Recruiting subjects for genetic studies: What study teams need to know

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June 2, 2025

1 pm – 2 pm

Outline

- Genetics 101
- Pedigree Drawing
- Informed Consent
- Typical subject recruitment questions
- NJACTS GREAT Core Consultation

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Genetics 101

- **Genetics vs. Genomics:** Genetics is the study of individual genes, whereas genomics is the study of the entire genome, or all of an organism's genes, interactions among genes, and the environment's role in affecting them

Genetics and Genomics 101

- **Why should we study genetics and genomics?**
  - Genetic makeup is associated with risk for health and disease (*who becomes sick or stays healthy?*)
  - Genetic makeup can also make us more or less responsive to medical treatments (*how do we treat patients?)*
    - The case of redheads and anesthetics (*MC1R* gene)

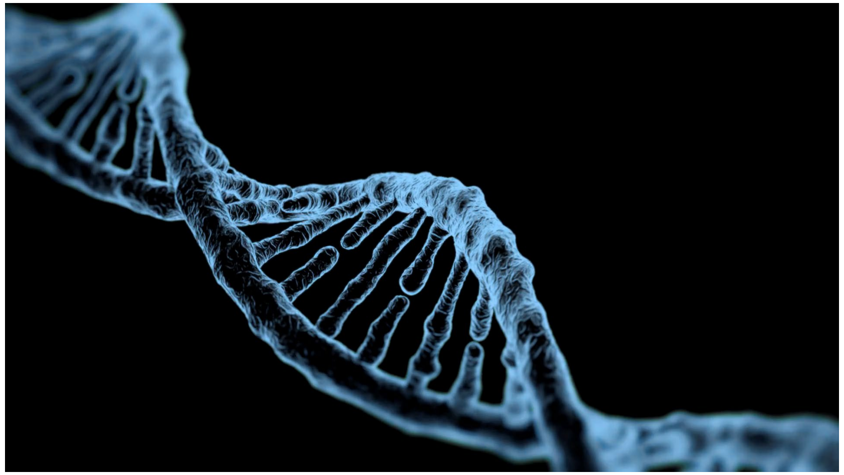
Genetics and Genomics 101

- **How do we study genetics and genomics?**

Latent/inferred genetic design

- Twins
- Adoption
- Family history

Molecular genetic designs



- Genome-wide genotyping and sequencing

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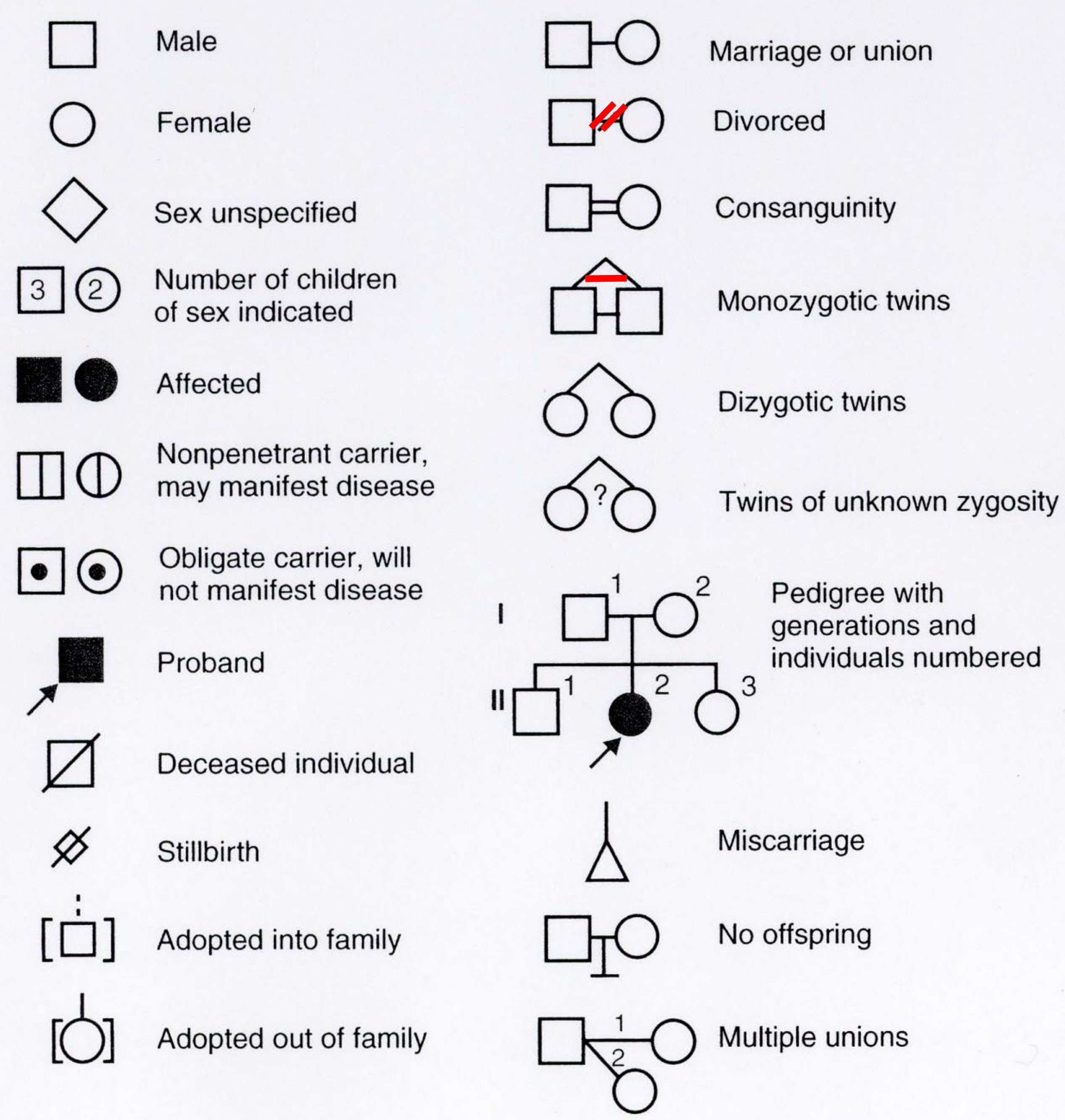
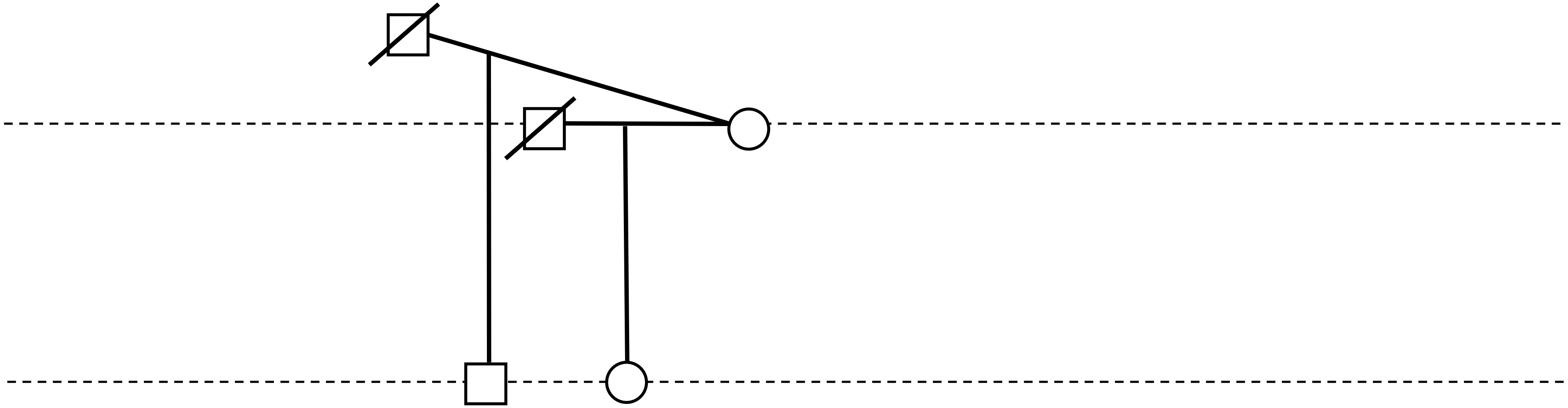
Pedigree Drawing

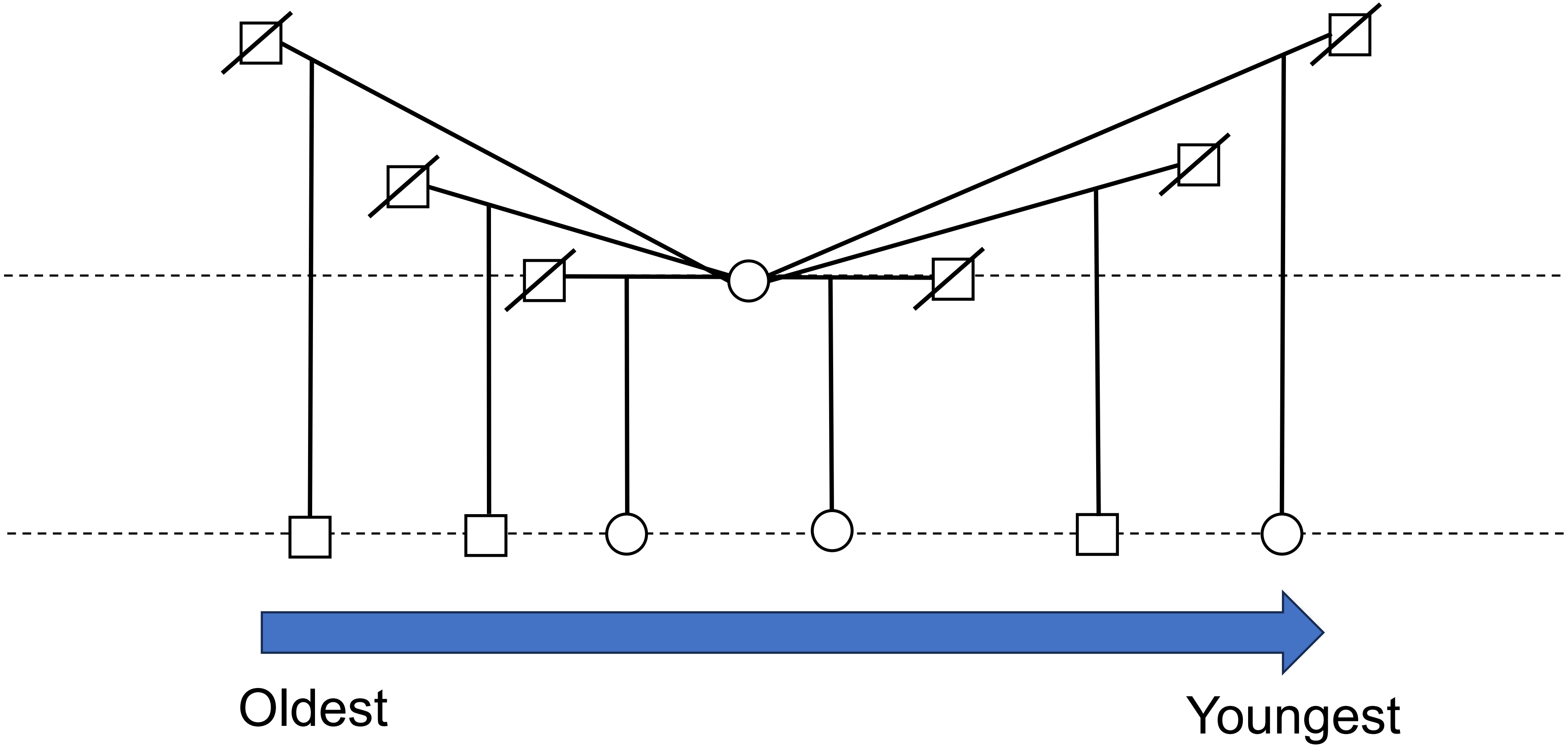
- Pedigree nomenclature
- Understanding family relationships
- How to ask the questions


Male
Female
Sex unspecified
3
2
Number of children of sex indicated
Affected
Nonpenetrant carrier, may manifest disease
Obligate carrier, will not manifest disease
Proband
Deceased individual
Stillbirth
Adopted into family
Adopted out of family
Marriage or union
Divorced
Consanguinity
Monozygotic twins
Dizygotic twins
?
Twins of unknown zygosity
I
II
1
2
1
2
3
Pedigree with generations and individuals numbered
Miscarriage
No offspring
1
2
Multiple unions

