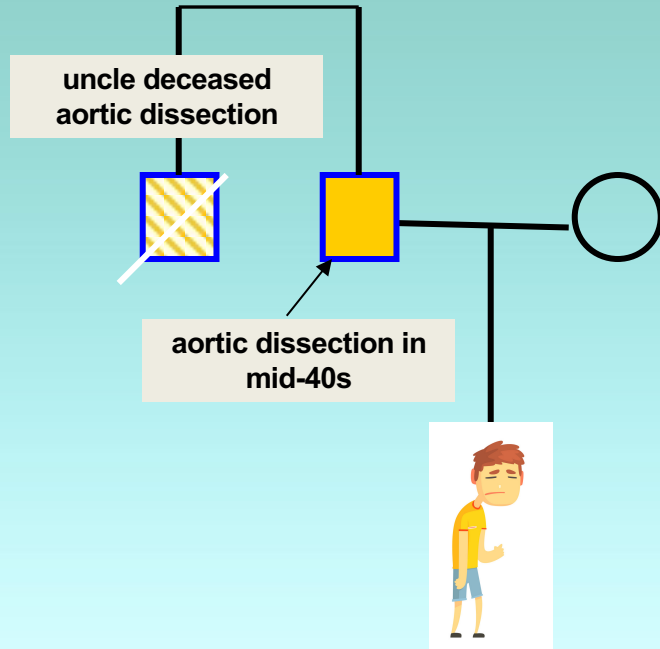
What would you do?

19yo M presents with chest pain



- Non-exertional chest pain. Recent URI with coughing, now resolved, not COVID. What's your workup?
- EKG: Normal. Troponin: Negative. Dimer: Negative.
- Next steps:
 - A. Treat with NSAIDs and discharge
 - B. Refer to a cardiologist
 - C. Further testing. If so, what?
 - D. Keep for observation
 - E. Other

With this information, what would you do?



So now, what would you do?

- A. Treat with NSAIDs and discharge
- B. Refer to a cardiologist
- C. Further testing. If so, what?
- D. Keep for observation
- E. Other

19 yo male with chest pain

- "We informed them that there was a history of aortic dissection in his family," said Tyler's mother, Debra.
 - No advanced testing or referrals were done
- Nine days after he first experienced chest pains, Tyler collapsed and died.
- 20% of aortic dissections have genetic/familial link

