MAIN IDEA: THE INHERITANCE OF A TRAIT OVER SEVERAL GENERATIONS CAN BE SHOWN AS A PEDIGREE

OBJECTIVE 1: DESCRIBE HUMAN GENETIC DISORDERS THAT ARE CAUSED BY INHERITANCE OF RECESSIVE ALLELES

A. Most genetic disorders are caused by recessive alleles and in order to be expressed, the person would need to be homozygous recessive.
B. An individual who is heterozygous for a recessive disorder is called a carrier.
C. Many of these alleles are rare, but a few are common in certain ethnic groups. (see page 297, Table 11.2)
D. Punnett squares can be used to calculate the chance that offspring will be born with some of these genetic disorders.
E. Cystic fibrosis
   1. The most common genetic disorder among white Americans.
   2. About 1 in 20 are carriers and 1 in 3500 children born to white Americans inherits the disorder.
   3. Defective protein in the plasma membrane results in the formation and accumulation of thick mucus in the lungs and digestive tract.
   4. Life expectancy is mid-twenties.
F. Albinism
   1. Genes do not produce the normal amount of pigment called melanin.
   2. Results in no color to skin, hair, and eyes (eyes are blue)
   3. Associated with vision problems and skin is highly susceptible to UV damage.
   4. There is no cure, but life span not affected.
G. Tay-Sachs
   1. Recessive disorder of the central nervous system in which there is an absence of an enzyme that normally breaks down a lipid produced and stored in tissues of the CNS. Causes accumulation of lipids in the cells.
   2. Life expectancy is approximately five years.
   3. Common in the U.S. among the Amish and those with Eastern European Jewish ancestors.
A. Galactosemia
   1. Recessive alleles cause the absence of an enzyme that breaks down galactose.
   2. Accumulation of galactose causes enlarged liver, kidney damage and mental disabilities.
   3. There is no cure but those that are homozygous recessive should limit their intake of lactose and galactose in the diet by avoiding milk products.

OBJECTIVE 2: DESCRIBE HUMAN GENETIC DISORDERS THAT ARE CAUSED BY THE INHERITANCE OF SINGLE DOMINANT ALLELE

A. Many traits are inherited just as the rule of dominance predicts
   1. Tongue rolling, hanging earlobes, almond shaped eyes, and thick lips
B. Some disorders are caused by a single dominant allele.
C. Huntington’s Disease is a lethal genetic disorder caused by a rare dominant allele.
   1. Occurs in 1 in 10,000 people in the U.S.
   2. Results in the breakdown of certain areas of the brain causing loss of brain function, uncontrollable movements, and emotional disturbances.
   3. Occurs in people between ages of 30-50, after they may have already had children and passed along the dominant allele.
   4. There is a test to determine if you are a carrier.….if positive you can decide if you want to risk having children…. you will also know that you will most likely develop the disease.
D. Another trait inherited by a single dominant allele is a form of dwarfism called achondroplasia.
   1. Occurs in 1, 25000 people in the U.S. and the gene that affects normal bone growth is dominant instead of recessive!
   2. People with achondroplasia have a small body size and limbs that are comparatively short. Most will have an adult height of four feet and an average life span.
   3. 75% of the people with achondroplasia are born to parents of average size. SO…. 
      a. In a dominant genetic condition, the genotype is seen as the phenotype
      b. When children with achondroplasia are born to parents of average height, the conclusion is that the condition occurred because of a new mutation or genetic change.
OBJECTIVE 3: DESCRIBE AND INTERPRET A PEDIGREE
A. A pedigree maps the inheritance of genetic traits from generation to generation.
B. A pedigree uses a set of symbols to identify gender, individuals affected by the trait being studied, and family relationships through several generations.
   1. circle - female
   2. square - male
   3. shaded symbols – individuals showing the trait being studied.
   4. Unshaded symbols – individuals not showing the specific trait
   5. Half-shaded symbol – carriers, heterozygous individuals
   6. Horizontal line – connects parents
   7. Vertical line – connects parents to their offspring
C. Each horizontal row designates a generation, with the most recent row at the bottom.
E. Pedigrees are used to infer genotypes from the observations of phenotypes.
   1. By knowing physical traits, genealogists can determine what genes an individual is most likely to have.
   2. Pedigrees can help genetic counselors determine whether inheritance patterns are dominant or recessive.
   3. When recessive traits are expressed, the ancestry of the person expressing the trait is followed for several generations to determine which parents or grandparents were carriers of the recessive allele.
F. If good records have been kept within families, disorders in future offspring can be predicted. However, more accuracy can be expected if several individuals within the family can be evaluated.

MAIN IDEA: COMPLEX INHERITANCE OF TRAITS DOES NOT FOLLOW INHERITANCE PATTERNS FOLLOWED BY MENDEL
OBJECTIVE 4: DISTINGUISH BETWEEN INCOMPLETELY DOMINANT AND CODOMINANT ALLELES AND GIVE EXAMPLES OF CODOMINANCE IN HUMANS
A. Many inheritance patterns are more complex than those studied by Mendel.
   1. Most traits are not simply dominant or recessive.
B. Incomplete Dominance
   1. Neither trait is dominant over the other, the traits appear to blend
   2. A red flower is crossed with a white flower makes a pink flower.
   3. The heterozygous genotype has a phenotype different than either homozygous dominant or homozygous recessive.

