1. Human Genetics
   1. The study of \_\_\_\_\_ as it occurs in human beings.
   2. Human genetics encompasses a variety of overlapping fields including: classical genetics, cytogenetics, molecular genetics, biochemical genetics, genomics, population genetics, developmental genetics, clinical genetics, and genetic counseling.
   3. The development of human genetics has been limited because of
   4. \_\_\_\_\_,
   5. limited \_\_\_\_\_,
   6. lack of ability to select \_\_\_\_\_,
   7. ~18 years between generations,
   8. \_\_\_\_\_ influences.
   9. Genetic Traits in Humans can be tracked through Family \_\_\_\_\_
2. The inheritance of human traits follows \_\_\_\_\_ laws.
3. A Pedigree
4. shows the inheritance of a trait in a family through multiple \_\_\_\_\_
5. demonstrates \_\_\_\_\_ or \_\_\_\_\_ inheritance, and
6. can also be used to deduce \_\_\_\_\_ of family members.
7. Symbols and Rules
   * 1. \_\_\_\_\_ =
     2. \_\_\_\_\_ =
     3. \_\_\_\_\_ =
8. How to Read a Pedigree Chart
9. \_\_\_\_\_: row
10. \_\_\_\_\_: squares
11. \_\_\_\_\_: circles
12. Horizontal lines between circle and square: \_\_\_\_\_
13. Vertical lines: \_\_\_\_\_ of a mating
14. Different colors or shading are used to show individuals with and without a trait.
    1. Human Health Often Relates to Genetics
15. There are major aspects related to personal health
16. physical, emotional, social, spiritual, vocational, and intellectual.
17. In order to be considered “well” or “\_\_\_\_\_”, it is imperative that none of these areas be neglected.
18. For the scope of this course, we will confine our study to genetically related diseases.
19. \_\_\_\_\_ have been developed to detect genetic diseases or deficiencies before and after birth.
20. Technologies provide Insight into one’s Genetic \_\_\_\_\_ & Genetically Related Diseases
21. \_\_\_\_\_ TESTS on the mother at 15–20 weeks of pregnancy can help identify fetuses at risk for certain birth defects.
22. \_\_\_\_\_ \_\_\_\_\_ enables a physician to examine a fetus directly for anatomical \_\_\_\_\_.
    1. The most common procedure is \_\_\_\_\_ Imaging, using sound waves to produce a picture of the fetus.
23. \_\_\_\_\_ \_\_\_\_\_ can detect diseases that can be prevented by special care and precautions.
24. New technologies raise \_\_\_\_\_ considerations that include
25. the \_\_\_\_\_ and potential use of results of genetic testing,
26. time and financial \_\_\_\_\_, and
27. the determination of what, if anything, should be done as a result of the testing.
28. Modern technologies offer ways to obtain genetic information
29. before \_\_\_\_\_, \_\_\_\_\_ pregnancy, \_\_\_\_\_ birth
30. Genetic Testing can identify prospective parents who are heterozygous carriers for certain diseases.
31. Certain technologies can be used for detecting genetic conditions in a fetus:
    1. \_\_\_\_\_ extracts samples of amniotic fluid containing fetal cells and permits:
32. \_\_\_\_\_ to detect chromosomal abnormalities such as Down syndrome
33. biochemical tests on cultured fetal cells to detect other conditions
    1. Human Genetic Patterns of Inheritance
34. Different patterns of inheritance are observed depending on the condition:
35. \_\_\_\_\_ \_\_\_\_\_ Inheritance (Sickle Cell, Phenylketonuria, Cystic Fibrosis)
36. \_\_\_\_\_ \_\_\_\_\_ Inheritance (Huntington’s, Achondroplasia, Aneuploidy)
37. \_\_\_\_\_-\_\_\_\_\_ inheritance (e.g. hemophilia, pattern baldness)
38. How do these patterns get diagnosed or discovered?
    * 1. A Prepared Human \_\_\_\_\_.
    1. In this arrangement, the chromosomes are organized like a police line-up.
    2. Where are the \_\_\_\_\_ chromosomes?
       1. \_\_\_\_\_ of \_\_\_\_\_ and
       2. \_\_\_\_\_ \_\_\_\_\_ chromosomes (\_\_\_\_\_ and \_\_\_\_\_)
    3. The sex chromosomes determine the gender.
       1. Males (\_\_\_\_\_)
       2. Females (\_\_\_\_\_)
       3. A karyotype is an \_\_\_\_\_ that \_\_\_\_\_ the chromosomes and allows the detection of \_\_\_\_\_.
39. Three Methods of Karyotyping
    * + 1. \_\_\_\_\_ Method of Karyotyping
           1. The blood culture is \_\_\_\_\_ (spun at a high speed) to \_\_\_\_\_ the blood cells from the culture fluid (blood plasma).
    1. The fluid is discarded and a hypotonic solution is mixed with the cells. This makes the \_\_\_\_\_ blood cells (RBC’s) swell and burst. The \_\_\_\_\_ blood cells (WBC’s) swell but do not burst, and their chromosomes spread out.
    2. Another centrifugation step separates the swollen WBC’s. The fluid containing the remnants of the RBC’s is poured off. A \_\_\_\_\_ (a preservative) is mixed with the WBC’s. A drop of the cell suspension is spread on a microscope slide, dried and stained.
    3. The slide with the WBC’s DNA is viewed with a microscope, and the images of the \_\_\_\_\_ chromosomes are sorted by \_\_\_\_\_ and \_\_\_\_\_ on a computer.
    4. The resulting display is the karyotype.
       1. The 46 chromosomes found indicates:
          1. \_\_\_\_\_ of autosomes and
          2. \_\_\_\_\_ chromosomes: X and Y.
       2. Each of the chromosomes consist of two sister chromatids lying close together.
       3. \_\_\_\_\_ \_\_\_\_\_
          * 1. Another karyotyping method uses different \_\_\_\_\_ to color the \_\_\_\_\_.
            2. The chromosomes are matched by comparing:
          1. \_\_\_\_\_ (length)
          2. \_\_\_\_\_ (stripe pattern)
       4. \_\_\_\_\_ Karyotying Method
40. The \_\_\_\_\_ process (\_\_\_\_\_ karyotyping) dyes the chromosomes different colors for ease of matching.
41. SKY adds \_\_\_\_\_ to the sorting process.
42. Sorting Chromosomes
    * 1. The \_\_\_\_\_ is the small section of \_\_\_\_\_ on the \_\_\_\_\_ of a chromosome.
      2. Every time the chromosome duplicates the telomere slightly \_\_\_\_\_.
      3. When the telomere completely disappears, the chromosome can no longer \_\_\_\_\_**.**
    1. Genetic Disorders
43. Overview
    * 1. Many \_\_\_\_\_ have a \_\_\_\_\_ aspect. Some, including many cancers, are caused by a mutation in a gene or group of genes in a person's cells.
      2. These \_\_\_\_\_ can occur randomly or because of an environmental exposure such as cigarette smoke.
      3. Other genetic disorders are \_\_\_\_\_. A mutated gene is passed down through a family and each generation of children can inherit the \_\_\_\_\_ that causes the disease.
      4. Still other genetic disorders are due to problems with the number of packages of genes called chromosomes.