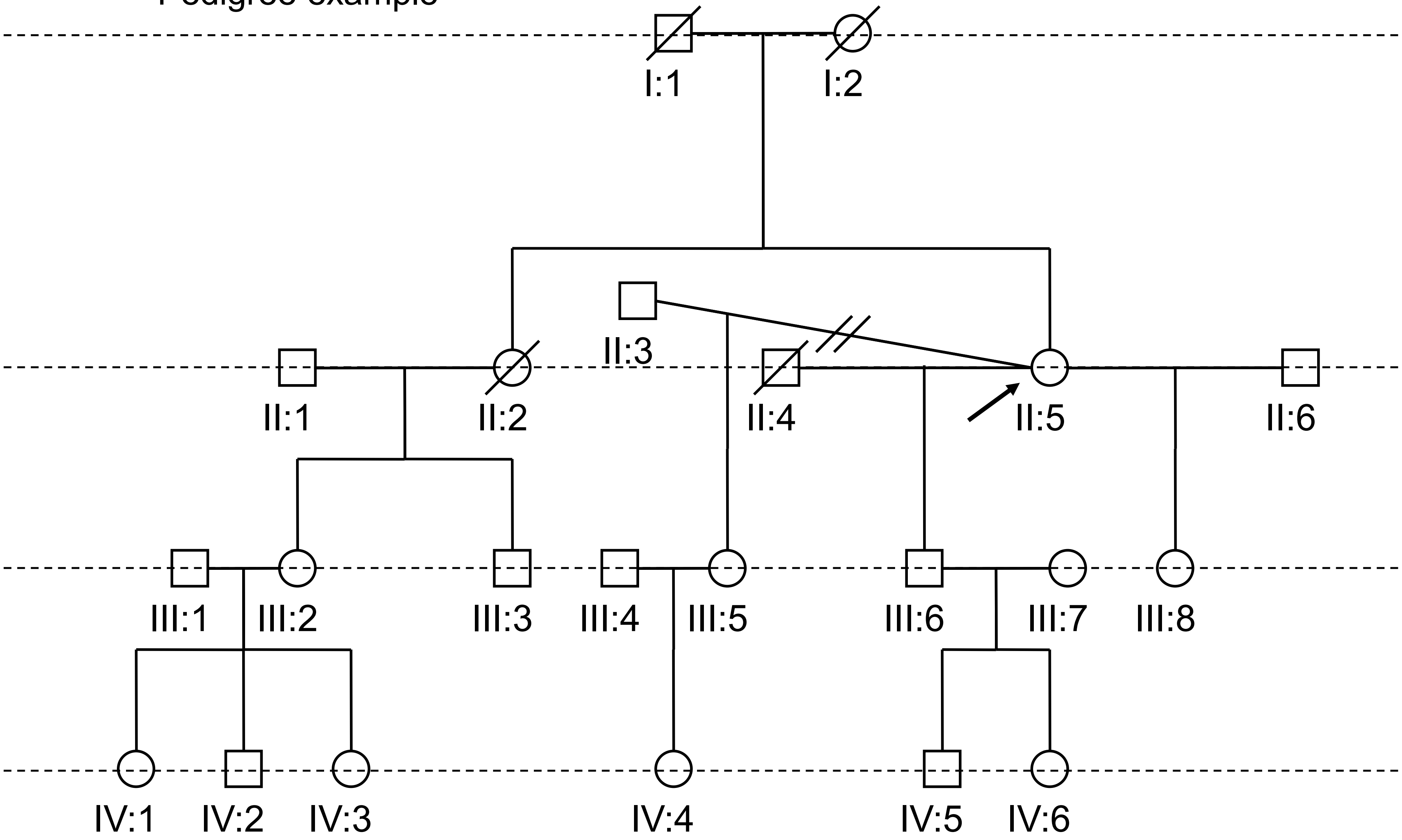
How to draw multiple spouses



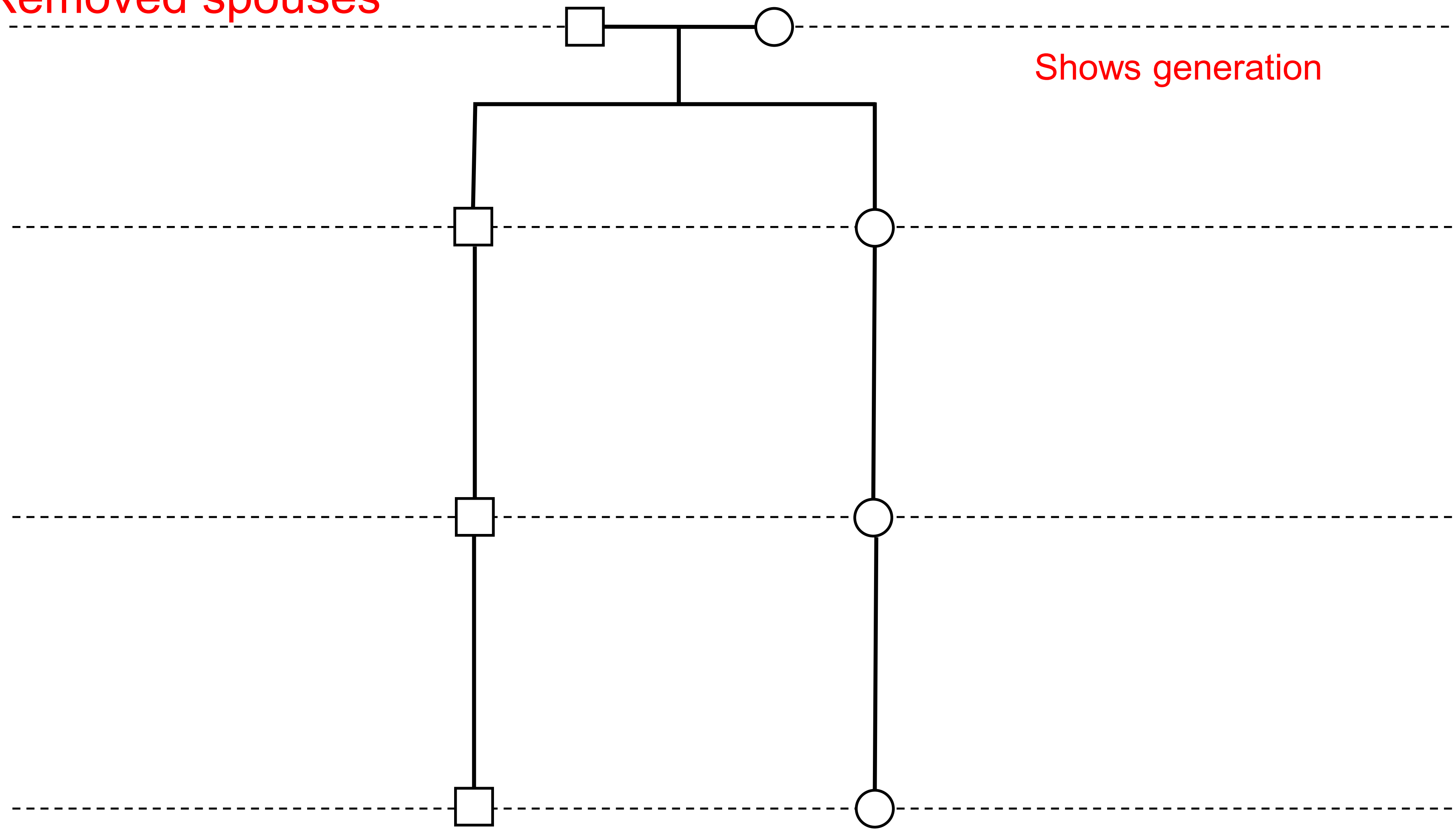
How to draw multiple spouses



Pedigree example

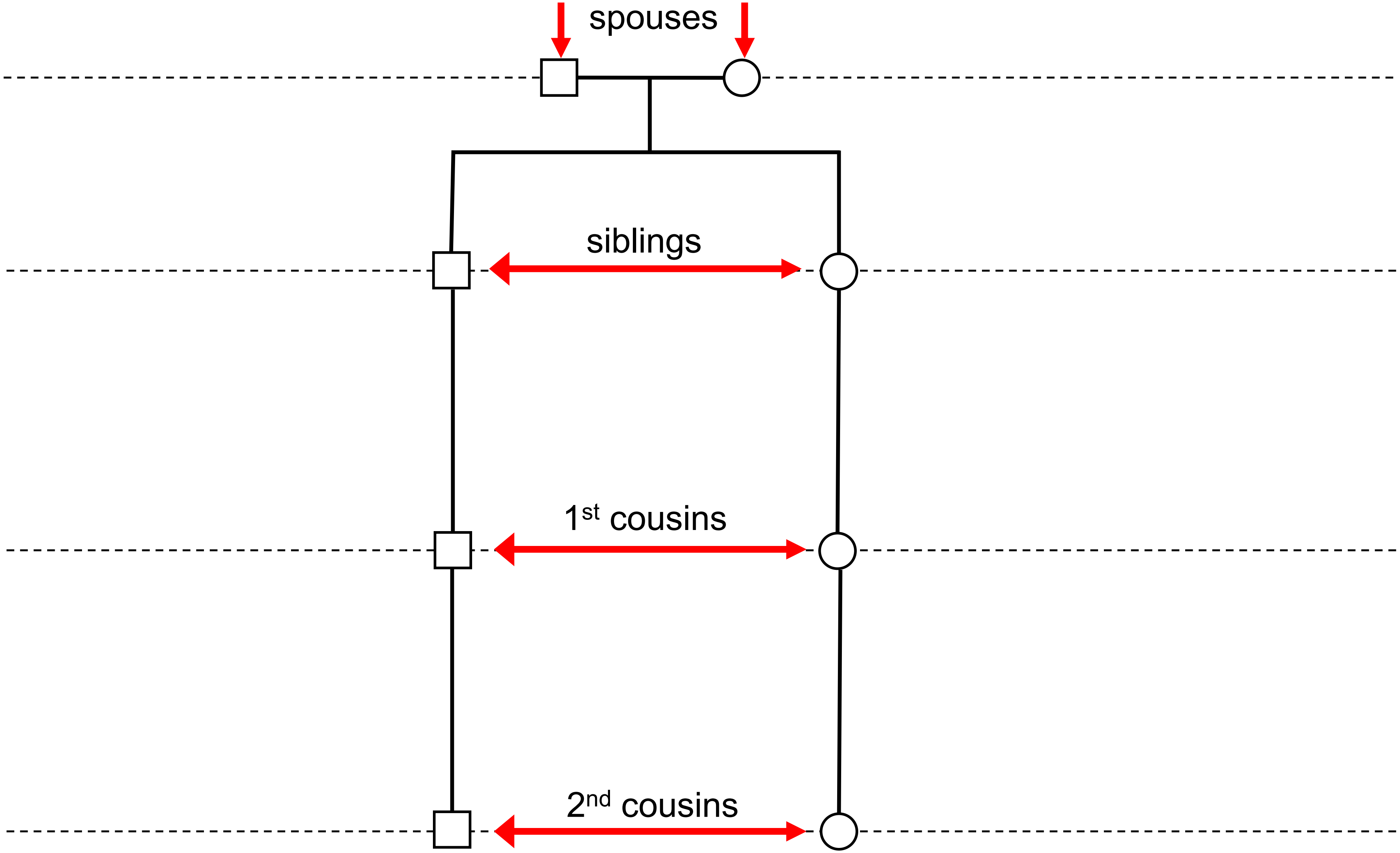


Removed spouses



Notice dashed lines

Shows generation

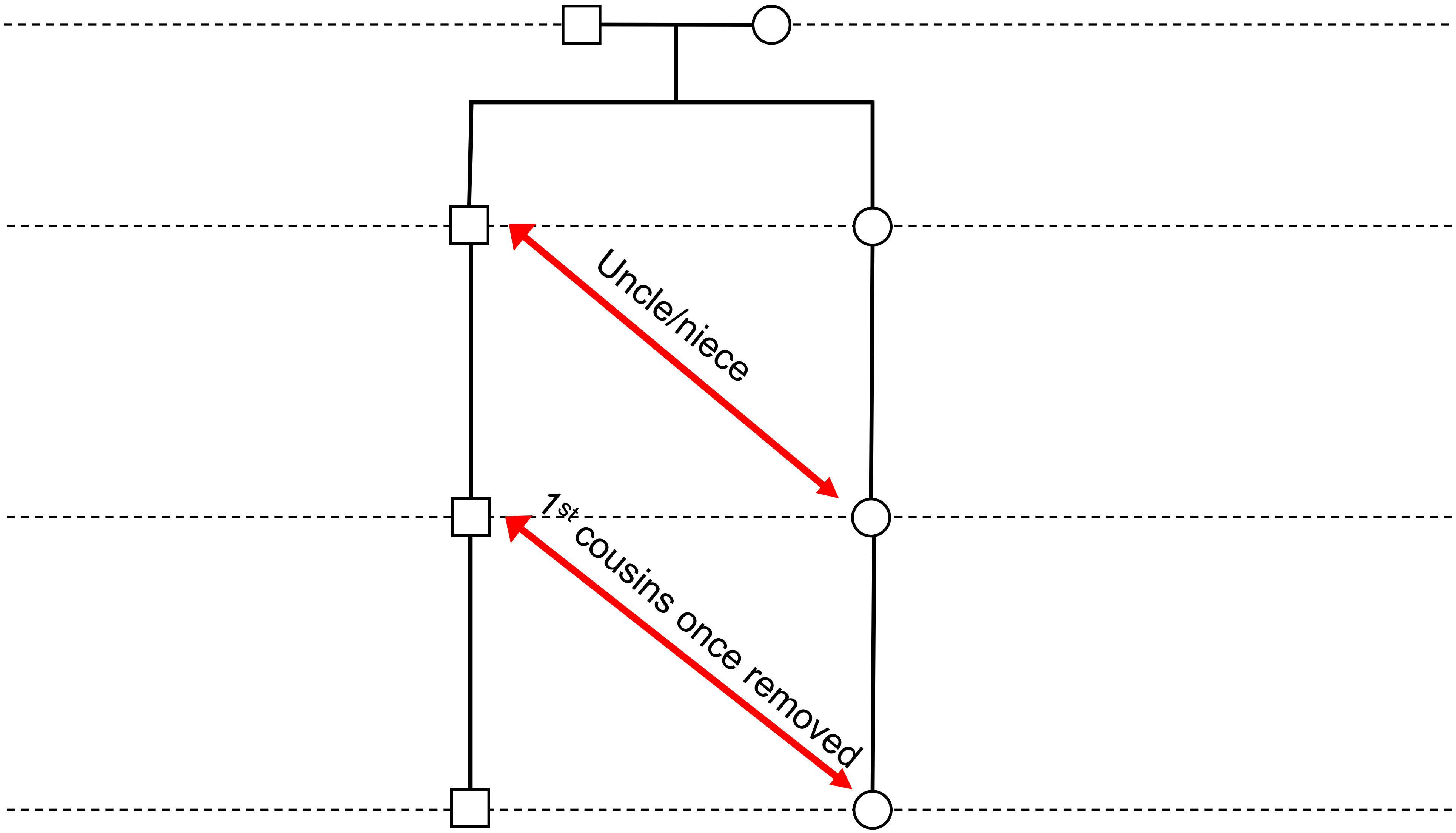


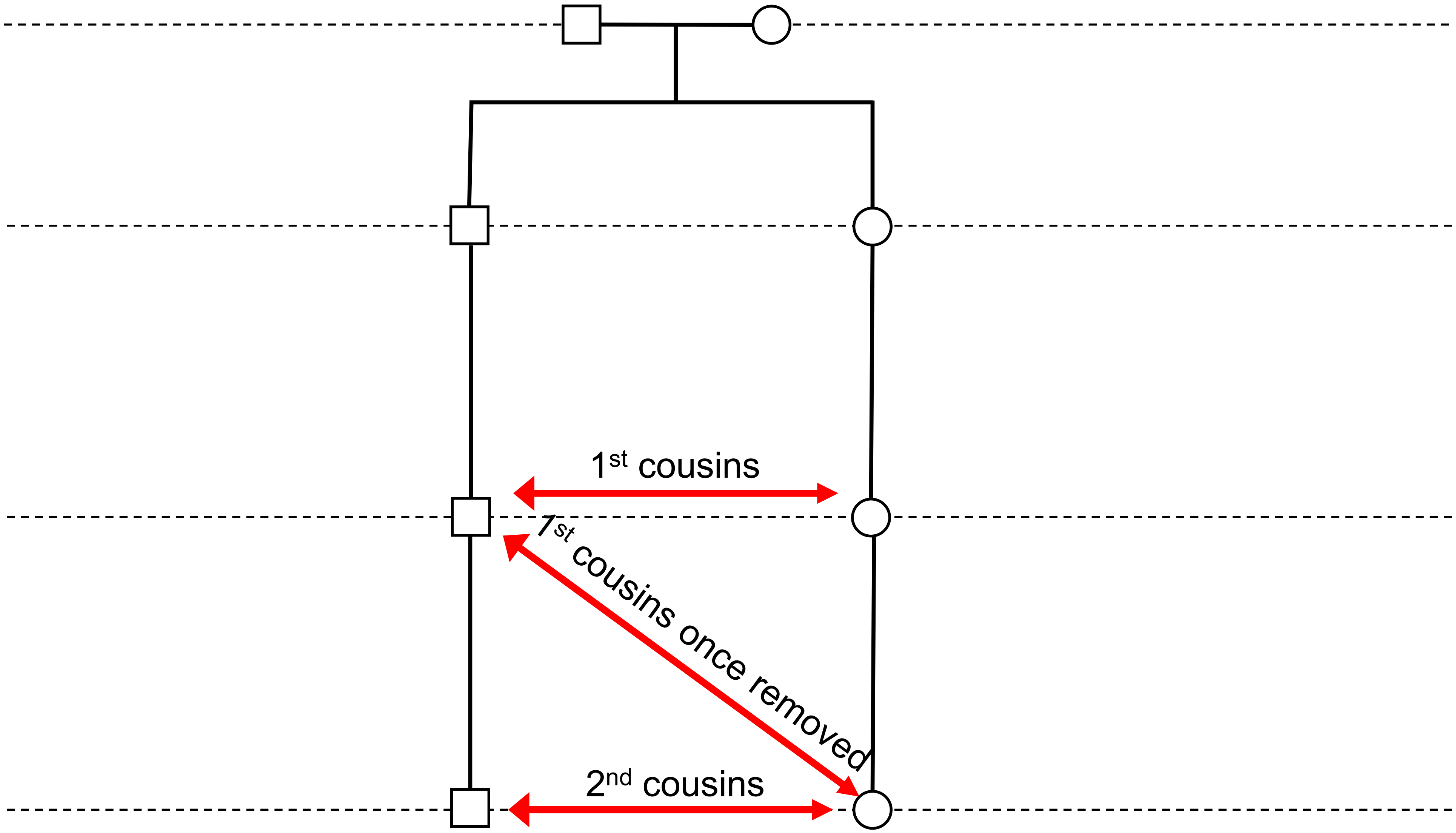
