- For example, if an allele is imprinted in a female and therefore silenced in the egg, then the new zygote will always only express the paternal allele received from the sperm for that trait

- Extranuclear genes found in mitochondria and chloroplasts are ALL inherited from your mother (from the egg cell; DNA is each is all maternal

Pedigree Analysis

- With long generation times and fewer offspring, human genetics is much more difficult to study
- Therefore, scientists collect information about a family's history and compile it into a family tree of sorts- a pedigree
- Study of a pedigree allows geneticists to determine if a trait is dominant or recessive, and also autosomal or sex-linked
- Pedigrees can also be used to predict genotypes of individuals
- Therefore, they are useful in predicting the outcomes of a future mating event
- Illustrate phenotypes
- Dominant: all generations, usually more than half the people, tt is unshaded
- Recessive: Skips generation(s), usually fewer people shaded, tt is shaded
- Find an intermediate family and analyze them
- In the case of sex-linked vs autosomal genes, there are a few specifics to remember and look for...
    - Most sex-linked genes are more specifically X-linked
    - Men only have one X, which they have to get from mom
        - Y comes from dad
    - Women have two X's, one from mom and one from dad
    - If a male has an X-linked trait...
        - NO sons will inherit it-not possible (give Y)
        - ALL daughter will at least be carriers
    - If a female is carrying an X-linked trait (just one)
        - All sons and daughters have a 50% chance of inheriting it
    - If a female is homozygous for an X-linked trait (both X's)...
        - ALL sons will inherit the trait
        - ALL daughters will at least be carriers

Catalog of Inheritable Disorders

- Autosomal recessive disorders:
    - Tay-Sachs Disease
        - Brain cells lack functional enzyme that normally metabolizes certain lipids in lysosome
        - Buildup of lipids causes seizures, blindness, brain degeneration, and death in just a couple of years
        - Common in interbreeding communities (Ashkenazic Jews-1 in 36,000...1 in 27 carrier... 100x greater than other populations)
        - Related mating leads to an increased chance of getting autosomal recessive

        - This one is actually somewhat incompletely dominant-heterozygote does have some deficit in the enzyme, but not enough to cause symptoms
    - Cystic fibrosis
        - Malfunctioning Cl- transporter in lung and digestive surfaces leads to buildup of thick mucus
        - Leads to digestive issues, breathing issues, and recurrent bacterial infections
        - 1 in 2500 of European descent, 1 in 25 carrier
        - Untreated live 0-5 years, treated live to 20's or 30's
    - Sickle-cell disease
        - Mutation in Hb gene leads to rods of hemoglobin that deform red blood cells
        - Affect O2 carrying capacity, plus may clump and clog small blood vessels
        - 1 in 400 African Americans, maybe 1 in 10 carrier
        - Also somewhat incompletely dominant-heterozygote is said to have "sickle cell trait"
        - Carrying one genes provides resistance to malarial parasite (Immunity to malaria; place with malaria have higher number of people with it)
- Autosomal dominant disorders
    - Most are lethal, particularly when homozygous
    - Maybe 10 total; very rare
    - Achondroplasia
        - Dwarfism caused by malformation of cartilage and bone
        - 1 in 25,000 people
        - 99.99% of the population is homozygous recessive
        - Normal height people are dd
    - Huntington's Disease
        - Case of a lethal dominant allele that can pass on because it causes death at an advanced age
        - Degenerative disorder of the nervous system with no phenotypic symptoms until middle age
        - 1 in 10,000 people
- X-linked disorders
    - Generally recessive, so seen almost exclusively in men
        - Female require two "broken" X's, while men only need one
    - Colorblindness
    - Duchenne muscular dystrophy
        - 1 in 3500 males in US
        - Lack of muscle protein dystrophin leads to progressive weakening of muscles, loss of coordination, and death in 20's
    - Hemophilia
        - Absence of protein(s) from the clotting cascade
        - "Royal Disease"
- <u>Chromosomal Abnormalities</u> can be the result of:
    - Abnormal chromosome number