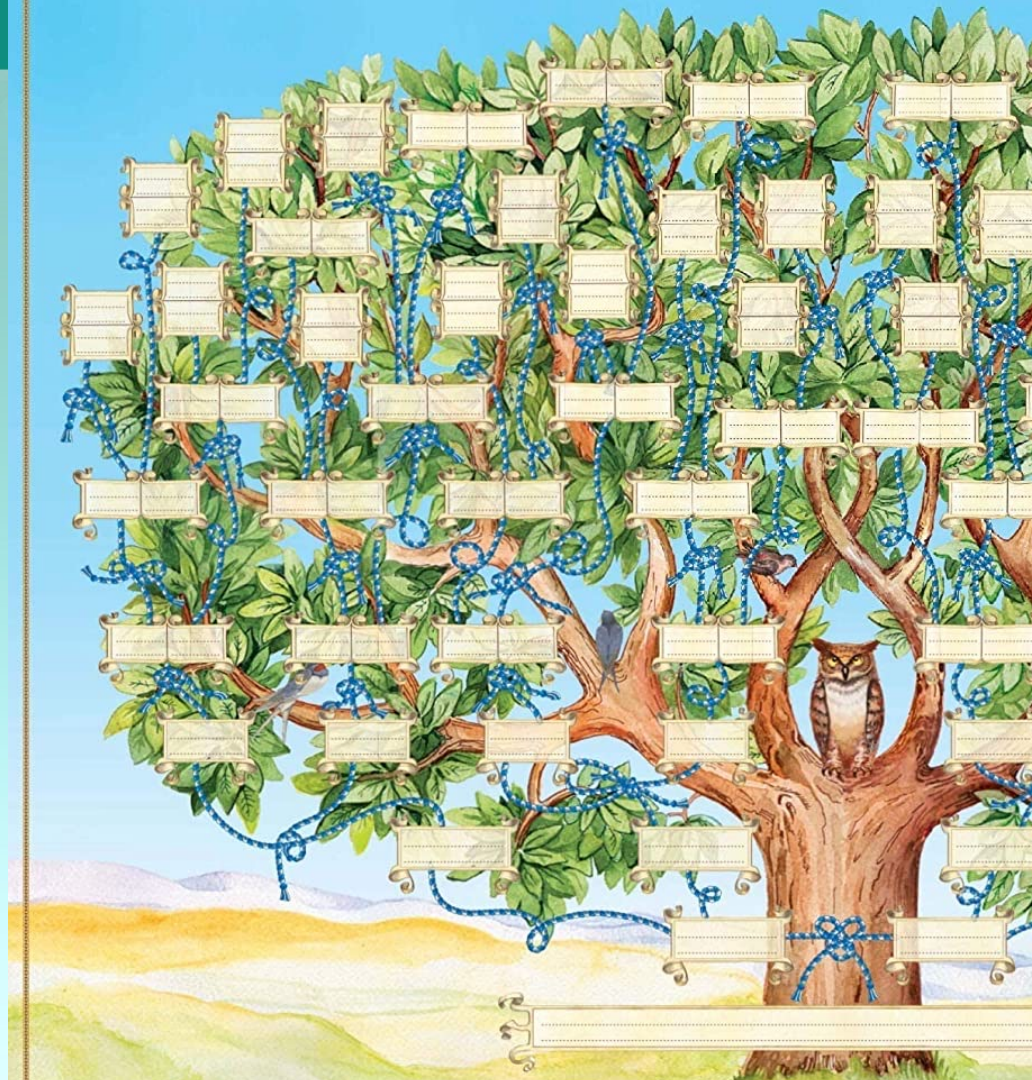


In memory Tyler Kahle

<https://bestcare.org/aortic-dissection-at-any-age-the-tyler-kahle-story>

<https://www.wsj.com/articles/SB106789437888762200>

Creating the Genetic Family Tree




Genetic Role in Healthcare

The background features a teal and light blue color scheme. A prominent graphic is a DNA double helix structure, with one strand in a lighter teal and the other in a darker teal. To the right of the DNA, there is a stylized bar chart with several vertical bars of varying heights. The overall aesthetic is clean and professional, typical of a medical or scientific presentation.

- PAs can act as “genetic gatekeepers”
- Not just in primary care – all specialties
 - Screening for inherited diseases
 - Assessing familial/genetic risk for prevention and early identification
 - Recommending genetic testing
 - Making referrals to genetic service providers

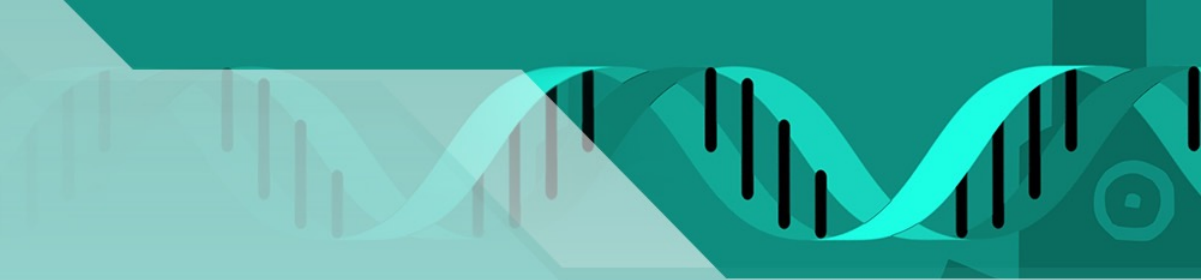
A Typical Family History Notation



- “Brother has asthma, heart disease and diabetes. Mother has lung cancer. Grandmother died of some type of cancer.”
- Thoughts?

Maybe a little more detail would help

- “Father had thromboangiitis obliterans and suffered amputations. Mother had diabetes and hypertension as did maternal grandmother. Both have succumbed to strokes. Arthritis of some type is noted in mother and an aunt.”
- Thoughts?



WHY IS FAMILY HISTORY IMPORTANT?