spouses

siblings

1st cousins

2nd cousins


Uncle/niece
1st cousins once removed

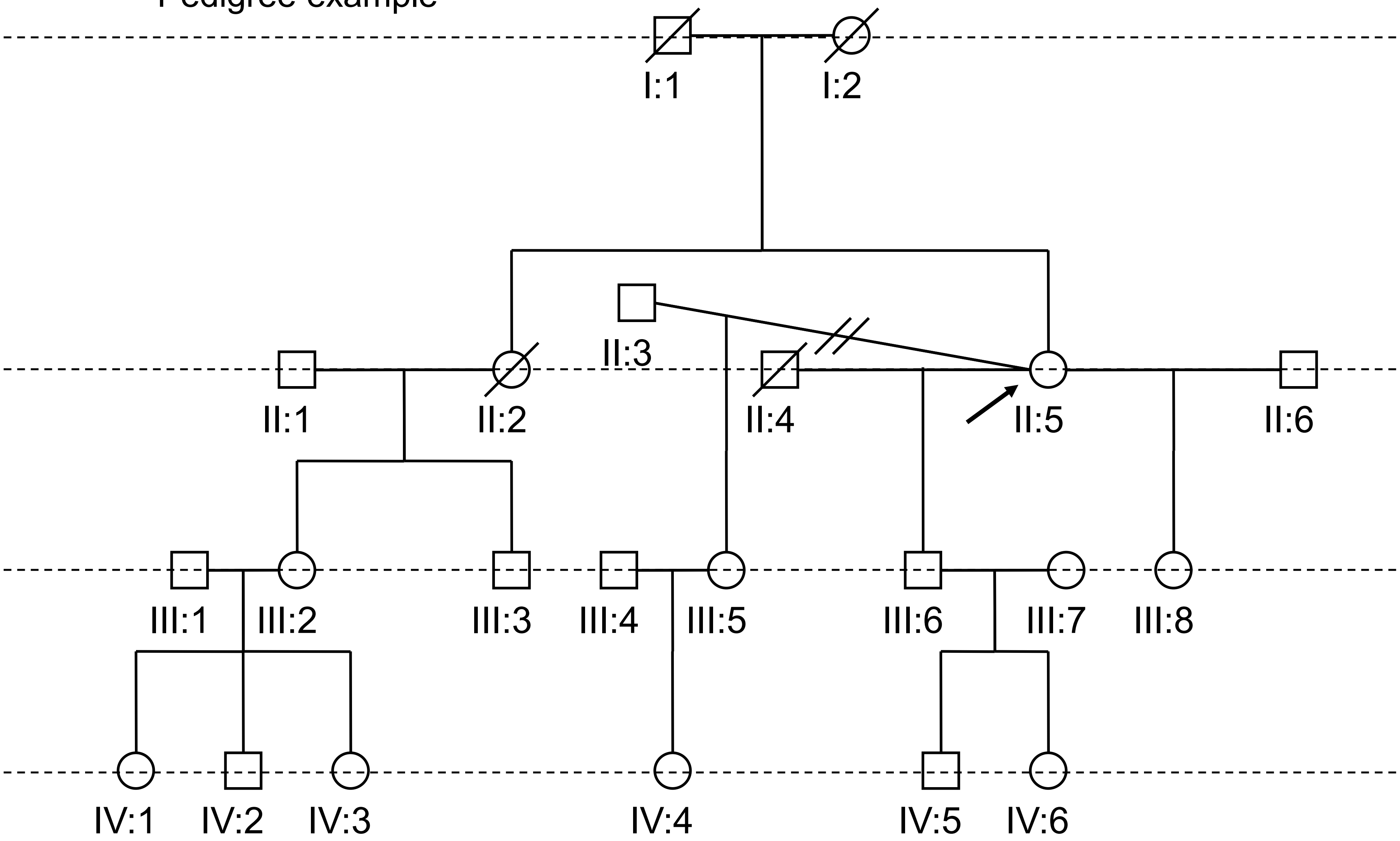


1st cousins

1st cousins once removed

2nd cousins

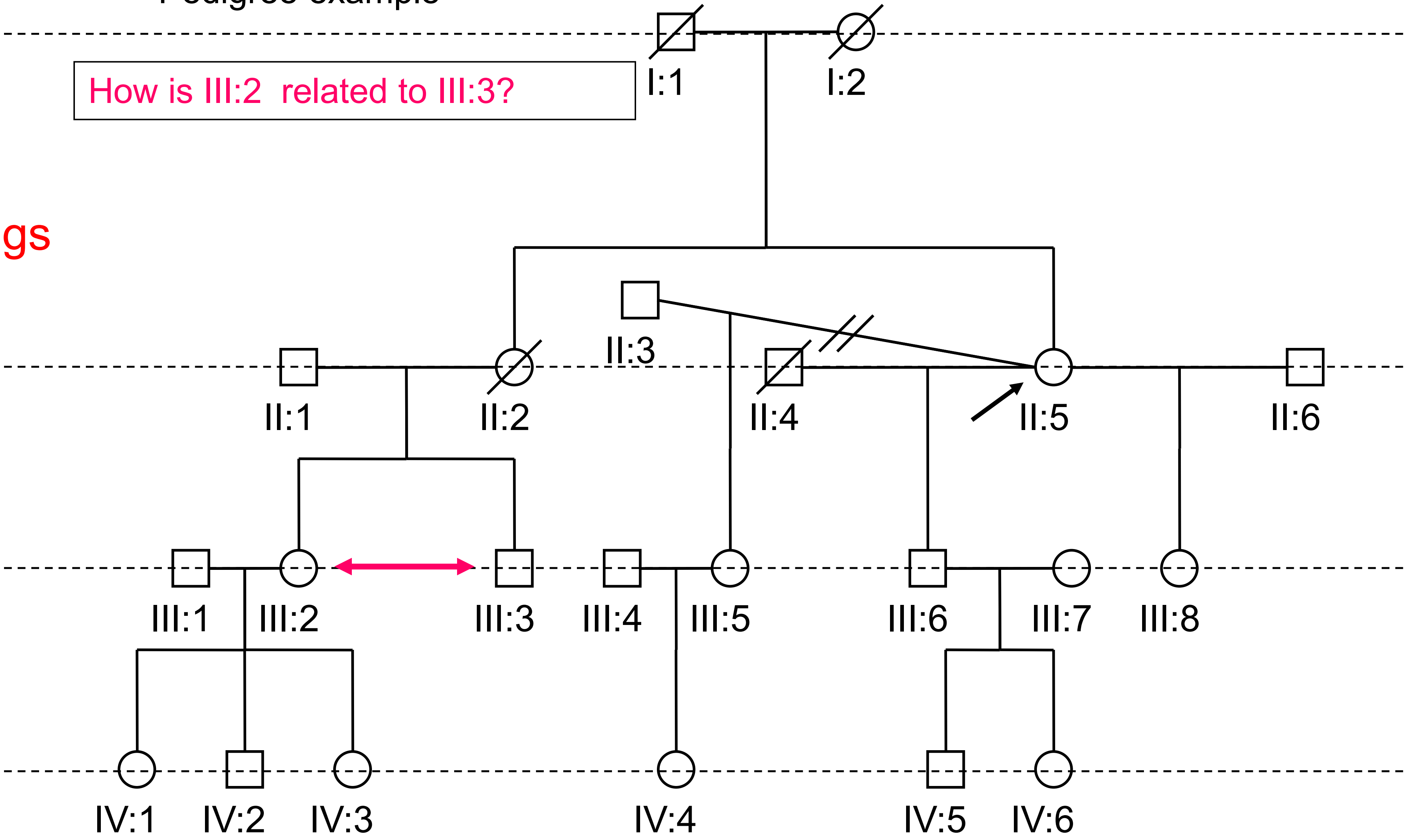
Pedigree example



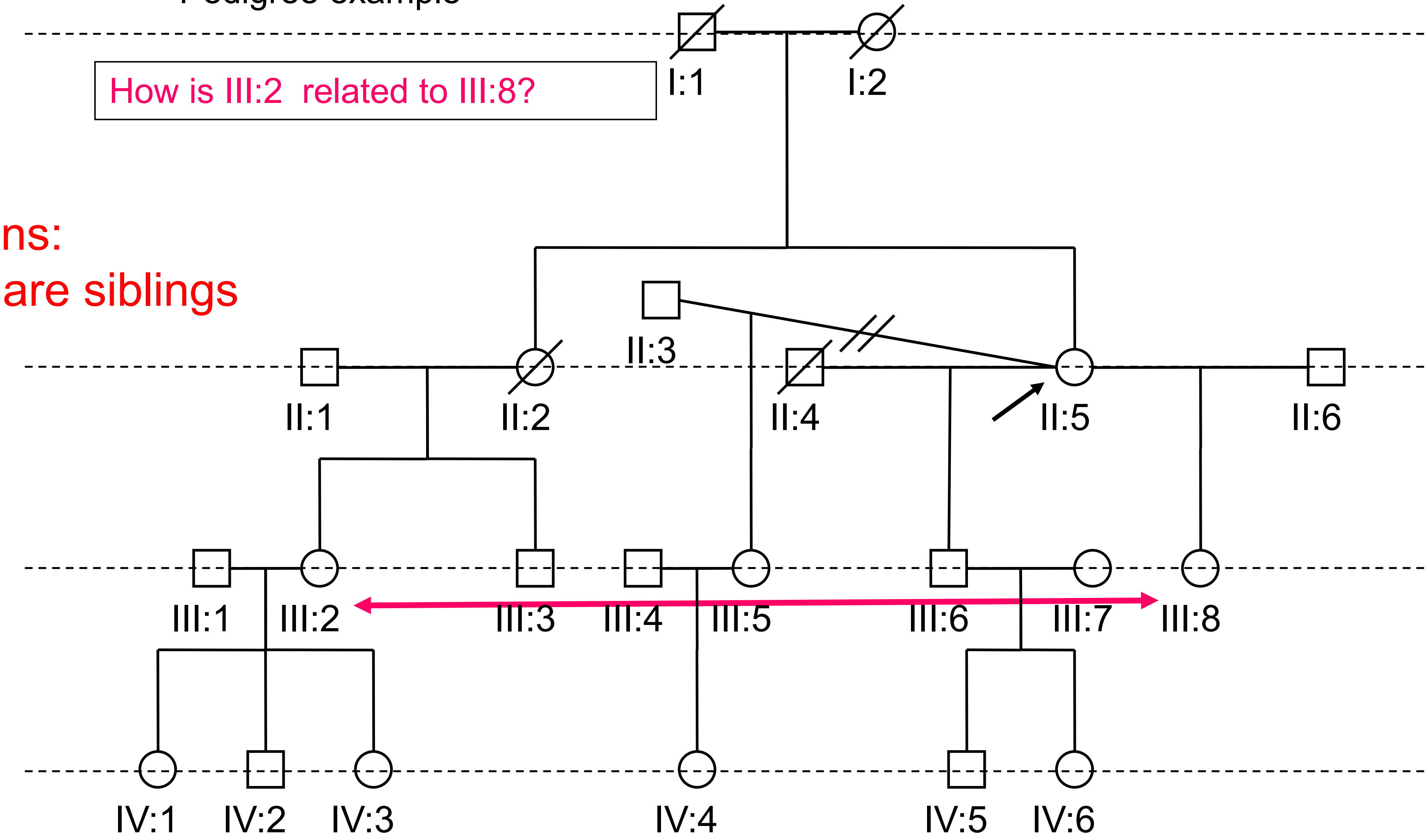
Pedigree example

How is III:2 related to III:3?

