DRAWING A FAMILY PEDIGREE
Independent Learning module:

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TERMINAL LEARNING OBJECTIVES:
After completion of this session the student should be able to:

- Given a family history, draw a 3 generation pedigree using standard symbols, and interpret the possible mode(s) of inheritance of a given trait in the family.

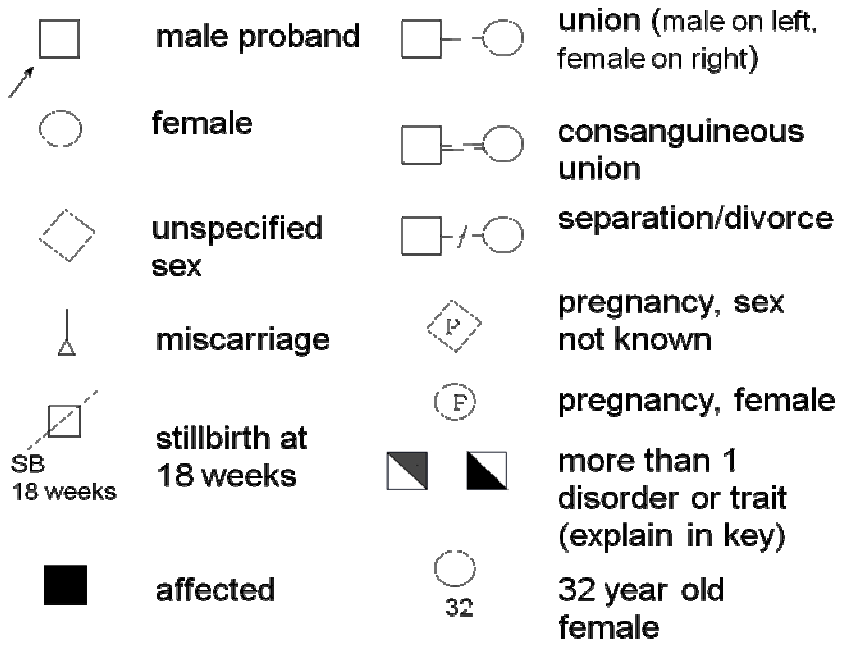
WEB-BASED LEARNING:
Complete the NHPEG powerpoint presentation on Family History Principles at
http://www.nchpeg.org/index.php?option=com_content&view=article&id=145&Itemid=64

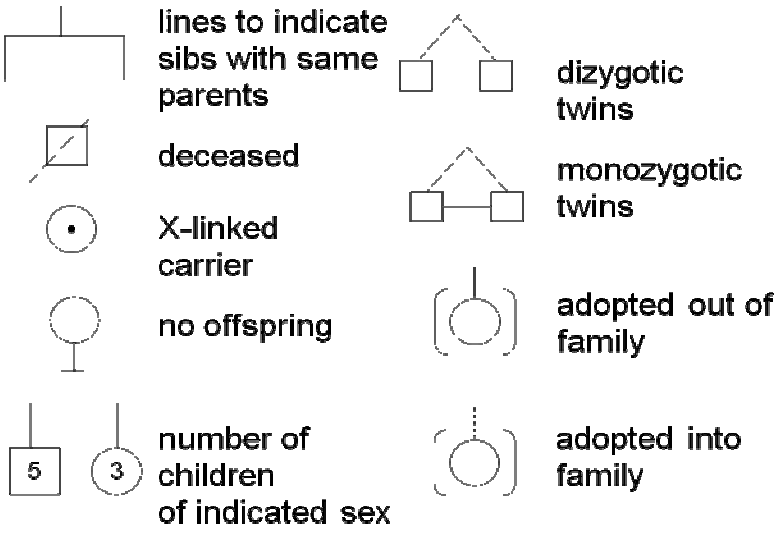
THE FAMILY PEDIGREE

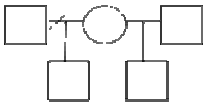
A. Steps in drawing a pedigree (use the template provided)