Family history is a risk factor for diseases throughout all stages of life

Birth defects
Blood disorders



Diabetes
Depression



Alzheimer's disease
Osteoporosis



Pre-conception

Infants

Children

Adolescents

Adults

Older adults

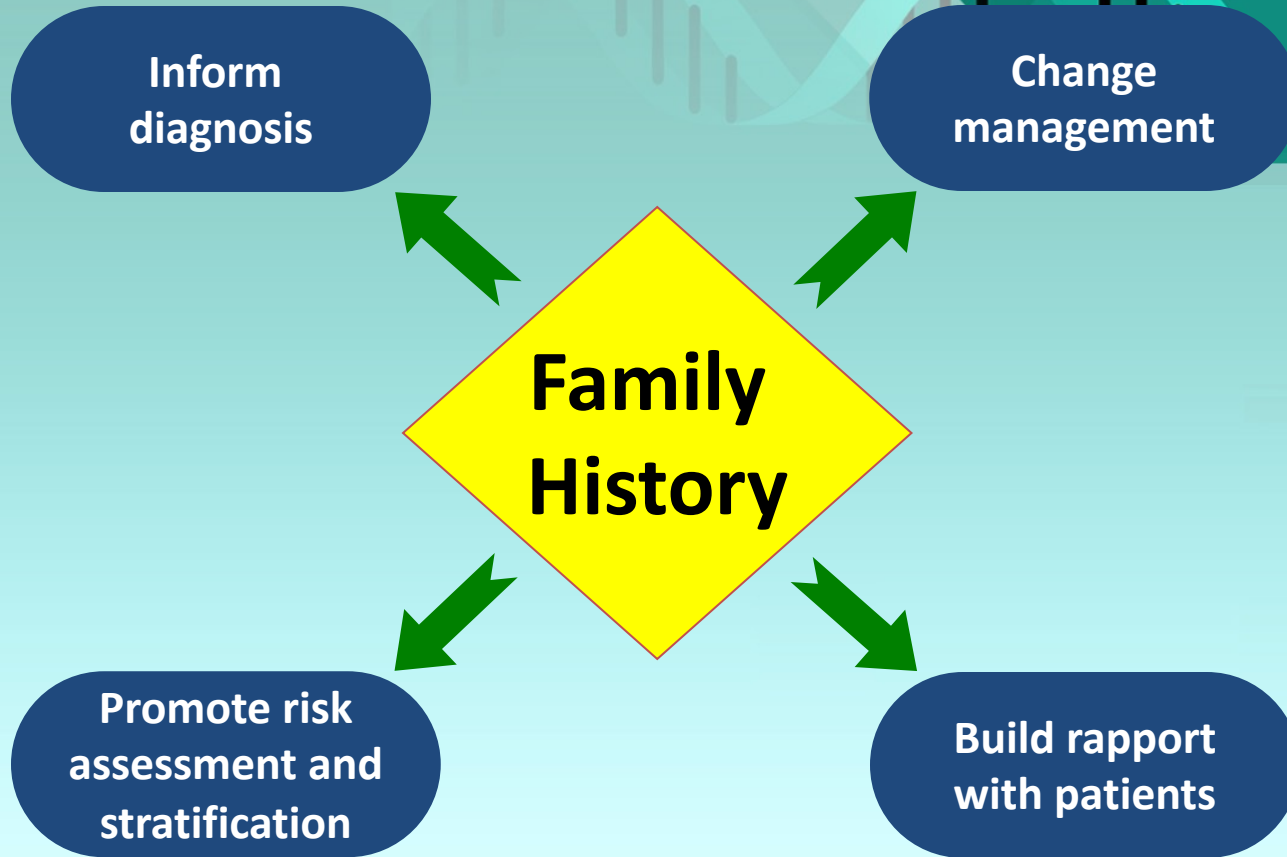


Asthma
Autism

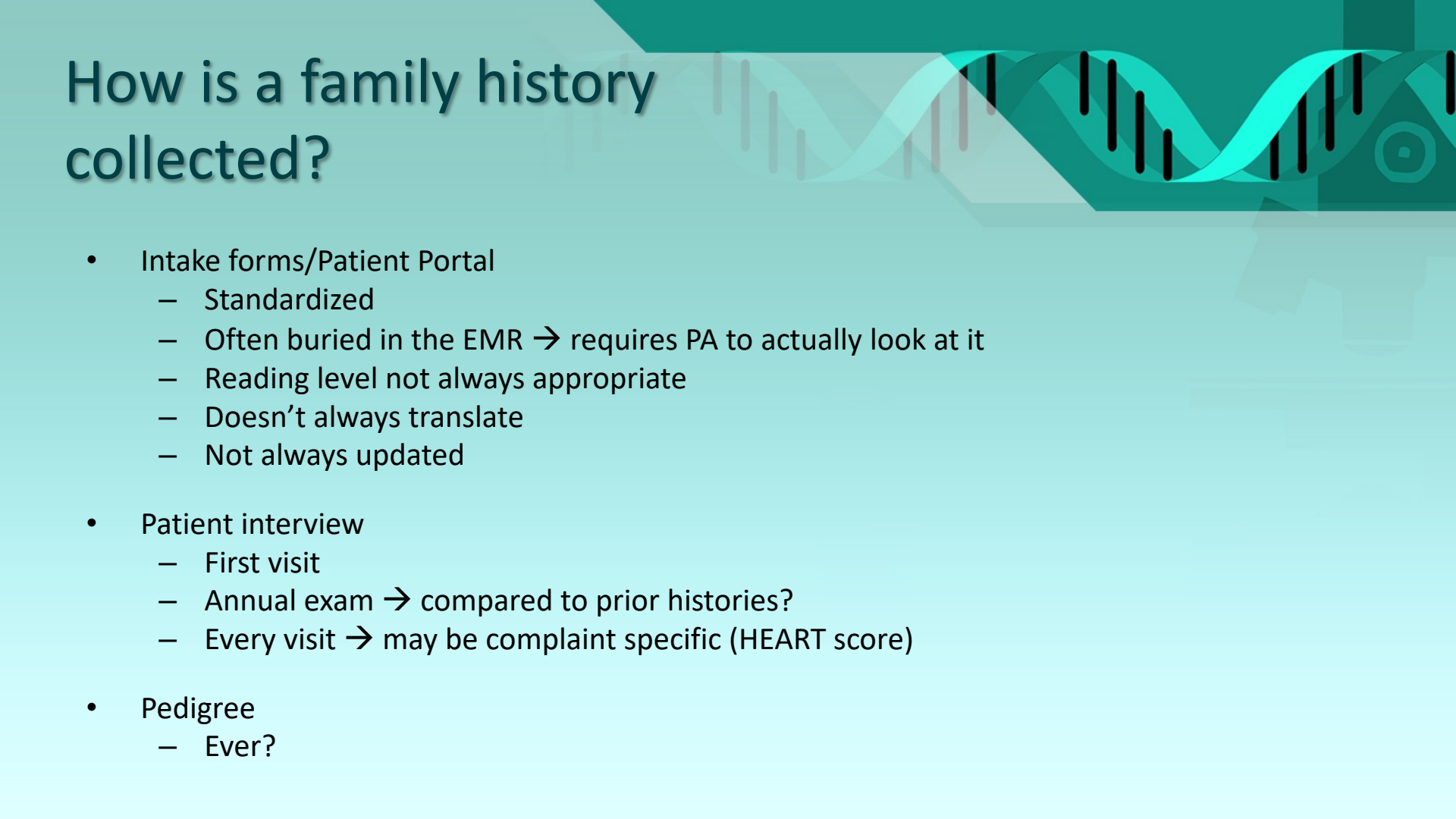
Cancer
Heart disease

Miscarriage
Maternal death
Congenital disease





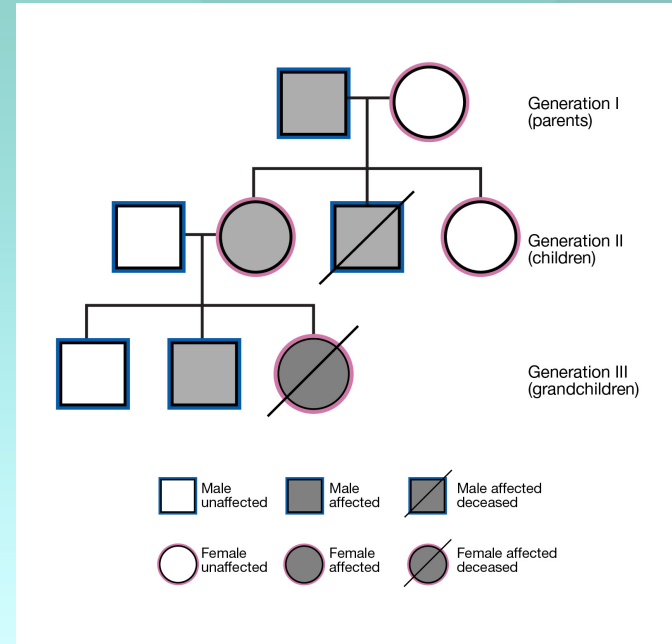
How is a family history collected?



- Intake forms/Patient Portal
 - Standardized
 - Often buried in the EMR → requires PA to actually look at it
 - Reading level not always appropriate
 - Doesn't always translate
 - Not always updated
- Patient interview
 - First visit
 - Annual exam → compared to prior histories?
 - Every visit → may be complaint specific (HEART score)
- Pedigree
 - Ever?

What is a pedigree?

- A standardized method of drawing a family history
- Relationships of family members are represented schematically



Advantages of a Pedigree



- Can be viewed at a glance
- Easier to interpret graphically
- Easily annotated and updated
- May demonstrate patterns of transmission of familial disorders
 - Helps to clarify or possibly identify risk of disease
 - Helps ascertain screening, diagnostic, surveillance, and/or treatment options
 - May distinguish genetic from other risk factors