Full siblings



Pedigree example

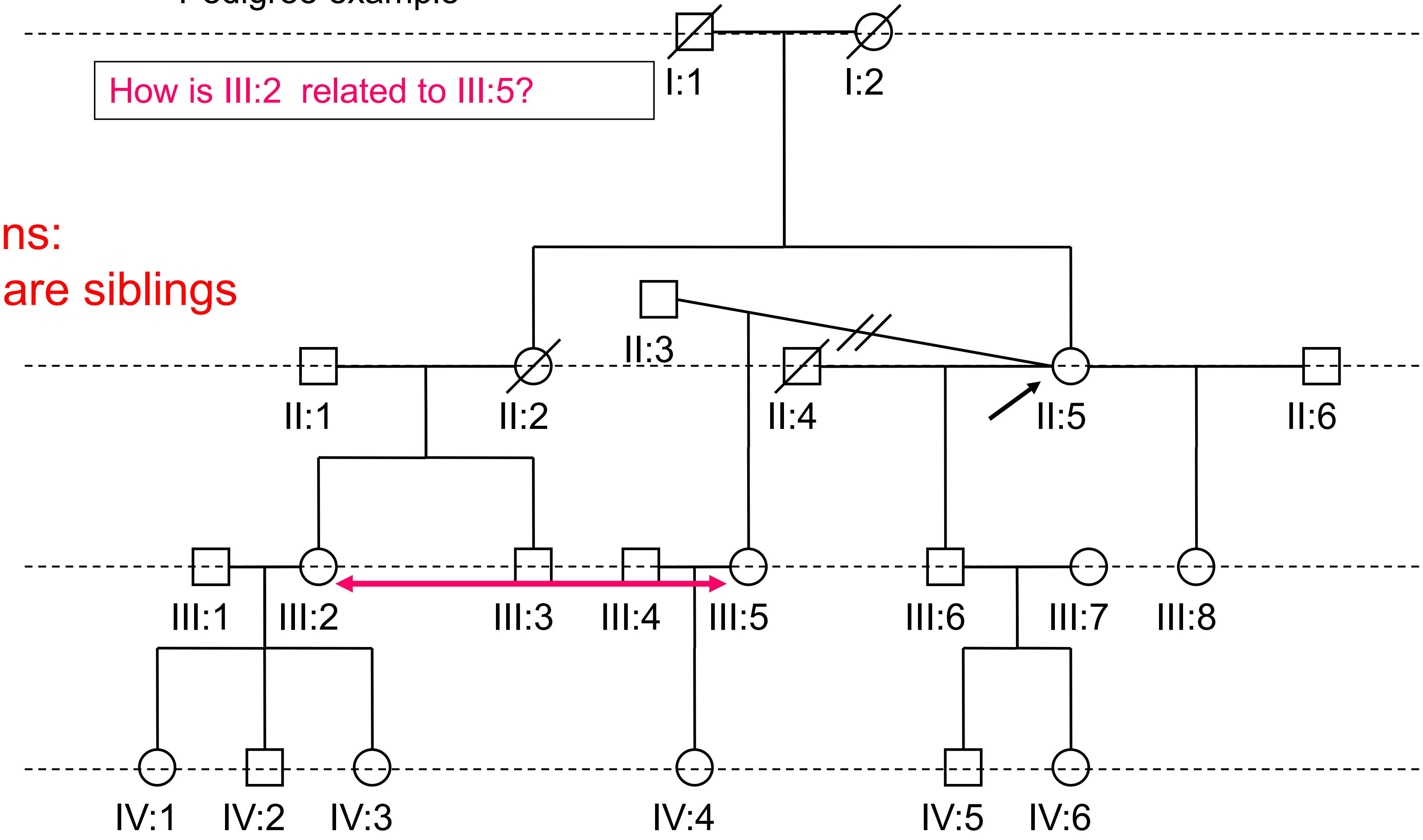

How is III:2 related to III:8?

1st cousins:
Parents are siblings

Pedigree example

How is III:2 related to III:5?

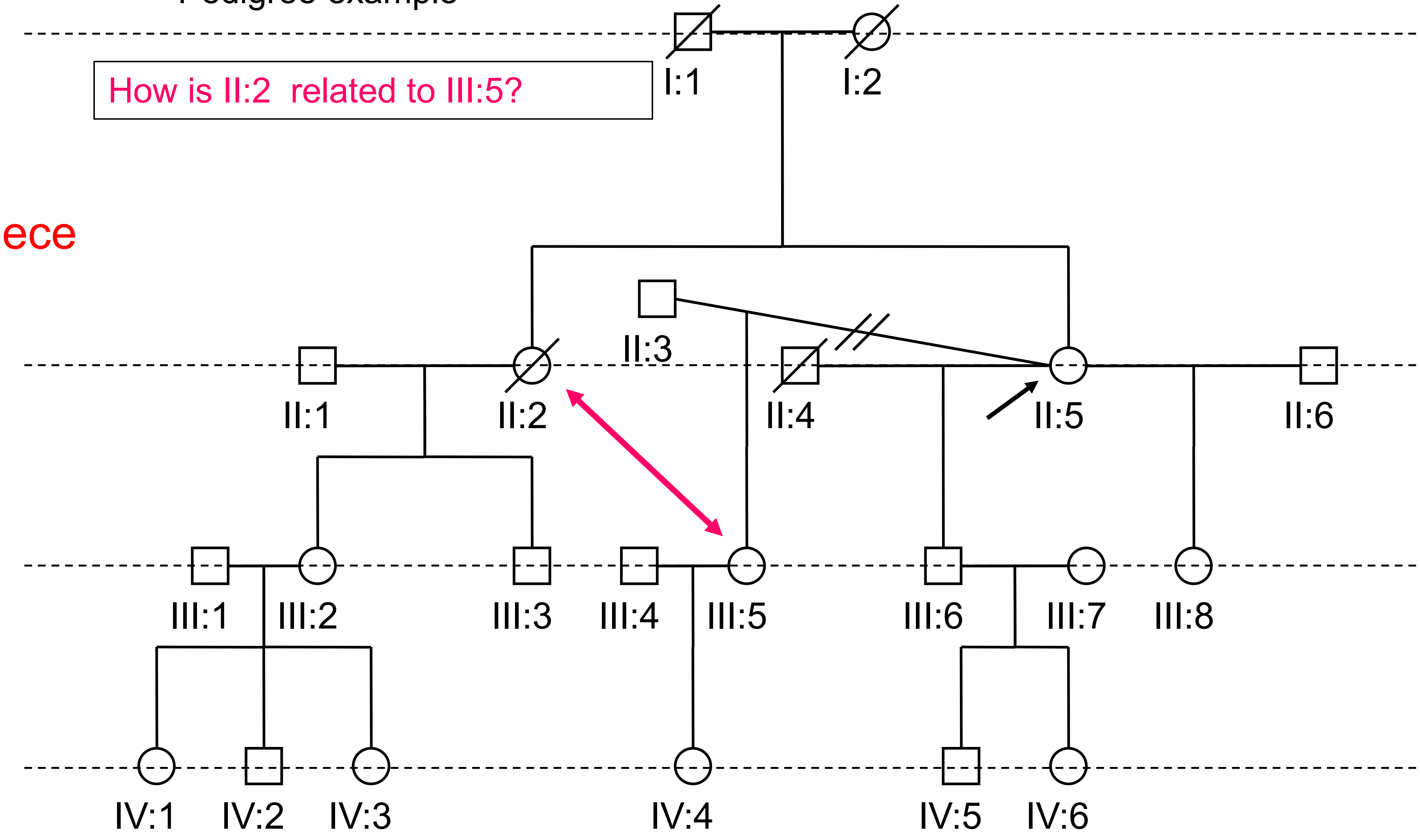
1st cousins:
Parents are siblings



Pedigree example

How is II:2 related to III:5?