1. In corner of the page, indicate date of consultation, surname of consultand (the person seeking advice regarding their risk), your name and the date that you create the pedigree
2. Draw in the **consultand** – the person seeking advice.
3. Draw in the consultand's parents and sibs or spouse and offspring and indicate their ages. N.B. With a couple, draw the male on the left side and the female on the right – this makes it easier to interpret a pedigree for possible X-linked inheritance. If a sib has children, but there is no pertinent information from the sib's spouse's side of the family, you don't need to draw in the spouse.
4. Indicate the **proband** (also known as propositus or index case) with an arrow. This is the person affected with a genetic disorder through whom a family is first brought to attention. For example, the relative with Down syndrome in a family seeking prenatal counseling or the parent who is affected with Marfan syndrome and wanting to know about risks for his or her children. In the latter case, the consultand and the proband are the same individual.
5. Expand the pedigree to include at least 3 generations
6. Indicate ethnic origin on both sides. Ask about consanguinity ("is there any chance that you are related by blood?" or "do you have any relatives in common?")
7. Create a key to denote features of interest in affected individuals.
8. Are there any relatives with identical or similar features to your index case or any other health concerns?
9. Are there any disorders that "run in the family"?
10. Has anyone in the family had miscarriages, stillbirths, or infant deaths?
11. Are there any relatives with developmental delay/learning problems/birth defects such as cleft lip and palate or structural heart problems?
12. If any deaths, what was the cause?

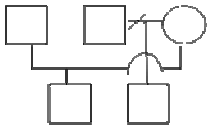
B. Standard symbols used in creating a pedigree:
(adapted from Bennett RL et al (1995) Recommendations for standardized pedigree nomenclature. J Genet Counsel 4:267-279)







female who has had 2 partners, no longer with first, has a son by each (either notation is acceptable)



C. Interpreting the pedigree

Throughout the Medical Genetics course, you will be learning about the typical features of various inheritance patterns. The following are some of the "clues" to look for in the family history.

Feature	**Possible clue to:**
recurrent miscarriages	familial chromosome rearrangement
stillbirths	chromosome anomaly or perinatal lethal single gene defect
affected relatives in different generations	autosomal dominant/X-linked
consanguinity/same ethnic origin	autosomal recessive
late maternal age	chromosomal aneuploidy
late paternal age	new dominant mutation
affected sibs, normal parents	autosomal recessive
affected males, unaffected mothers	X-linked recessive
maternal transmission only	mitochondrial

PRACTISE DRAWING A PEDIGREE: Review the standard notation and tutorial on constructing and interpreting pedigrees, as above

Draw your own family pedigree. Complete the Family Health Portrait at https://familyhistory.hhs.gov/fhh-web/home.action and compare the results with what you have drawn.

Draw a family pedigree using the information given below, using the template on the next page.

Mr. and Mrs. Kruger, a husband and wife of Dutch origin, are distantly related. They are concerned about their child who has been diagnosed with Hurler syndrome. The mother is one of a sibship of 4. One of mother's 2 brothers had a daughter who died in infancy. Father has a 4 year old son from a previous relationship. Father's sister has had 3 miscarriages and one healthy son.