Other Benefits

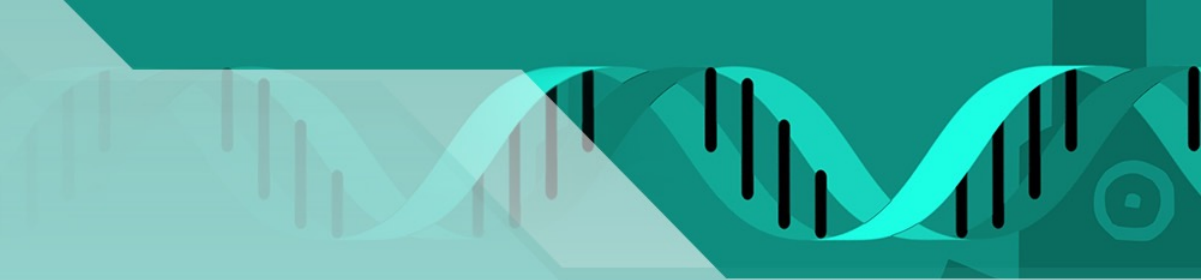
The background features a teal and light blue color scheme. A prominent graphic is a DNA double helix structure, with one strand in a lighter teal and the other in a darker teal. To the right of the DNA, there is a stylized bar chart with several vertical bars of varying heights. The overall aesthetic is clean and scientific.

- Helps to establish rapport with patients
- Family relationships graphically emerge--could alert you to sensitive issues and family dynamics
- Shows shared environmental or lifestyle factors that can interact with genetic risk
- Family education (esp. prevention)
- Will alert you to families that may require assistance (e.g., support groups, resources, referrals)

Barriers



- Time (and reimbursement)
- Underestimation of its value
 - What people know or understand about their families isn't always accurate
 - Medical literacy
 - Bias/distrust of diagnoses
 - Genetic discrimination in disability or life insurance?
 - GINA* protects for health insurance
 - Confidentiality issues
- Not always incorporated into EMRs
 - Scanned document
 - Written list



BREAKING DOWN BARRIERS

Barrier removal strategies



- Gather history over several visits
- Interactive software for patients to record their family history at home before an appointment*
- EMR updates
- Stress the importance to staff/colleagues
- FHx can increase complexity of MDM
 - Can bill for higher level

* https://www.cdc.gov/genomics/famhistory/knowning_not_enough.htm

Practicalities

- Family histories are dynamic and require updating
- Use family history questionnaires for expediency
- Interpretation of the pedigree requires some experience and expertise—when in doubt, refer, refer, refer!



Approach to taking a pedigree





















The background of the slide features a teal and light blue color scheme. On the right side, there is a stylized DNA double helix structure in shades of teal and black. Below it, a faint, light-colored silhouette of a microscope is visible, suggesting a medical or scientific context.




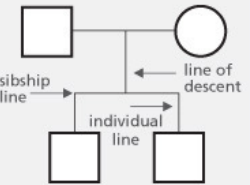
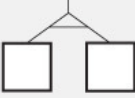

- Explain to patient
 - Why it's important
 - How you plan to use it
 - The confidential nature of collecting and documenting it

Approach (cont)

- Start with the patient (consultand, proband)
- Asking about the health of 3-generations pedigree is obtained:
 - Patient
 - Siblings
 - Parents
 - +/- Children (depending on age)
 - +/- Grandparents (depending on age)

How to graph the pedigree

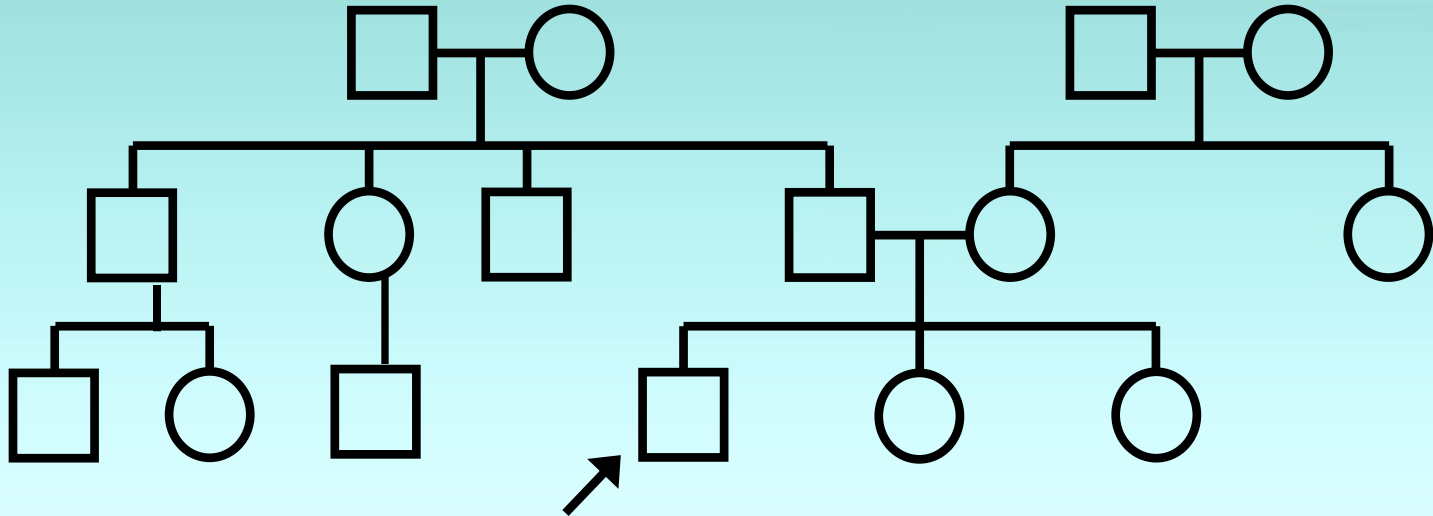
	Male	Female	Sex Unknown
Individual			
Affected individual (symbol coloured in)			
Multiple individuals			
Deceased			
Pregnancy			
Miscarriage	 male	 female	
Person providing pedigree information			