* In Down syndrome, for example, there is an extra copy of chromosome 21.

1. Commonly used terms
2. Affected persons have a \_\_\_\_\_ or condition.
3. Carriers do not have a genetic disease, but carry the \_\_\_\_\_ which causes the disease
   1. They have one \_\_\_\_\_ allele + one \_\_\_\_\_ for a disease.
   2. There is \_\_\_\_\_ effect of the recessive allele on a carrier, because it has a normal/dominant allele that is expressed.
4. Unaffected persons do not have the \_\_\_\_\_ and do not \_\_\_\_\_ for it either.

C. Many Inherited Traits in Humans are controlled by a \_\_\_\_\_ Gene

1. Because a trait is dominant does NOT mean that it is “\_\_\_\_\_” or more common than a recessive trait.

2. \_\_\_\_\_-type Traits are those most often seen in nature and not necessarily specified by dominant alleles.

1. These genes are all located on \_\_\_\_\_.
   1. Autosomal \_\_\_\_\_ Traits
2. Genes located on Autosomes control autosomal traits and disorders.
   * 1. Thousands of human genetic disorders — ranging in severity from relatively mild, such as \_\_\_\_\_, to invariably fatal, such as \_\_\_\_\_ \_\_\_\_\_ — are inherited as recessive traits.
     2. Most people who have recessive disorders are born to \_\_\_\_\_ parents who are both \_\_\_\_\_, \_\_\_\_\_ of the recessive allele for the disorder, but are phenotypically normal.
3. \_\_\_\_\_ Cell Anemia
4. Transmitted in an autosomal recessive fashion.
5. Most common inherited disease seen in individuals of \_\_\_\_\_ descent.
6. This disease affects the type of \_\_\_\_\_ produced and the \_\_\_\_\_ of red blood cells.
7. Caused by a mutation in one of the proteins that makes up hemoglobin.
8. This is a substitution mutation in one of the protein chains of hemoglobin.
9. This results in the amino acid valine being inserted into this chain instead of the normal glutamic acid.
10. This replacement of glutamic acid with valine results in a hemoglobin molecule with abnormal structure distorting the shape of the \_\_\_\_\_ blood cells.
11. As a result hemoglobin cannot carry \_\_\_\_\_ properly.
12. \_\_\_\_\_ Advantage
13. The condition of the heterozygous form being \_\_\_\_\_ against some disease or illness.
14. This is the protection that the heterozygous condition can give to people who are \_\_\_\_\_ of a \_\_\_\_\_ allele.
15. Example: \_\_\_\_\_ Cell Disease and \_\_\_\_\_. Carriers of sickle-cell disease have increased resistance to malaria.
16. \_\_\_\_\_ (PKU)
17. Transmitted in an \_\_\_\_\_ fashion
18. Inability to break down the amino acid \_\_\_\_\_.
19. Enzyme which breaks down the amino acid \_\_\_\_\_ is defective.
20. Caused by a mutated gene which code for this enzyme.
21. Therefore, phenylalanine builds up in the brain, causing \_\_\_\_\_ and other problems.
22. Requires the \_\_\_\_\_ of this amino acid from the diet.
23. Cystic Fibrosis –
24. Transmitted in an \_\_\_\_\_ fashion.
25. Single most common inherited disease among \_\_\_\_\_.
26. Results in increased (thick) airway and digestive tract \_\_\_\_\_ that causes \_\_\_\_\_ problems and recurrent \_\_\_\_\_; Lethal.
27. Caused by a defective membrane chloride channel protein.
28. Tay Sachs Disease –
29. Nervous system destruction due to the absence of an enzyme (hexosaminidase A) needed to break down lipids necessary for normal brain function.
30. Early onset and common in infantile form results in blindness, seizures, paralysis, and early death.
    1. Autosomal \_\_\_\_\_ Traits
       * 1. Based on a dominant allele
       1. Human disorders include
31. \_\_\_\_\_ Disease, a degenerative disorder of the \_\_\_\_\_ system and
32. Achondroplasia, a form of \_\_\_\_\_ in which the head and torso of the body develop normally butthe arms and legs are short.
    * 1. Congenital Genetic Diseases
33. Disease a child is \_\_\_\_\_ with due to a mutation in the DNA.
34. May be a Point or Chromosomal Mutation and based on \_\_\_\_\_ allele in chromatid pair.
35. Many are called \_\_\_\_\_ because the chromosome abnormalities result in common problems associated with the specific chromosomal mutation.
36. There are many examples of dominant allele genetic disorders called: \_\_\_\_\_.
    * 1. autosomal dominance
37. Individuals have one \_\_\_\_\_ or \_\_\_\_\_ chromosome
38. (2*n* + 1 or 2*n* - 1)
39. Caused by \_\_\_\_\_-\_\_\_\_\_ during meiosis
40. Major cause of human reproductive failure
41. Most human miscarriages are aneuploids

C. \_\_\_\_\_ Syndrome

1. Trisomy of chromosome \_\_\_\_\_
2. \_\_\_\_\_ impairment and a variety of additional defects.
3. Can be detected before birth.
4. Risk of Down syndrome increases dramatically in mothers over age \_\_\_\_\_.

D. \_\_\_\_\_ Syndrome

1. Inheritance of only one X (\_\_\_\_\_)
2. 98% spontaneously miscarried
3. Survivors are short, infertile females
   1. No functional \_\_\_\_\_
   2. Secondary sexual traits reduced
   3. May be treated with hormones, surgery

E. \_\_\_\_\_ Syndrome

1. \_\_\_\_\_ condition
2. Results mainly from nondisjunction in mother (67%)
3. Phenotype is tall males
4. \_\_\_\_\_ or nearly so
5. \_\_\_\_\_ traits (sparse facial hair, somewhat enlarged breasts)
6. Treated with testosterone injections
   1. Sex-linked Traits
      * 1. Sex-linked traits are produced by genes only on the sex chromosomes (gametes).
        2. They can be Dominant or Recessive.
      1. A = dominant a = recessive
      2. What would be the genotypes of a male and female that have a Sex-linked Dominant trait and do not express the trait?
         1. Expresses Trait: Male - XA Y Female - XA XA or XA Xa
         2. No Expression: Male - Xa Y Female - Xa Xa
      3. What would be the genotypes of a male and female that have a Sex-linked Recessive trait and do not express the trait?
         1. Expresses Trait: Male - Xa Y Female - Xa Xa
         2. No Expression: Male - XA Y Female - XA XA or XA Xa (Carrier)
      4. Most Sex-linked traits are Recessive.