FAMILY PEDIGREE

Origin _____/_____ _____/_____

I

II

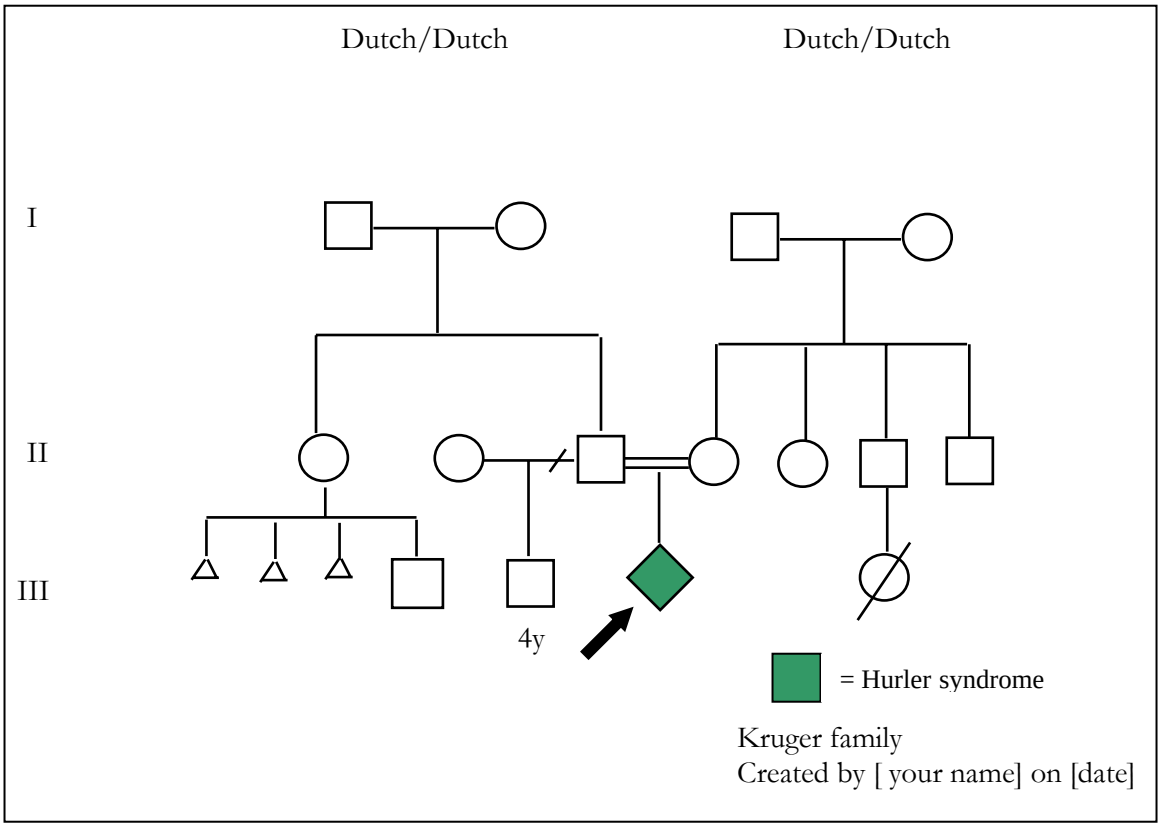
KEY

III

_____ family

Created by _____ on _____

How does your completed pedigree match with the following?



Take your own family history from a relative, and draw the pedigree based on information you obtain. Use the template. This will give you practice asking the appropriate questions, and recording the information in pedigree form. If possible, record the interview, so that you can refer back to it to clarify any details. Follow the steps in drawing a pedigree (under Introduction to Medical Genetics), and be sure to use standard notation. Is there any genetic condition in the pedigree? Can you identify any pattern of inheritance?

CASE STUDY: "Royal blood"

Queen Victoria married her first cousin, Albert, and had several children. Of her children:
(a) One son, Leopold, had hemophilia A. (b) Daughter Beatrice had a daughter, Victoria, who married King Alfonso of Spain and gave birth to 7 children: the oldest and youngest children (boys) had hemophilia A, the second child (girl) had congenital deafness, the 3rd and 5th were healthy girls, the 4th was a stillborn male and the 6th, Juan (a healthy male), became the father of King Juan Carlos of Spain. (c) Daughter Alice had a daughter, Alexandra who married Nicholas II, Czar of Russia and had 4 healthy daughters and a son, Nicholas, who had hemophilia A. (d) Daughter Victoria married Friedrich, emperor of Russia and had healthy children. (e) Son Albert was the grandfather of King George V.
Draw Queen Victoria's family pedigree. How does your pedigree match with the pedigree located at www.lhsc.on.ca/Patients_Families_Visitors/Genetics/Resources/index.htm"