Marriage/partnership	
Divorce/separation	
Where the partners are blood relatives (consanguineous relationship)	
Children/siblings	
Identical twins (monozygotic)	
Non-identical twins (dizygotic)	

See online resources for additional symbols <https://www.genomicseducation.hee.nhs.uk/taking-and-drawing-a-family-history/>
<https://www.aafp.org/afp/2005/0801/p441.html>

To start

- Identify the person providing the history with an arrow



Collection

The background features a stylized DNA double helix in shades of teal and light blue. To the right, there is a bar chart with vertical bars of varying heights, also in teal and light blue. The overall aesthetic is clean and scientific.

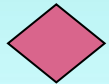
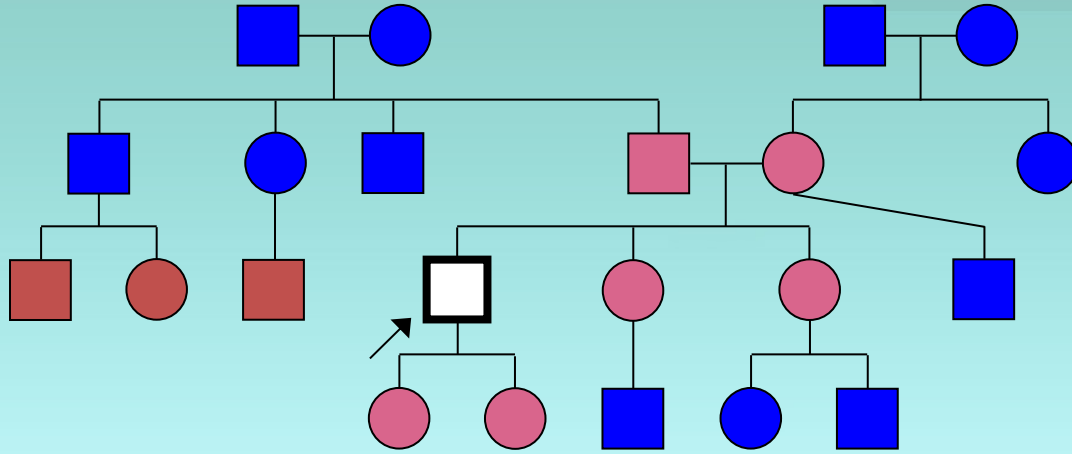
- Also indicate:
 - Date of collection (or date of update), and name of collector (or updater)
 - Legend or key, if symbols are used to designate disease

Non-directed vs Targeted Pedigree

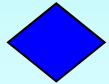
The background features a teal and light blue color scheme. A prominent graphic is a DNA double helix structure, with one strand in a lighter teal and the other in a darker teal. To the right, there is a faint, stylized silhouette of a human figure, possibly representing a patient or a doctor. The overall aesthetic is clean and medical.

- Non-directed
 - not related specifically to an HPI or other complaint, but collected for a new patient encounter or during annual exam
- Targeted
 - very specific to the complaint or more immediate need of a patient