4. Cross two heterozygotes:

   |   R   |   R'  |
---|------|------|
   R |      |      |
   R' |      |      |

Genotypic ratio =
Phenotypic ratio =

C. Codominance
   1. Both alleles of the pair are expressed, but in different places. Some dogs have brown and white hairs.
   2. Causes the phenotypes of both alleles to be produced in heterozygous individuals; both alleles are expressed equally.
   3. An example of codominance in humans is sickle-cell anemia. It is a blood disorder with abnormal hemoglobin (the oxygen carrying protein) that causes red blood cells to deform and block small blood vessels. This blockage causes pain and tissue damage. Sickle-shaped cells do not survive as long as normal red blood cells so the person is anemic.
   a. Individuals that are heterozygous for the allele produce both normal and sickle-celled hemoglobin. Symptoms are not as severe as those that are homozygous.
   b. The sickle cell allele increases resistance to malaria
OBJECTIVE 5: COMPARE MULTIPLE ALLELIC AND EPISTASIS INHERITANCE AND BE ABLE TO GIVE EXAMPLES OF EACH

A. It is common for more than two alleles to control a trait in a population.

B. Traits controlled by more than two alleles have **multiple alleles**

1. Example: Four alleles of a single gene govern fur color of rabbits resulting in ten possible genotypes and four phenotypes. (see page 304, figure 11.7)

2. Human blood types are determined by the absence or presence of certain proteins on the surface of red blood cells, and there are three alleles.

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<tr>
<th>GENOTYPES</th>
<th>SURFACE PROTEIN</th>
<th>PHENOTYPE</th>
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<tbody>
<tr>
<td>I^AIA^A or I^Ai</td>
<td>A</td>
<td>A</td>
</tr>
<tr>
<td>I^BI^B or I^Bi</td>
<td>B</td>
<td>B</td>
</tr>
<tr>
<td>I^AIl</td>
<td>A and B</td>
<td>AB</td>
</tr>
<tr>
<td>ii</td>
<td>none</td>
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3. Determining blood type is necessary before receiving transfusions: Different surface proteins will cause the RBC’s to clot.

4. Determining blood type may be useful in determining parentage but it cannot prove with certainty man is definitely the father; only DNA testing can determine actual parentage.

a. Who could be the father of baby Jane? Jane is type O, her mother is type A and her two possible fathers are Jim (type B) or John (type AB). Prove your answer in a punnett square.

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Genotypic ratio =

Phenotypic ratio =

C. **Epistasis** is an interaction in which one allele hides the affects of another allele as in coat color of Labrador retrievers. (see page 305, figure 11.8)

1. Coat color is controlled by two sets of alleles:
   a. The dominant allele E determines whether the fur will have dark pigment (E_bb, E_B_)
   b. Alleles ee cause no pigmentation.
   c. Allele B determines how dark the pigment will be

OBJECTIVE 6: DETERMINE THE SEX OF AN ORGANISM BY ITS CHROMOSOMES

A. Humans have 46 chromosomes or 23 homologous pairs

B. 22 of the matching pairs are called **autosomes**.

C. The 23rd pair differ in males and females and are called the **sex chromosomes**.

1. XX = female
2. XY = male

D. Because males produce two kinds of gametes during meiosis, either X or Y, males determine the sex of the offspring.

1. Do a punnet square to determine the ratio of expected males to females after fertilization.

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<td>X</td>
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<td>X</td>
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Genotypic ratio =

Phenotypic ratio =

2. Because fertilization is governed by probability, the ratio usually is not exactly 1:1 in a small population.
OBJECTIVE 7: EXPLAIN DOSAGE COMPENSATION AND ITS RELATIONSHIP TO CHROMOSOME ACTIVATION
A. X chromosomes are larger than Y chromosomes and the X chromosome carries genes necessary for the development of both females and males. The Y chromosome has genes mainly for male development.
B. Because females have two X chromosomes and males have just one, to balance the difference in the dose of X-related genes, one of the X chromosomes stops working in the females body cells.
   1. This is called dosage compensation or X-inactivation
   2. Inactivation is a completely random event and occurs in all mammals.
C. Chromosome inactivation results in different coat colors of a calico cat.
D. The inactivated X chromosomes can be observed in cells. Darkly stained, inactivated X chromosomes are called Barr bodies and are seen as a condensed, darkly stained structure in the nucleus. Only females will have Barr bodies in their nucleus.

OBJECTIVE 8: BE ABLE TO EXPLAIN WHAT A SEX-LINKED TRAIT IS AND DISCUSS ITS IMPACT ON THE MALE POPULATION
A. Sex-linked traits are those traits controlled by genes located on a sex chromosome
   1. In humans it usually is on the X chromosome and is recessive
   2. Because the X and the Y are not homologous, the Y chromosome has no corresponding allele to one on the X chromosome.
B. Males have only one X chromosome so even if their defective allele is recessive they will have the trait. Females could have a normal second X and only be a “carrier” for the trait; they have to have two defective alleles in order for the sex-linked trait to be expressed.
C. Sex-linked traits in humans are certain forms of color blindness (red/green) see page 307, hemophilia (see page 308), one form of muscular dystrophy, and fragile X syndrome.

