Taking a Family History

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Learning objectives

• Understand why family histories are useful
• Recognize and recall proper nomenclature used to take a family history
• Create a complete family history
• Recognize inheritance pattern in a family history
Where have you encountered family histories in your life?

- Personal genealogy research
- Medical appointments
- Religious genealogy
How are family histories useful in clinical care settings?

- Quick and accurate visual record of a family
- Put the patient in charge and build rapport
- Suggest an inheritance pattern and identify at-risk family members
- Aid in making a diagnosis
- Reveal information about familial relations
Terminology

• **Pedigree**: family history drawn in picture format using standard symbols

• **Proband**: affected family member who brought the family to medical attention

• **Consultand**: person seeking genetic evaluation

Sometimes consultand and proband are the same person!
Basic symbols

- Female
- Male
- Other or unspecified
- AMAB (Male sex assigned at birth, female gender)
- AFAB (Female sex assigned at birth, male gender)
- AFAB (Non-binary person, female sex at birth)
Basic symbols

- 36
- 4 mo
- 5 wk
Basic symbols

N 5 P

13 weeks

Abortion
Spontaneous Abortion
Ectopic pregnancy
Product of conception
Basic symbols

Key

Colon cancer

Thyroid cancer

Prostate cancer

Sickle cell trait

Intellectual disability
Basic symbols

Proband and/or consultand
Basic symbols

d. 82
COD: Heart attack

Death
Quick quiz!

- What does this symbol represent?

5 wk
Quick quiz!

• What does this symbol represent?

5 week old female
Quick quiz!

• Draw the symbol for a 52-year-old man diagnosed with hypothyroidism and atrial fibrillation.
Quick quiz!

• Draw the symbol for a miscarriage that occurred at 13 weeks.
Quick quiz!

• Draw the symbol for a miscarriage that occurred at 13 weeks.

13 wk
Basic Pedigree

- Paternal grandfather
- Paternal grandmother
- Aunt
- Father
- Sister
- Child
- Mother
- Brother
- Uncle
- Maternal grandfather
- Maternal grandmother
- First cousin
Line of descent

Relationship line

Identical twins

Sibship line

Line of descent

Infertility

No children by choice
Half siblings

- Paternal half sister and half brother
- Maternal half brother
Elizabeth Taylor’s pedigree

How many biological children did Elizabeth Taylor have?

Figure 3.6 A pedigree of actress Elizabeth Taylor demonstrating how to illustrate multiple marriage partners, stepchildren, and half siblings. (Source: www.celebsite.com)
Elizabeth Taylor’s pedigree

How many biological children did Elizabeth Taylor have?

4 children and one miscarriage
Adoption

Adopted in

Adopted out
Quick quiz!
Identify relationship of A, B, C, D, and E to the proband
Quick quiz!

A: Full sister
B: Half brother
C: Uncle
D: Cousin
E: Grandmother
Quick quiz!

• How would you draw two parents who recently adopted a 5-year-old girl?
Quick quiz!

• How would you draw two parents who recently adopted a 5-year-old girl?
Quick quiz!

- How would you draw a couple who has not been able to conceive due to fertility issues?
Quick quiz!

• How would you draw a couple who has not been able to conceive due to fertility issues?
Taking a family history

• Provide brief explanation of the purpose and process
• Indicate what information you are interested in collecting
• Use empathy in response to traumatic experiences
Taking a family history

• Chronological order
  – Consultand
  – Children or pregnancies
  – Siblings
  – Parents
  – Mother’s side
  – Father’s side
Taking a family history

• Make sure to record family member’s
  – Age
  – Major health issues including age of diagnosis or symptom onset
  – Genetic diagnosis or test results
  – Targeted medical history questions
Pedigree questions

• Ancestry specific when concerned about relative risk
  – Example: Ashkenazi Jewish ancestry
  – Is there any Ashkenazi Jewish ancestry in your family?

• Consanguinity
  – Is there any possibility your parents (or you and your spouse) are related by blood?
Consanguinity denied

Ashkenazi Jewish ancestry
Tools for creating a pedigree

• Electronic genetic pedigree software

• Pedigree stencils

• My Family Health Portrait
Review of Inheritance Patterns

- Autosomal dominant
- Autosomal recessive
- X-linked recessive
Autosomal dominant

• Only need one copy of a mutation to inherit a condition

• Pedigree clues
  – There is an affected family member in every generation
  – An affected individual will have an affected parent
Autosomal recessive

• Need 2 copies of a mutation to inherit a condition

• Pedigree clues
  – 2 unaffected parents with an affected child
  – Condition may skip a generation
  – More likely to see affected individuals in consanguineous families
Autosomal Recessive

Parents

Children

Affected
Carrier
Carrier
Unaffected
X-linked recessive

• Females have two X chromosomes while males only have one
• Pedigrees clues
  – More likely to see affected males than affected females
  – Affected males cannot have affected sons
  – Condition may skip a generation
X-Linked Recessive

Parents

Father Affected  Mother Unaffected

Children

Son Unaffected  Daughter Carrier  Son Unaffected  Daughter Carrier

NIH  U.S. National Library of Medicine
Congenital Hearing Loss

- All newborns are screened for hearing loss at birth hospital
- 1-2 in every 1,000 babies with a detectable level of hearing loss
- 50-60% of newborn/congenital hearing loss believed to be genetic
Congenital Hearing Loss

Most is Autosomal Recessive

- □ = carrier
- Ⓞ = affected with hearing loss
Family history of Queen Victoria

- Queen Victoria reigned England from 1837-1901
- Hemophilia
Hemophilia

X-linked recessive

Prince Albert

Queen Victoria

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3
Time to draw your own!

- Practice drawing your 3 generation pedigree.
- Make sure to ask yourself stepwise questions to lead you through the process (ex: how many kids do you have, how old are they, etc).
- After ~10 minutes we will come back together to discuss.
References


• https://www.scientificamerican.com/article/born-to-the-purple-the-st/

• https://medlineplus.gov/genetics/understanding/inheritance/inheritancepatterns/
My Family Health Portrait