Genetics Disorders

- Many disorders in humans are genetic in origin and follow Mendel’s laws of inheritance.
- These genetic disorders are often controlled by a single pair of alleles.

A karyotype is simply a picture of a person’s chromosomes. In order to get this picture, the chromosomes are isolated, stained, and examined under the microscope. Most often, this is done using the chromosomes in the white blood cells. A picture of the chromosomes is taken through the microscope.
Amniocentesis is a medical procedure used in prenatal diagnosis of chromosomal abnormalities and fetal infections, and also used for sex determination in which a small amount of amniotic fluid, which contains fetal tissues, is sampled from the amniotic sac surrounding a developing fetus, and the fetal DNA is examined for genetic abnormalities. A needle is usually inserted through the mother’s abdominal wall, then through the wall of the uterus, and finally into the amniotic sac. With the aid of ultrasound-guidance, a physician extracts approximately 20ml of amniotic fluid. If used for prenatal genetic diagnosis, then fetal cells are separated from the extracted sample. The cells are grown in a culture medium, then fixed and stained. Under a microscope the chromosomes are examined for abnormalities.

- Karyotyping and biochemical tests of fetal cells and molecules can help people make reproductive decisions.
- Fetal cells can be obtained through (1) amniocentesis...

Chorionic villus sampling (CVS), sometimes called “chorionic villous sampling” (as “villous” is the adjectival form of the word “villus”), is a form of prenatal diagnosis to determine chromosomal or genetic disorders in the fetus. It entails sampling of the chorionic villus (placental tissue) and testing it for chromosomal abnormalities, usually with FISH (fluorescence in situ hybridization is a cytogenetic technique used to staining DNA) or PCR (polymerase chain reaction is a technology in molecular biology used to amplify DNA samples). CVS usually takes place at 10–12 weeks’ gestation, earlier than amniocentesis or percutaneous umbilical cord blood sampling. It is the preferred technique before 15 weeks.

- …and (2) Chorionic villus.

Ultrasound is an oscillating sound pressure wave with a frequency greater than the upper limit of the human hearing range. Ultrasound can be used for medical imaging. Ultrasound has been used by radiologists and sonographers to image the human body for at least 50 years and has become a widely used diagnostic tool. The technology is relatively inexpensive and portable, especially when compared with other techniques, such as magnetic resonance imaging (MRI) and computed tomography (CT). Ultrasound is also used to visualize fetuses during routine and emergency prenatal care. Such diagnostic applications used during pregnancy are referred to as obstetric sonography. As currently applied in the medical field, properly performed ultrasound poses no known risks to the patient.

- Ultrasound is also a very helpful diagnostic tool for examining a fetus.

Most genetic disorders are caused by autosomal recessive alleles, i.e. cystic fibrosis and sickle-cell disease.

Some genetic disorders are caused by autosomal dominant disorders, i.e. achondroplasia and Huntington’s disease.

A pedigree shows the pattern of inheritance for a particular condition; genetic counselors can use a pedigree to determine whether a condition is dominant or recessive. Consider the two possible patterns of inheritance above. In pattern I, the child is affected, but neither parent is; this can happen if the condition is recessive and both parents are Aa. Notice that the parents are carriers, because they appear normal (do not express the trait) but are capable of having a child with the genetic disorder. In pattern II, the child is unaffected, but the parents are affected. This can happen if the condition is dominant and the parents are Aa.
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- **Family pedigrees** are used to determine patterns of inheritance and individual genotypes.

Pedigree showing inheritance of deafness in a family from Martha’s Vineyard. In the pedigree, squares represent males, and circles represent females; colored symbols here indicate deafness. The earliest generation studies is at the top of the pedigree. Notice that deafness did not appear in this generation and that it showed up in only two of the seven children in the generation at the bottom. By applying Mendel’s principles, we can deduce that the deafness allele is recessive.

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<tr>
<th>Generation</th>
<th>Individuals</th>
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How would you know that the individual at the asterisk is heterozygous?

- **Some genetic disorders** are caused by alleles that are located on the sex chromosomes, i.e., Hemophilia and red-green color blindness.

- The X sex chromosome is larger than the Y and therefore carries many alleles that have little to do with gender.

- The inheritance of X-linked genes follow special rules.

Genetics Disorders

- A male receives a single X chromosome from his mother and therefore will have the X-linked disorder if inherited.

- A female receives two X chromosomes and therefore would need to receive the X-linked disorder from both parents.

Sex-linked genes exhibit a unique pattern of inheritance. In fruit flies eye color is a sex-linked characteristic. The inheritance pattern of this gene reflects the fact that males have one X chromosome and females have two. These figures illustrate inheritance patterns for white eye color (r) in the fruit fly, an X-linked recessive trait.
Color blindness, or color vision deficiency, is the inability or decreased ability to see color, or perceive color differences, under normal lighting conditions. The most usual cause is a fault in the development of one or more sets of retinal cones that perceive color in light and transmit that information to the optic nerve. This type of color blindness is usually a sex-linked condition. The genes that produce photopigments are carried on the X chromosome; if some of these genes are missing or damaged, color blindness will be expressed. Color blindness will be expressed in males with a much higher probability than in females because males only have one X chromosome.

Hemophilia is a bleeding disorder that slows the blood clotting process. A high incidence of hemophilia has plagued the royal families of Europe. The two major forms of hemophilia occur much more commonly in males than in females. Hemophilia A is the most common type of the condition occurring in 1 in 4000 to 1 in 5000 males worldwide. Hemophilia B occurs approximately 1 in 20,000 newborn males worldwide. Both Hemophilia A and hemophilia B are inherited in an X-linked recessive pattern. Hemophilia is less common in females, because they would need to receive two altered copies of this gene to exhibit the disorder.