OBJECTIVE 9: DESCRIBE POLYGENIC INHERITANCE AND GIVE SOME EXAMPLES
A. Polygenic inheritance is the inheritance pattern of a trait that is controlled by two or more genes. (see page 309)
   1. Genes may be on the same or on different chromosomes. (RRTtSsQq)
   2. Examples are eye color, skin color, height, or size/color of fruit
   3. The phenotypes of these traits will show a continuous range of variability from the minimum value of the trait to the maximum. Results in a bell-shaped curve. (see page 326)
      a. All heterozygotes are intermediary and all alleles represented by an upper case letter contributes a small, but equal, portion to the trait being expressed.

OBJECTIVE 10: SUMMARIZE HOW INTERNAL AND EXTERNAL ENVIRONMENTS AFFECT GENE EXPRESSION
A. The genetic makeup of an organism at fertilization determines only the organism’s potential to develop and function.
B. As the organism develops, many factors can influence how the gene is expressed, or even whether it is expressed at all.
C. Genes interact with each other and the environment to form a complete picture of inheritance.
   1. individuals known to have a particular gene fail to express the phenotype specified by that gene.
D. External environmental influences include temperature, nutrition, light, water, chemicals, and infectious agents.
   1. Example: leaves may have different color and shape depending on how much light they receive
E. Internal environments of males and females are different because of hormones and structural differences.
   1. Example: feather color in peacocks are expressed differently in males and females because of hormones, which are determined by different sets of genes.
   2. Age also affects gene function as the internal environment changes as an organism ages.
F. By studying twins, scientists can better determine if inheritance patterns are influenced by genetic or environmental contributions.
   1. Traits that appear frequently in identical twins are at least partially controlled genetically.
   2. Presume traits that are expressed differently in identical twins are strongly influenced by environment.
   3. A percentage of twins that both express a given trait is called a concordance rate (see page 310). A large difference between fraternal twins and identical twins show a strong genetic influence.
MAIN IDEA: CHROMOSOMES CAN BE STUDIED USING KARYOTYPES

OBJECTIVE 11: DISTINGUISH NORMAL KARYOTYPES FROM THOSE WITH ABNORMAL NUMBERS OF CHROMOSOMES AND DEFINE AND DESCRIBE THE ROLE OF TELOMERES

A. Scientists study whole chromosomes by using images of chromosomes stained during metaphase.
B. The staining bands identify or mark identical places on homologous chromosomes.
C. The pairs of homologous chromosomes are arranged in decreasing size to produce a micrograph called a **karyotype**, (see page 311, figure 11.17) which is useful in pinpointing unusual chromosomes numbers and sizes.
D. Scientists have found that chromosomes end in protective caps called **telomeres**.
   1. Telomere caps consist of DNA associated with proteins.
   2. Telomeres might be involved with both aging and cancer.

OBJECTIVE 12: DESCRIBE NONDISJUNCTION AND ITS EFFECTS ON AUTOSOMAL AND SEX CHROMOSOMES IN HUMANS

A. Mistakes in meiosis, usually due to **nondisjunction**, may result in abnormal number of chromosomes. Autosomal or sex chromosomes can be affected. (see page 312, fig. 11.18)
   1. Nondisjunction can occur in Meiosis I, Meiosis II, or both.
   2. Trisomy of 21st pair – Down’s Syndrome (only autosomal trisomy in which affected individuals survive to adulthood.)
   3. X, XXX, XXY, XYY – any individual with at least one Y chromosome is male, and any individual without a Y is a female. Most lead normal lives, but are sterile and may suffer from varying degrees of mental retardation. (see page 314, Table 11.4)

OBJECTIVE 13: ASSESS THE BENEFITS AND RISKS OF DIAGNOSTIC FETAL TESTING

A. Tests for assessing the possibility of genetic and chromosomal disorders are available. (see page 315, Table 11.5)
B. Some of the available tests are amniocentesis, choronic villi sampling, and fetal blood sampling.
C. Common benefits include diagnosis of chromosome abnormalities and genetic defects.
D. Common risks include miscarriage and risk of infection.
E. When considering fetal testing the doctor would need to consider previous health problems of the mother and the health of the fetus.

SUMMARY: VARIATION OF TRAITS

A. Variation among individuals of the same species can be explained by both genetic and environmental factors.
B. Individuals within a species have similar, but not identical, genes.
C. In sexual reproduction, variations in traits between parent and offspring arise from the particular set of chromosomes (and their respective multiple genes) inherited, with each parent contributing half of a chromosome pair.
D. Genes along with environmental factors can modify an individual’s specific development, appearance, behavior and likelihood of producing offspring.
E. The set of variations of genes present, together with the interactions of genes with their environment, determines the distribution of variation of traits in a population.