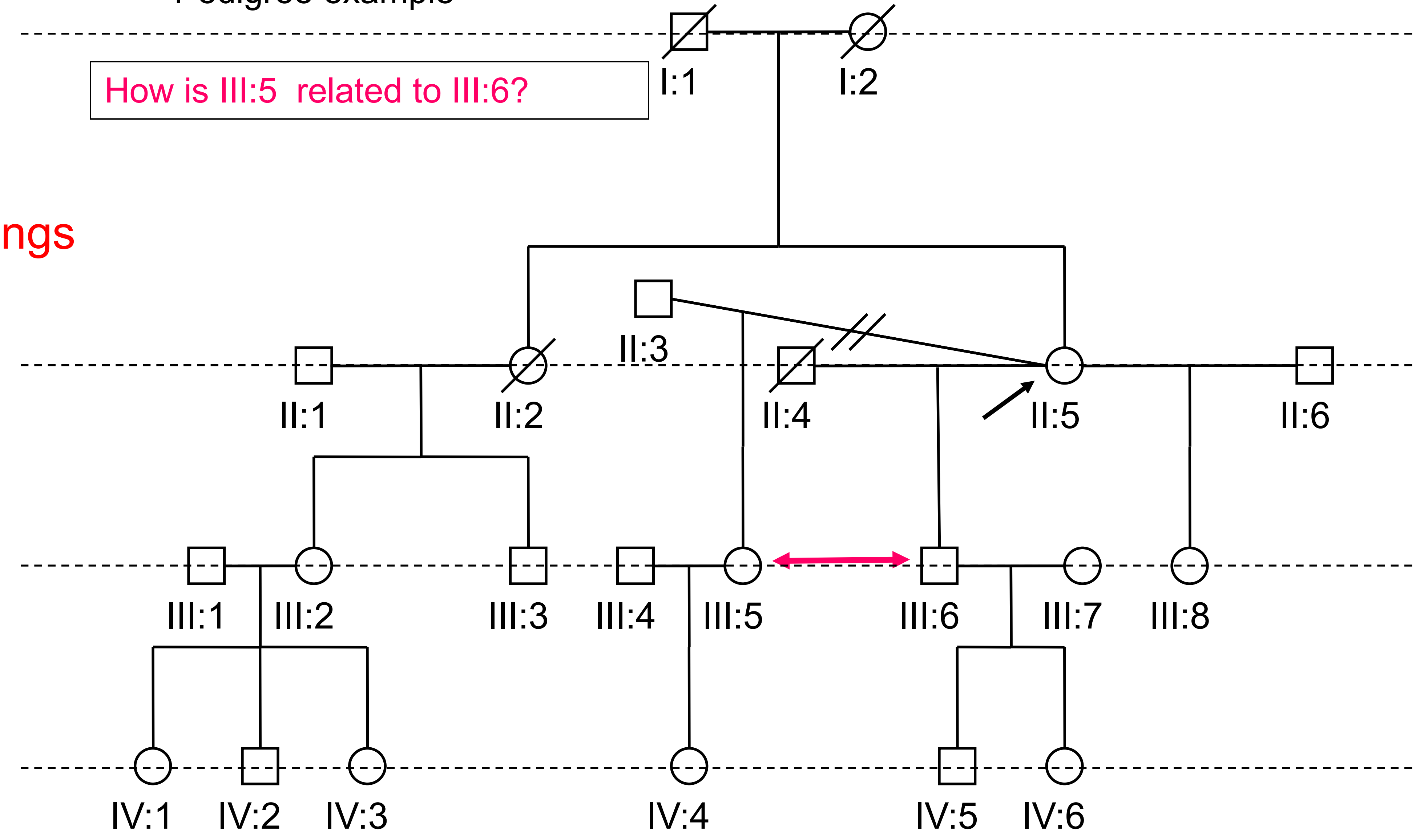
Aunt - niece



Half siblings

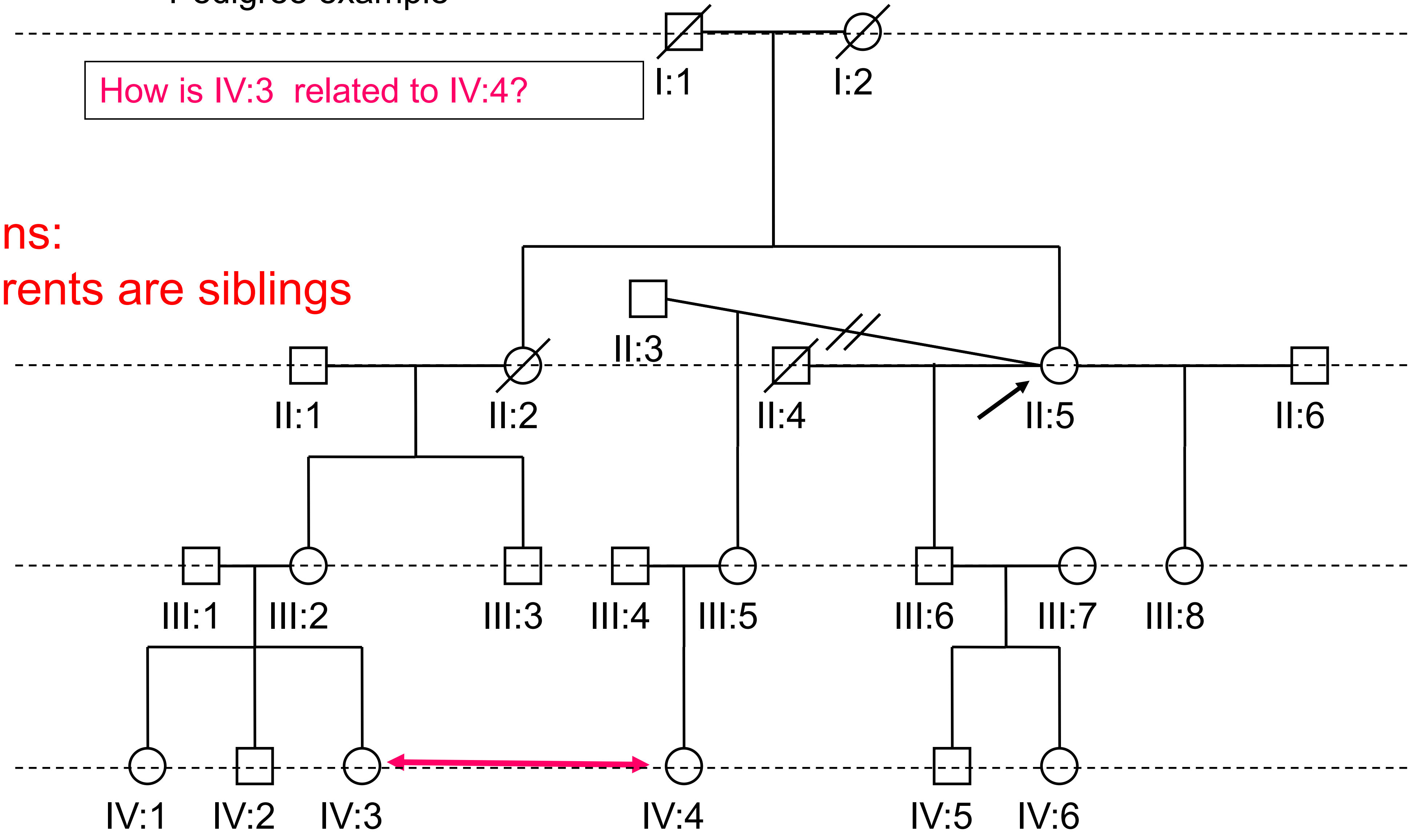
Pedigree example

How is III:5 related to III:6?



Pedigree example

How is IV:3 related to IV:4?

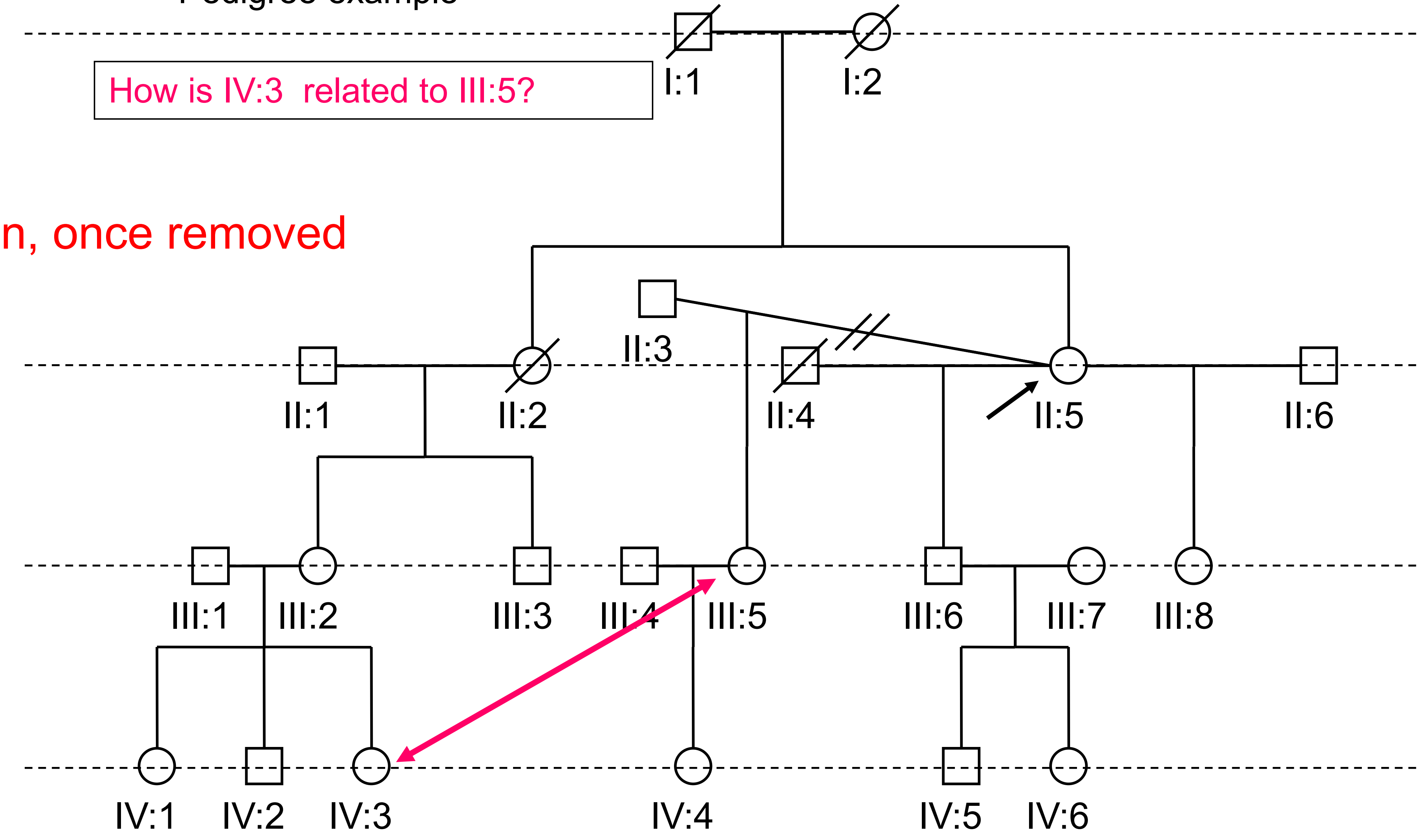


2nd cousins:
Grandparents are siblings

Pedigree example

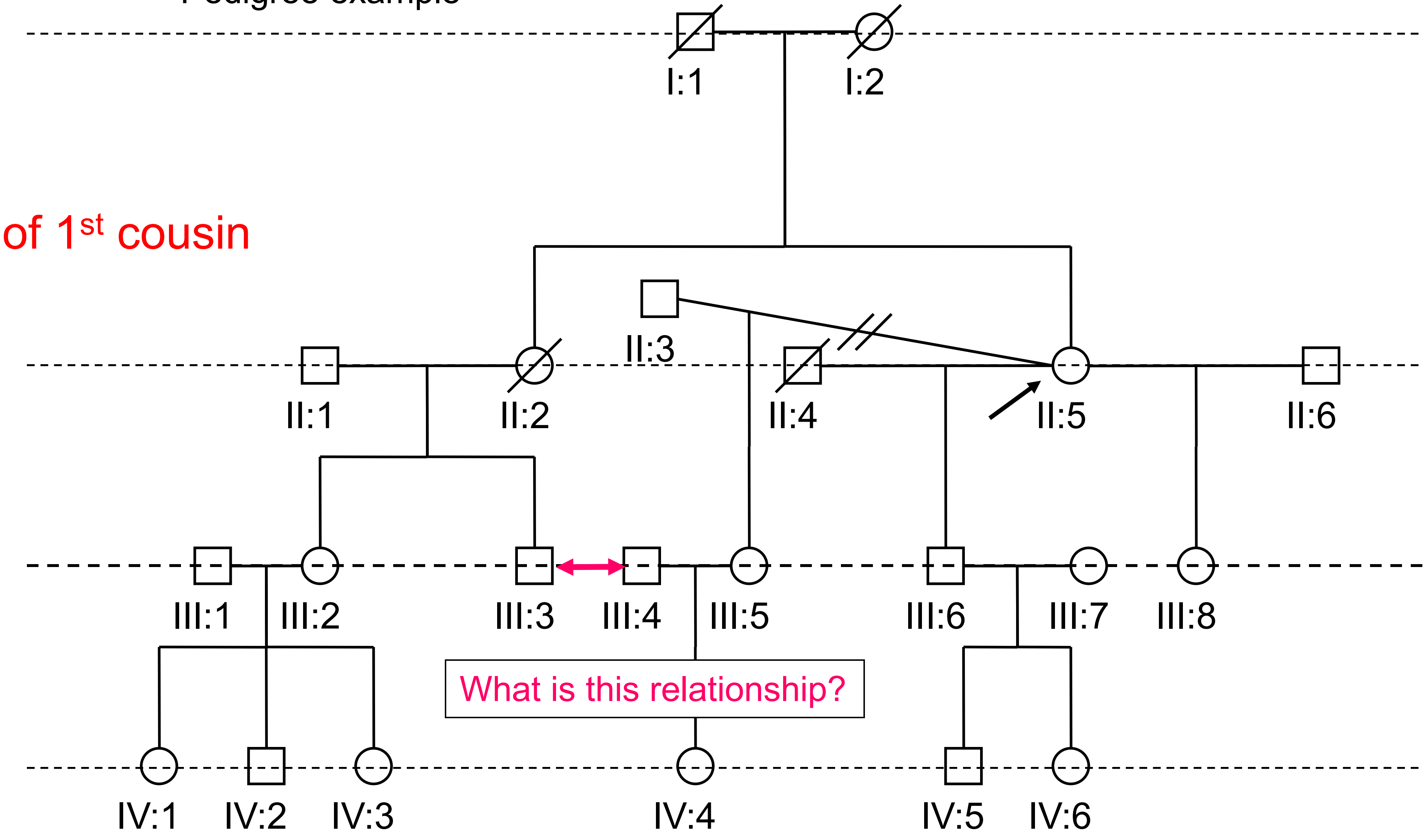
How is IV:3 related to III:5?