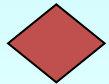
Degrees of relationship



First-degree relatives: parents, siblings, children

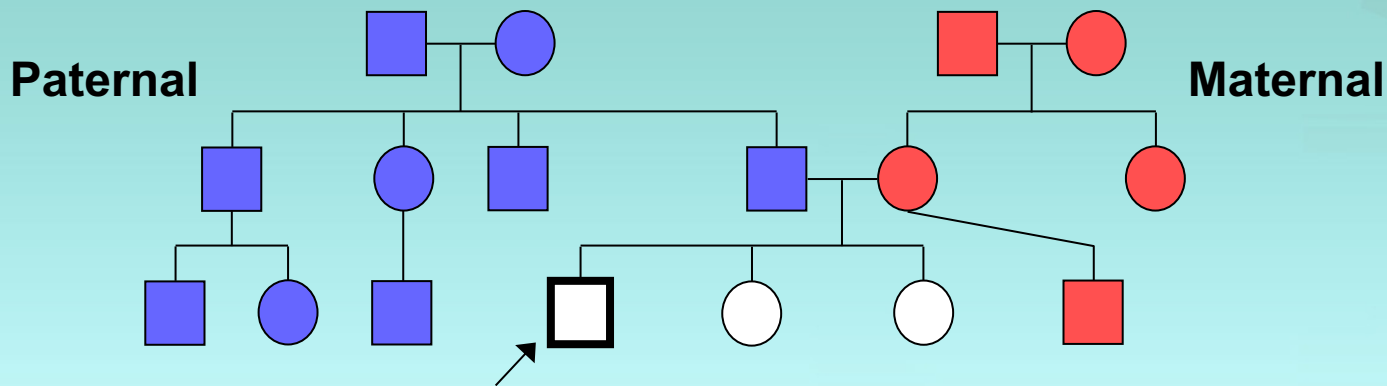


Second-degree relatives: half-siblings, aunts, uncles, grandparents, nieces & nephews



Third-degree relatives: first cousins

Maternal and paternal relatives



Issues to be careful about

The background features a teal and light blue color scheme. On the right side, there is a stylized DNA double helix with black vertical bars of varying heights extending from it, resembling a bar chart or a genomic map. The overall aesthetic is clean and scientific.

- Don't make assumptions
 - Children don't always have the same mother and/or father
 - Miscarriages may not be discussed if not asked about explicitly
 - Infertility vs. no children by choice
 - Adoptions (into and out of family)
 - Consanguinity

Pay attention to



- Ethnicity
 - Document ethnicity as it may confer increased risk for certain conditions (e.g., Tay Sachs disease, cystic fibrosis)
 - Be aware of your own bias, e.g. sickle cell “only” affects individuals who are Black
- Cultural awareness
 - May feel it is disrespectful to talk about deceased relatives, especially for conditions with more stigma (e.g. mental health)
 - Idea of confidentiality

<https://store.samhsa.gov/sites/default/files/d7/priv/sma16-4931.pdf>
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6396634/>

What and how to inquire

The background features a teal and light blue color scheme. A prominent graphic is a DNA double helix structure, with one strand in a lighter teal and the other in a darker teal. To the right of the DNA, there is a bar chart with several vertical bars of varying heights. The overall aesthetic is clean and scientific.

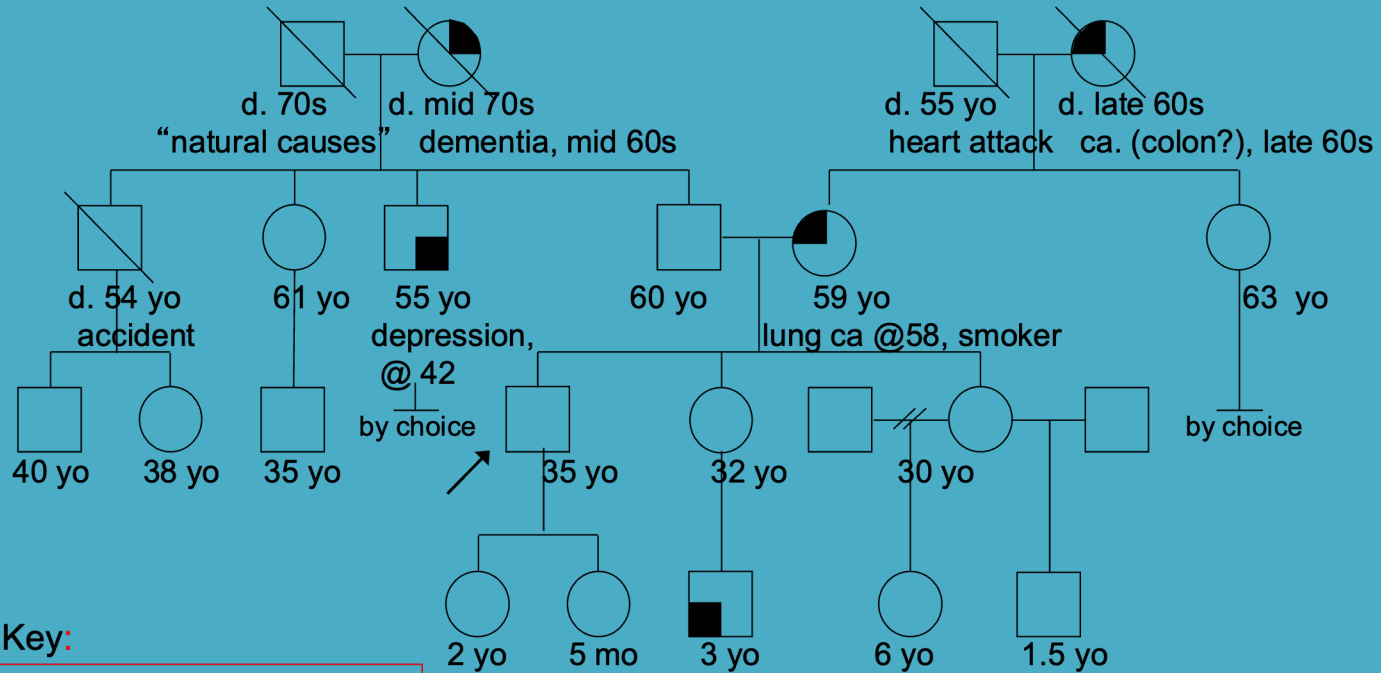
- Depends on patient (proband)
 - e.g., prenatal vs pediatric vs adult
- Questionnaires exist for different patients
 - Good starting point
 - Inflexibility of structure decreases thoroughness
- Make sure to start with condition(s) that patient is most concerned about

