Pedigrees and Karyotypes
A pedigree shows the relationships within a family and it helps to chart how one gene can be passed on from generation to generation. Pedigrees are tools used by genetic researchers or counselors to identify a genetic condition running through a family, they aid in making a diagnosis, and aid in determining who in the family is at risk for genetic conditions.
On a pedigree:
- A circle represents a female
- A square represents a male
- A horizontal line connecting a male and female represents a marriage
- A vertical line and a bracket connect the parents to their children
- A circle/square that is shaded means the person **HAS** the trait.
- A circle/square that is not shaded means the person **does not have** the trait.
- Children are placed from oldest to youngest.
- A key is given to explain what the trait is.
Marriage

Male-DAD  Female-MOM

Has the trait

Male-Son  Female-daughter  Female-daughter  Male-Son

Oldest to youngest
Steps:
• Identify all people who have the trait.

• For the purpose of this class all traits will be given to you. In other instances, you would have to determine whether or not the trait is autosomal dominant, autosomal recessive, or sex-linked.

• In this example, all those who have the trait are homozygous recessive.

• Can you correctly identify all genotypes of this family?
  • F - Normal
  • f - cystic fibrosis

Key:
- affected male
- unaffected male
- affected female
- unaffected female
PKU
P- Unaffected
p- phenylketonuria

Key:           affected male                 affected female
unaffected male
unaffected female

PP or Pp
pp
Pp
pp
Pp
Pp or Pp
H-huntington’s disease
h-Unaffected

Key:  
- affected male
- unaffected male
- affected female
- unaffected female
To analyze chromosomes, cell biologists photograph cells in **mitosis**, when the chromosomes are fully condensed and easy to see (usually in **metaphase**).

The chromosomes are then arranged in homologous pairs.
Karyotypes

- The homologous pairs are then placed in order of descending size. The sex chromosomes are placed at the end.

- A picture of chromosomes arranged in this way is known as a karyotype.
The karyotype is a result of a haploid sperm (23 chromosomes) fertilizing a haploid egg (23 chromosomes).

The diploid zygote (fertilized egg) contains the full 46 chromosomes. (in humans)
Normal Human Male Karyotype: 46,XY
Normal Human Female Karyotype: 46,XX
Labeling a Karyotype

● To label a karyotype correctly, first list the number of chromosomes found in the karyotype. Ex. 46

- Normal Human Female: 46, XX

- Normal Human Male: 46, XY

● Secondly, list the type of sex chromosomes found in the karyotype. Ex. XX

● Lastly, list any abnormalities at the appropriate chromosome number.
What are abnormalities?

- Sometimes, during meiosis, things go wrong.
- The most common error is **nondisjunction**, which means “not coming apart”.
- If **nondisjunction** occurs, abnormal numbers of chromosomes may find their way into gametes, and a disorder of chromosome numbers may result.
Autosomal Chromosome Disorders

- Two copies of an autosomal chromosome fail to separate during meiosis, an individual may be born with THREE copies of a chromosome.

- This is known as a “Trisomy”

- Trisomy 13, Trisomy 18, Trisomy 21.
Down Syndrome

- Most common, Trisomy 21 (also known as Down Syndrome)
- 1 in 800 babies born in U.S. with Trisomy 21.
- Mild to severe mental retardation
- Increased susceptibility to many diseases and a higher frequency of other birth defects.

Male: 47, XY, +21
Female: 47, XX, +21
Sex Chromosome Disorders

- Turner’s Syndrome (nondisjunction)
  - Female inherits only one X chromosome
  - Karyotype: 45, X
  - Women are sterile, sex organs do not develop at puberty.

- Klinefelter’s Syndrome (nondisjunction)
  - Males receive an extra X chromosome
  - Karyotype: 47, XXY
  - The extra X chromosome interferes with meiosis and prevents individuals from reproducing.
Other Genetic Disorders

- **Sickle Cell Disease**
  - Characterized by the bent and twisted shape of the red blood cells.
  - More rigid and get stuck in capillaries. Blood stops flowing and can damage cells, tissues, and organs.
  - Produced physical weakness and damage to the brain, heart, and spleen...could be fatal.
  - Most commonly found in African Americans (can be linked to the incidence of malaria).
Other Genetic Disorders

- **Duchenne Muscular Dystrophy**
  - Sex-linked, defective gene for muscle protein.
  - Progressive weakening and loss of skeletal muscle.
  - In U.S., 1 out of every 3000 males born has condition.