1st cousin, once removed



Pedigree example

Spouse of 1st cousin

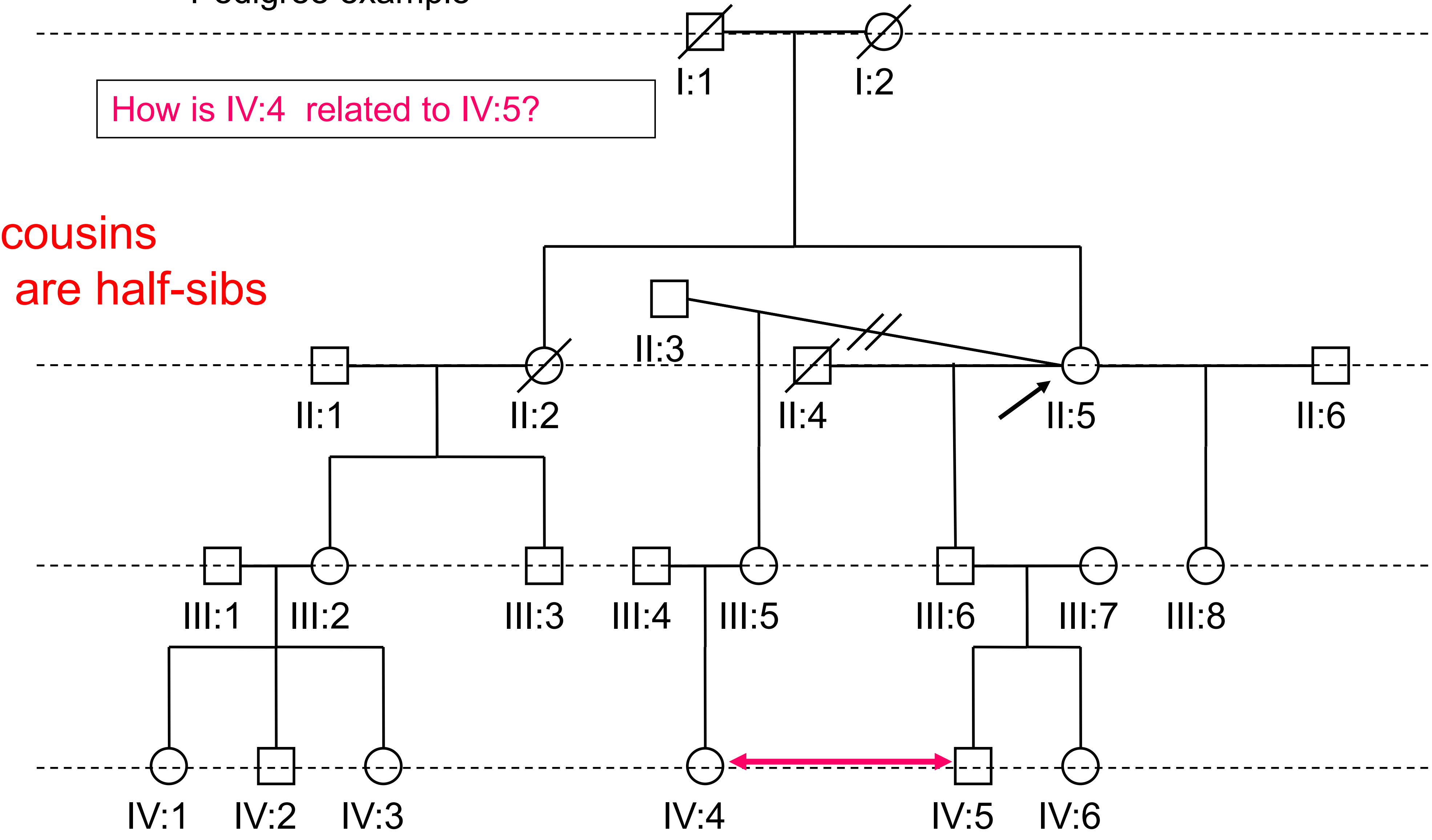


What is this relationship?

Pedigree example

How is IV:4 related to IV:5?

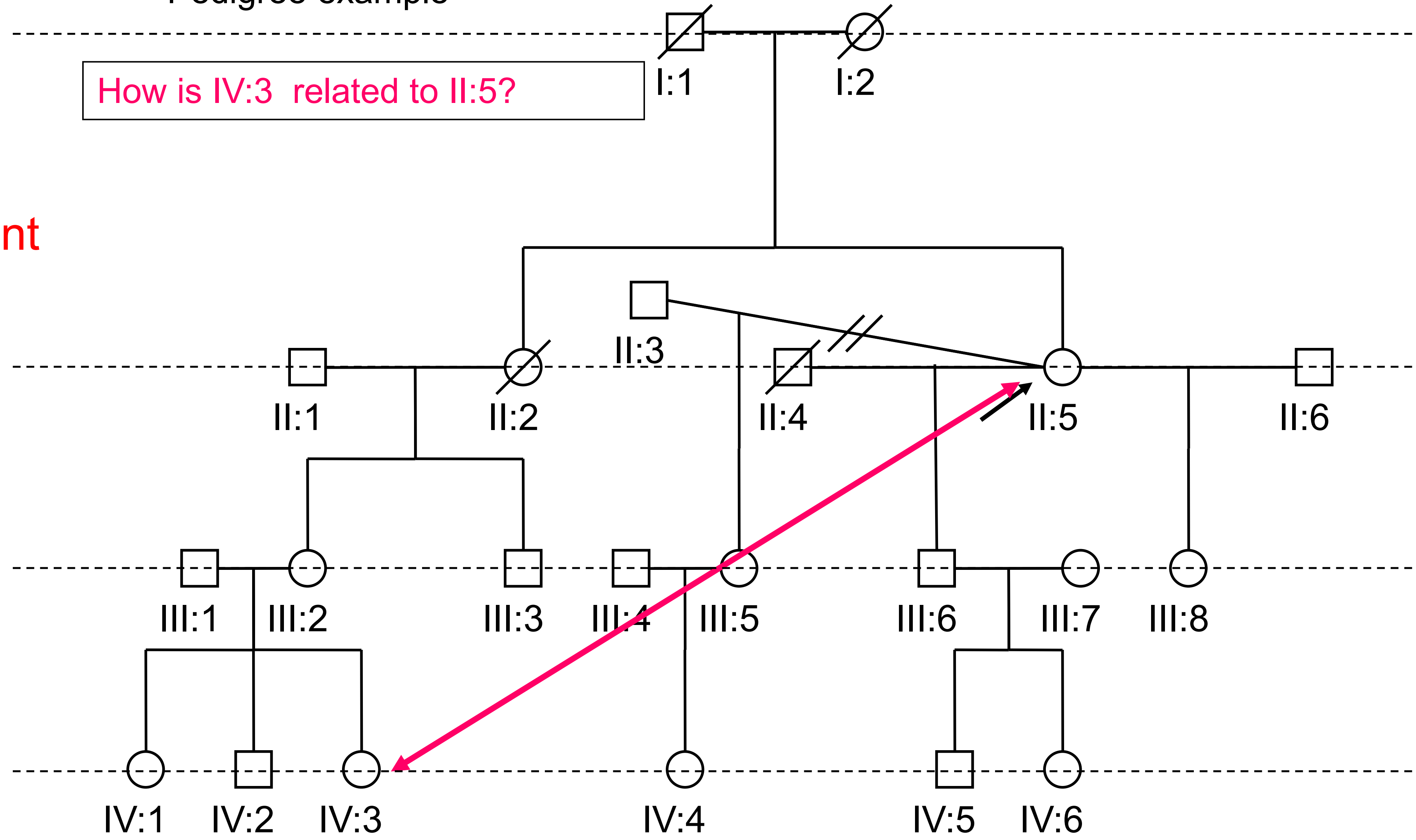
Half 1st cousins
Parents are half-sibs



Pedigree example

How is IV:3 related to II:5?

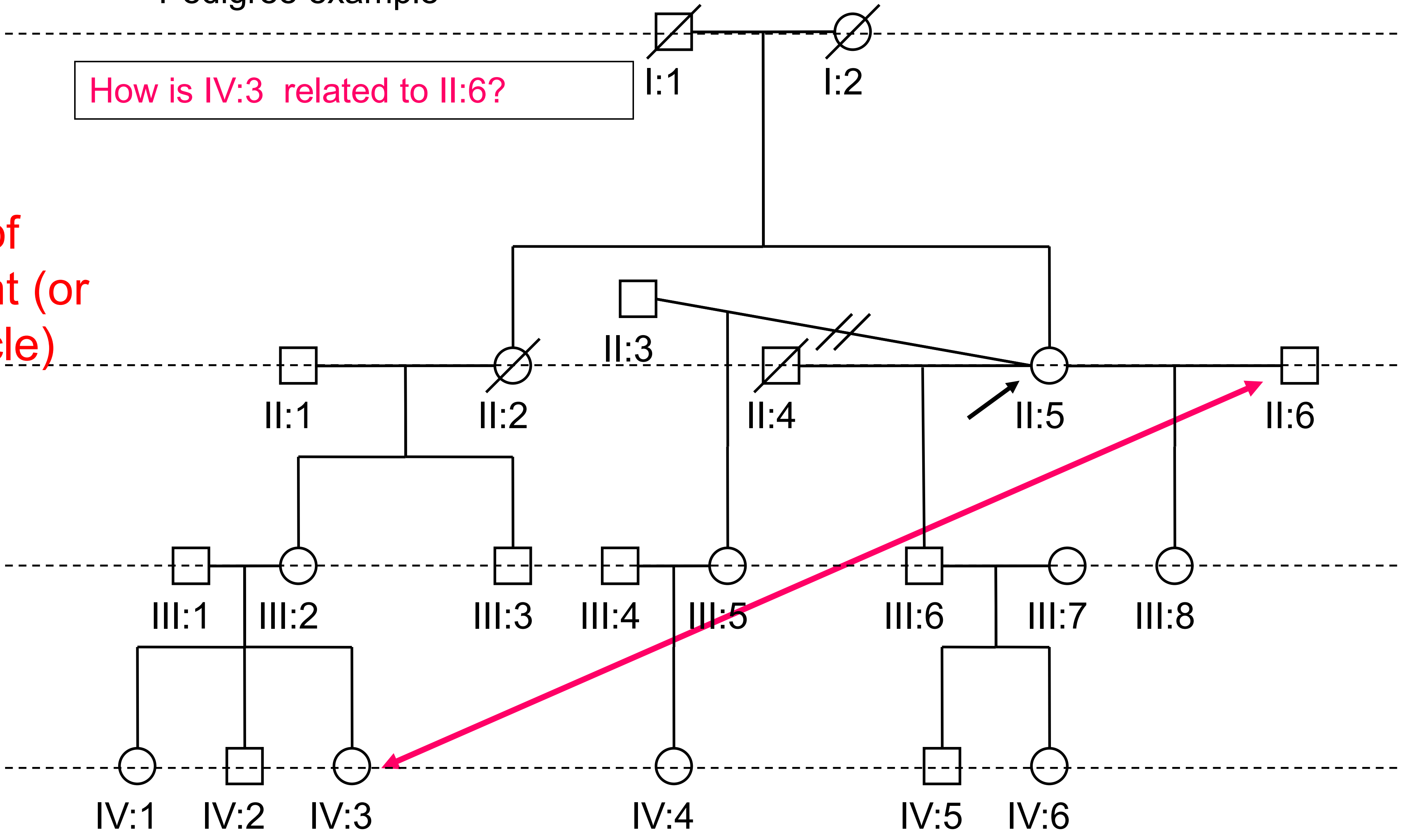
Great Aunt



Pedigree example

How is IV:3 related to II:6?