Example

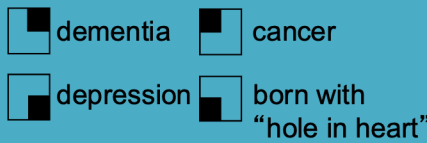
no consanguinity reported

N. European

German, English, Native American



Key:

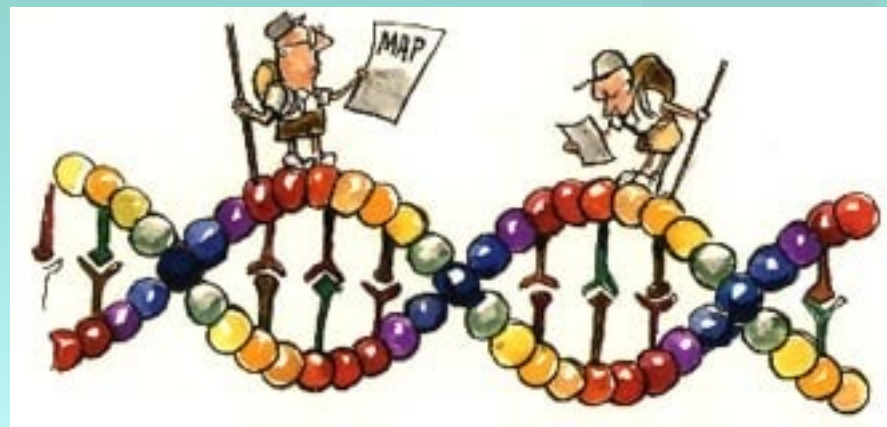


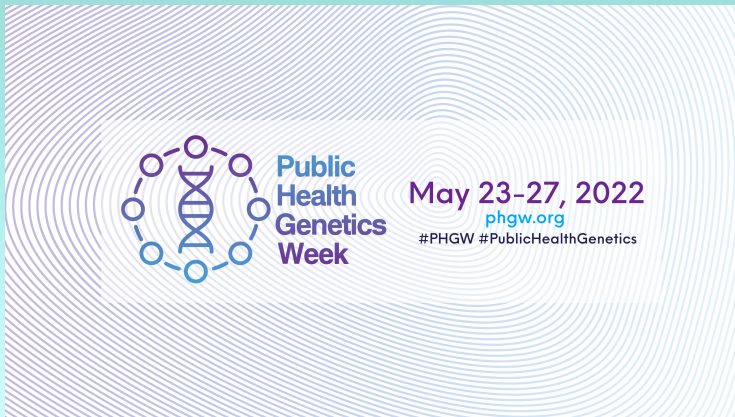
Collected by: Jane Doe
 Collected on: August 20, 2021

Implementation

The background features a teal and light blue color scheme. A prominent graphic is a DNA double helix structure, with one strand in a lighter teal and the other in a darker teal. To the right of the DNA, there is a stylized bar chart with several vertical bars of varying heights. The overall aesthetic is clean and scientific.

- Take a pedigree now (e.g., practice doing one on your own family or a friend)
- Pedigrees, red flags, and interpretation during return visits





Panel Discussion

Consider joining the Society of PAs in Genetics and Genomics
<https://spagg.wildapricot.org/>