Spouse of great aunt (or great uncle)



Standard system

- Do one side of the family first (e.g., dad’s side)
- Don’t let the person take control by indicating different people in the family. It becomes disorganized.
- When asking about each person, ask about their health. Then, at the end, ask about anything that you missed. Others in the family with pertinent symptoms.
- Two ways to remember.

Standard system

- If they say, “my cousin X”, you need to make sure how they are related.
- Do not assume that cousin is really related or is a 1st cousin.
- “So, let me make sure how that person is related. Is she your mom’s, brother’s child?”
- Some cultures use the other terms for any distant relative (e.g., “**Auntie**” for any older woman)

Standard questions

- How many children does XX have?
- Are the all from the same 2 parents?
- If “YES”, who is the eldest?
- If “NO”, are any from the same two parents?
  - Are any from the same two parents
  - Who is the eldest?

- NOTE: get the structure in your mind BEFORE drawing

Issues that might arise

- “It used to be John, but now it is Jane” –
  - Which symbol do you use?
  - **NOTE**: Use correct symbol and add text below symbol explaining (social construct)
- How old is your daughter?
  - “She is 15”
  - Do you ask if she has kids?

Issues that might arise

- If you run out of space on a page (e.g., 10 aunts/uncles), get a second (or third) page and keep going
- Draw the connecting lines across pages
- At the end, number the pages at the top so you know the order

More info about taking a pedigree

- https://www.youtube.com/watch?v=Nmp_gLIMAFM
- https://www.youtube.com/watch?v=lKjuMX0qhmw

- Updated standard pedigree nomenclature (centered on sex and gender inclusivity)
  - https://onlinelibrary.wiley.com/doi/10.1002/jgc4.1621

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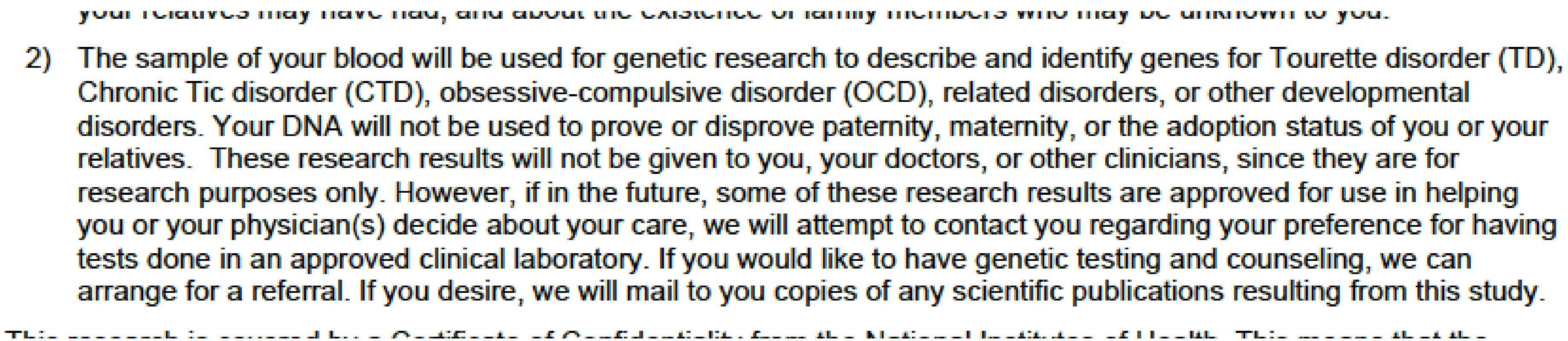
Informed Consent

- Provide informed consent (see example consent form)
- Review the consent form with the subject to ensure they understand what is involved (informed consent)
    - Need to understand everything that is in the consent form
    - Each study is different (e.g., do they return results?)
    - Indicate who funded the study

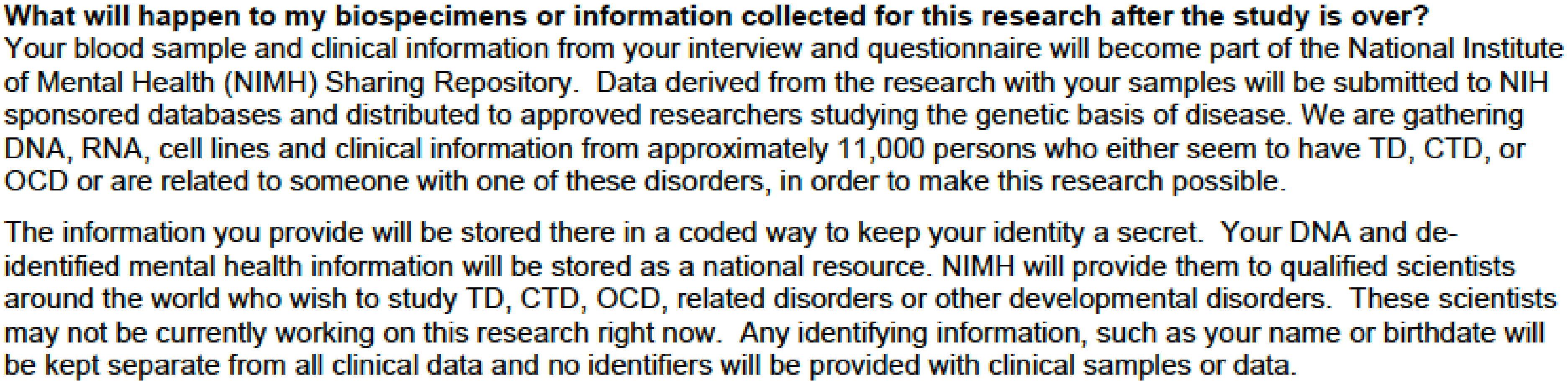
Snippets from Consent Form



your relatives may have had, and about the existence of family members who may be unknown to you.

2) The sample of your blood will be used for genetic research to describe and identify genes for Tourette disorder (TD), Chronic Tic disorder (CTD), obsessive-compulsive disorder (OCD), related disorders, or other developmental disorders. Your DNA will not be used to prove or disprove paternity, maternity, or the adoption status of you or your relatives. These research results will not be given to you, your doctors, or other clinicians, since they are for research purposes only. However, if in the future, some of these research results are approved for use in helping you or your physician(s) decide about your care, we will attempt to contact you regarding your preference for having tests done in an approved clinical laboratory. If you would like to have genetic testing and counseling, we can arrange for a referral. If you desire, we will mail to you copies of any scientific publications resulting from this study.

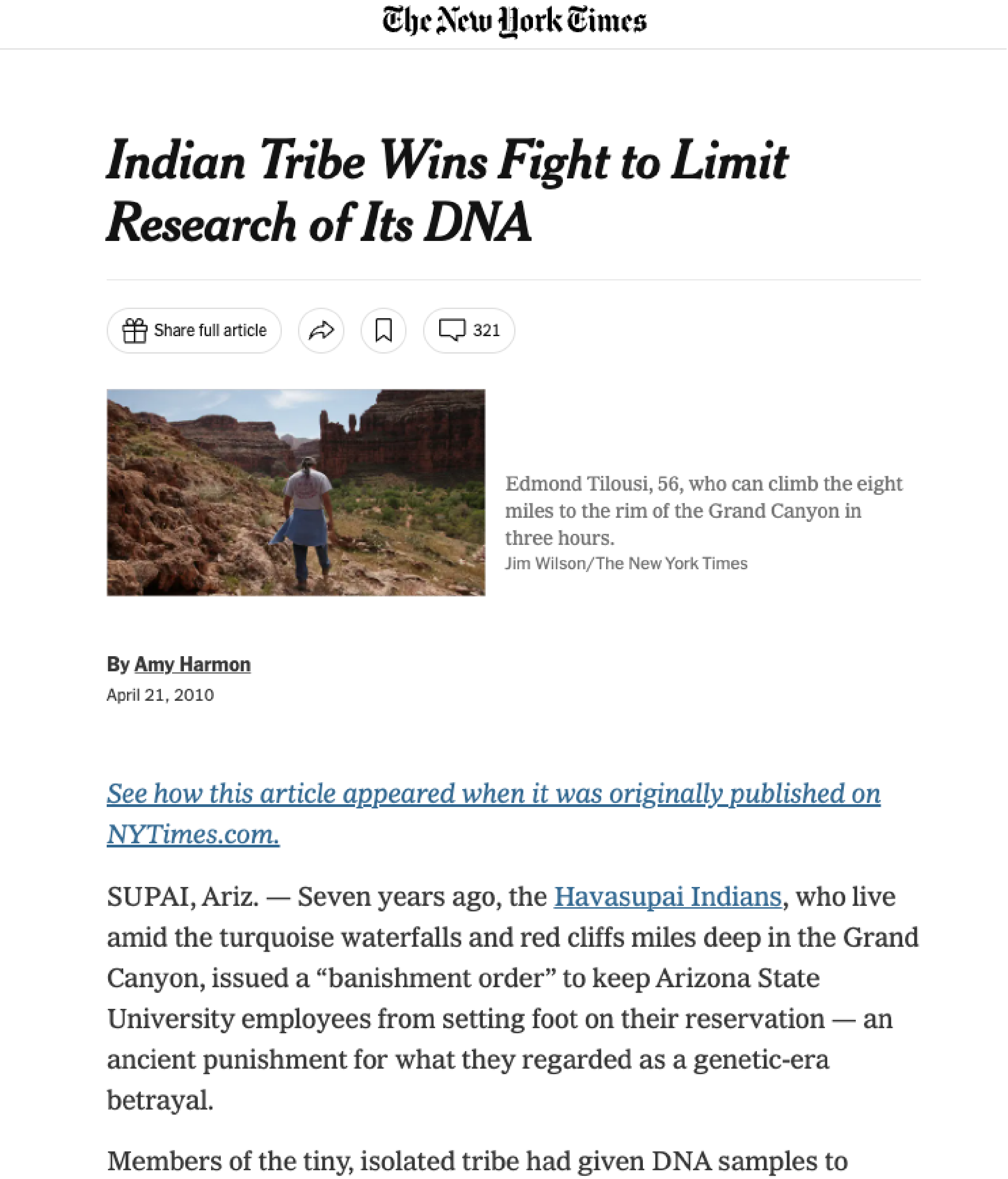
This research is covered by a Certificate of Confidentiality from the National Institutes of Health. This means that the



What will happen to my biospecimens or information collected for this research after the study is over?
Your blood sample and clinical information from your interview and questionnaire will become part of the National Institute of Mental Health (NIMH) Sharing Repository. Data derived from the research with your samples will be submitted to NIH sponsored databases and distributed to approved researchers studying the genetic basis of disease. We are gathering DNA, RNA, cell lines and clinical information from approximately 11,000 persons who either seem to have TD, CTD, or OCD or are related to someone with one of these disorders, in order to make this research possible.

The information you provide will be stored there in a coded way to keep your identity a secret. Your DNA and de-identified mental health information will be stored as a national resource. NIMH will provide them to qualified scientists around the world who wish to study TD, CTD, OCD, related disorders or other developmental disorders. These scientists may not be currently working on this research right now. Any identifying information, such as your name or birthdate will be kept separate from all clinical data and no identifiers will be provided with clinical samples or data.

Consent as a Process



The New York Times

Indian Tribe Wins Fight to Limit Research of Its DNA

Share full article 321

Edmond Tilousi, 56, who can climb the eight miles to the rim of the Grand Canyon in three hours.
Jim Wilson/The New York Times

By Amy Harmon
April 21, 2010

See how this article appeared when it was originally published on NYTimes.com.

SUPAI, Ariz. — Seven years ago, the Havasupai Indians, who live amid the turquoise waterfalls and red cliffs miles deep in the Grand Canyon, issued a “banishment order” to keep Arizona State University employees from setting foot on their reservation — an ancient punishment for what they regarded as a genetic-era betrayal.

Members of the tiny, isolated tribe had given DNA samples to

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Typical subject recruitment questions

Questions that subjects often ask in genetic studies during recruitment

Will I receive the results of the genetic testing?

- **Depends on the study, and you need to discuss with your PI (see consent form)**
- If NOT returning results, possible answers include:
  - Currently, there is no available test. We are trying to find the causes so that people have the option to get tested. Not all people want to know.
  - Remind the subject that this is not a test. A test means results. Instead, we hope that our research may help develop a test in the future. Once the study finds information that may be helpful to those with the disorder and their relatives, provide foundations and participants with what it means and the options available.
  - We hope that this study will provide individuals with the option to get testing. If so, each person can decide to get tested or not. If testing becomes available, we can refer you to genetic counselors who can work with you to get testing

Will I find out if I have the gene or which side of family it comes from?

- **This is not a test** (see return of results slide).
- Currently, XXX is only diagnosed clinically. We hope that our research may help develop a test in the future. Therefore, we will not be able to provide you with this information.
- We are using a research lab that does not have the same strict protocols as a lab for patient testing. If you want to get tested, I can refer you to a genetic counselor.

How will this research help me?

- **Typical consent form language**
- This study will not benefit you. Although you personally will not receive any direct benefit from this project, individuals who might develop XXX in the future, their family members, and future generations may benefit if we can find genes that lead to such disorders. We do not expect to discover any information of direct clinical relevance to your condition or treatment during the next few years. We hope that the results of our research will enable the development of new treatments that are specifically designed to treat the cause or causes. Hopefully, this will occur before the next generation begins to have symptoms.

Will the samples be used for other disorders (sharing repository)?

- **This is study-specific. Discuss with PI and review the consent form (it may indicate other disorders).**
- Find out what ultimately happens with DNA and data (become part of a sharing repository)
- Your specific identity is kept confidential by the PI of the study you participate in, even if the samples are used to study other disorders.
- Having other investigators use the samples is a good thing - it helps speed up research, so we find answers more quickly

Do I or my child have to give a blood sample or is saliva sample sufficient?

- This is a study-specific question- ask your PI

Will you clone me?

- We will not clone you.
- Firstly, we can’t: it’s really hard to clone a human, the technology isn’t there, and it may never be. Even if we hypothetically could, the resources (money and lab infrastructure) would be too expensive for any lab to afford human cloning.
- Instead, we are interested in finding the cause for the disorder in the hopes to develop better treatments

Can you test for paternity?

- This would need to be discussed with the PI,
- It depends on a number of factors, including the study design. For example, it is possible in family-based studies for non-paternity to be identified. However, whether that information would be returned to research participants would depend on the IRB-approved study protocol.
- Typically, the answer is: NO, we do not return non-paternity findings.

Can you test for paternity? (continued)

- The sample of your blood will be used for genetic research to describe and identify genes for XXX.
- Your DNA will not be used to prove or disprove paternity, maternity, or the adoption status of you or your relatives. These research results will not be given to you, your doctors, or other clinicians, since they are for research purposes only. However, if in the future, some of these research results are approved for use in helping you or your physician(s) decide about your care, we will attempt to contact you regarding your preference for having tests done in an approved clinical laboratory. If you would like to have genetic testing and counseling, we can arrange for a referral. If you desire, we will mail to you copies of any scientific publications resulting from this study.

Will my genetic information be protected?

- **There should be a section of consent for dealing with this**
- There is a federal law called the **Genetic Information Nondiscrimination Act** (GINA) that helps protect against genetic discrimination. In general, this law makes it illegal for health insurance companies, group health plans, and most employers to discriminate against you based on your genetic information. This law generally will protect you in the following ways: (1) health insurance companies and group health plans may not request your genetic information that we get from this research; (2) health insurance companies and group health plans may not use your genetic information when making decisions regarding your eligibility or premiums; and (3) employers with 15 or more employees may not use your genetic information that we get from this research when making a decision to hire, promote, or fire you or when setting the terms of your employment. However, it does not protect you against discrimination by companies that sell life insurance, disability insurance, or long-term care insurance.

Will my genetic information be protected?

- The information you provide will be kept completely confidential. All personal identifying information will be stored in our office in locked filing cabinets and will not be released to anyone outside the research staff. Your name and address will be entered into a research database accessible only by using a password that is unknown to individuals outside of our research staff. For laboratory and statistical analyses, and publication of any results from the study, you will be identified by a number, and no personal information will be used.

Will my genetic information be protected? (continued)

- None of the information you provide will be released to any of your family members without your permission. Similarly, none of the information your relatives provide will be released to you without their permission. This includes information about medical conditions you or your relatives may have had, and about the existence of family members who may be unknown to you.

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NJacts

Rutgers Institute
for Translational
Medicine & Science

Genetically Informed Research, Education, and Treatment (GREAT):
Introduction to a new resource to expand research capacity

http://njacts.rbhs.rutgers.edu

RUTGERS
UNIVERSITY

PRINCETON
UNIVERSITY

NJIT
New Jersey's Science &
Technology University

RWJBarnabas
HEALTH

NJ acts

Overview

- A new component of our core program focused on the impact of genetically informed approaches to research and treatment on translational research and translational science.
- The GREAT core will provide tailored instruction, mentoring, and analytic support for both translational research and translational science.
- This support is designed to address the complexities inherent to conducting genetically informed research in diverse populations.

RUTGERS UNIVERSITY

PRINCETON UNIVERSITY

NJIT New Jersey's Science & Technology University

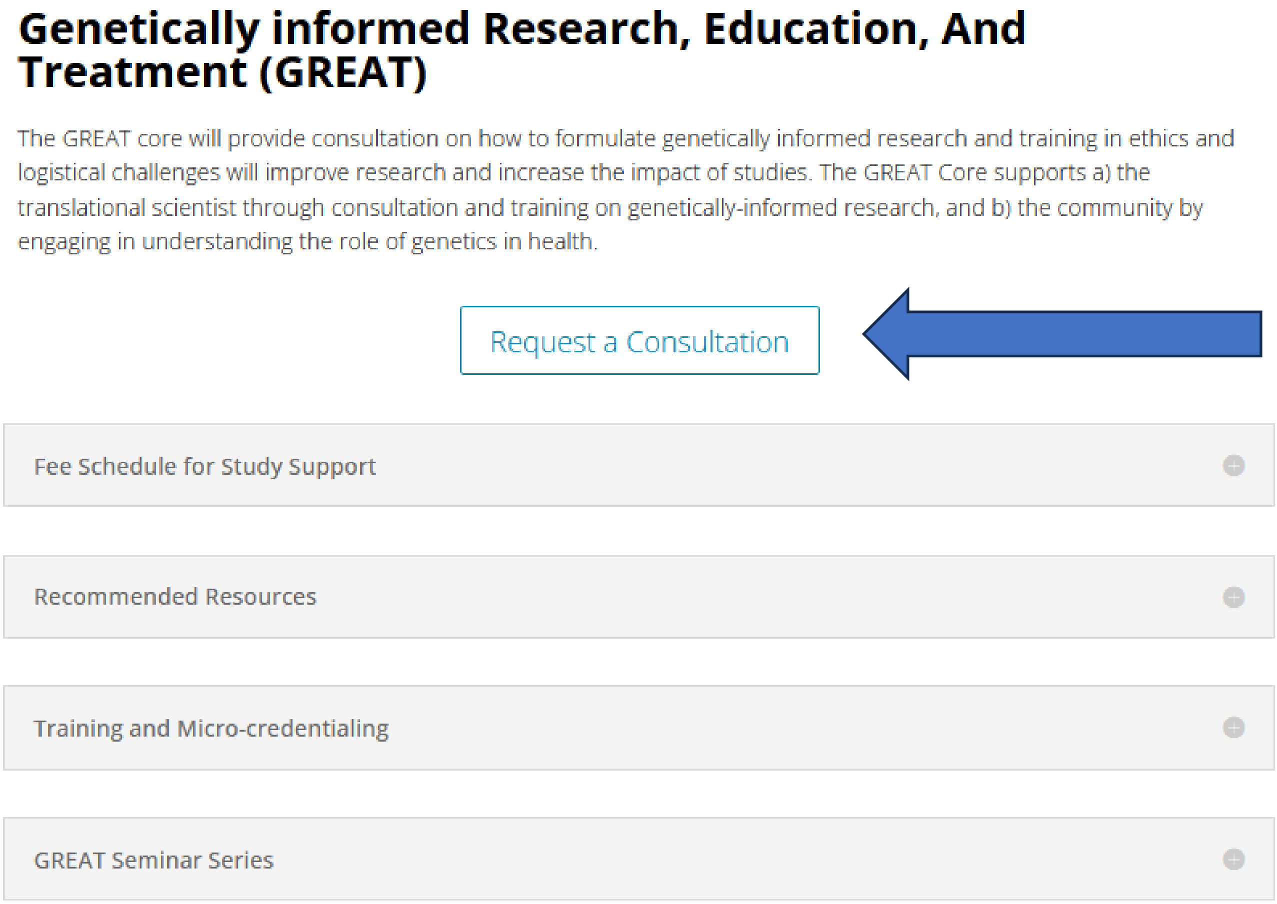
RWJBarnabas HEALTH

Overall Goals

- Provide consultation on how to formulate genetically informed research, and training in ethics and logistical challenges will improve research and increase the impact of studies.
- Supports:
  a) translational scientists through consultation and training on genetically-informed research,
  b) the community by engaging in understanding the role of genetics in health.

The GREAT Team is here to help!

Request a Consultation via our website



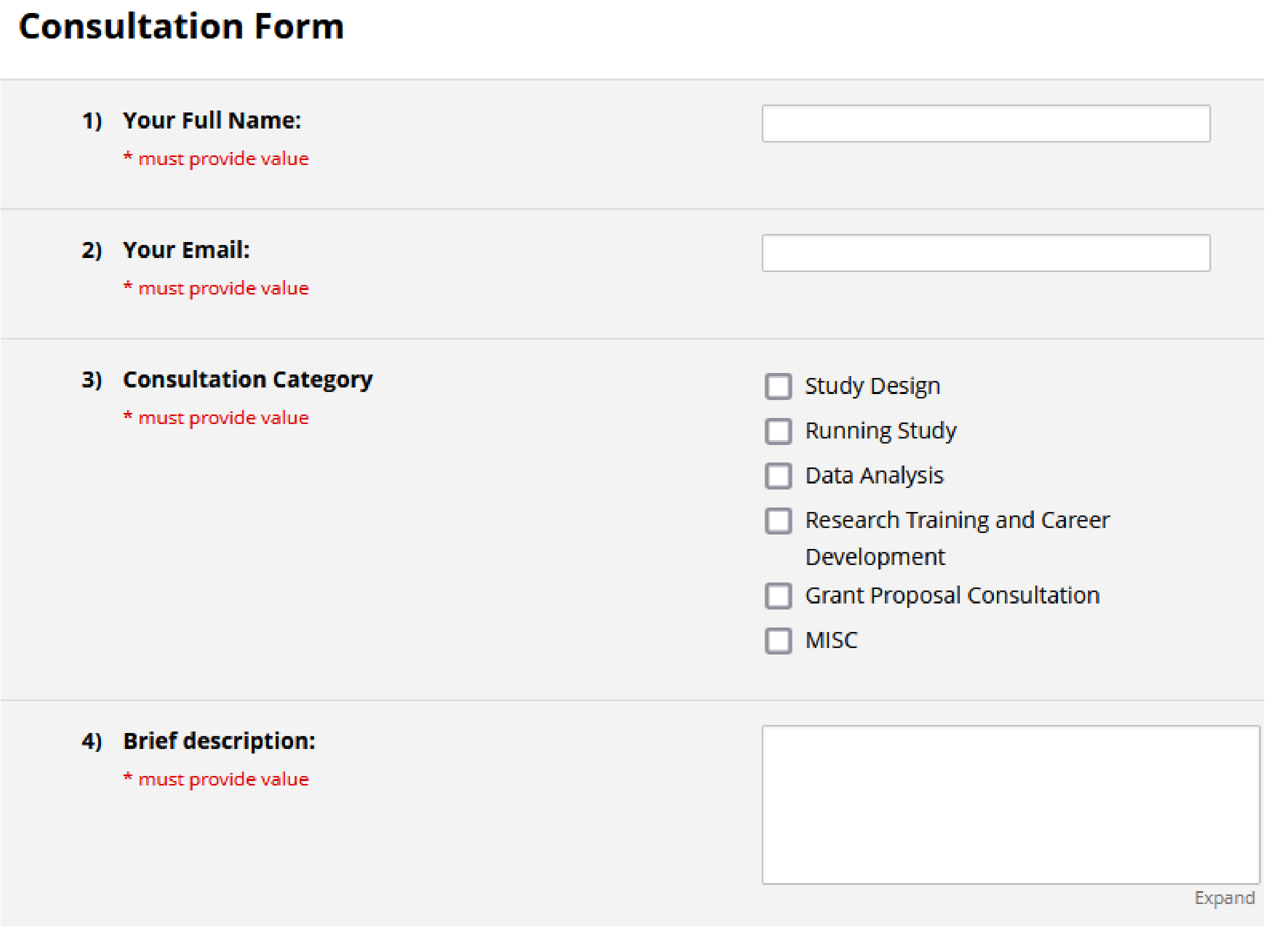
Genetically informed Research, Education, And Treatment (GREAT)

The GREAT core will provide consultation on how to formulate genetically informed research and training in ethics and logistical challenges will improve research and increase the impact of studies. The GREAT Core supports a) the translational scientist through consultation and training on genetically-informed research, and b) the community by engaging in understanding the role of genetics in health.

Scan me!

https://njacts.rbhs.rutgers.edu/investigator-resources/great/

Consultation Request



Consultation Form

1) **Your Full Name:** * must provide value

2) **Your Email:** * must provide value

3) **Consultation Category** * must provide value

- ☐ Study Design
- ☐ Running Study
- ☐ Data Analysis
- ☐ Research Training and Career Development
- ☐ Grant Proposal Consultation
- ☐ MISC

4) **Brief description:** * must provide value

Questions?

Thank you!