Human genetics: pedigree analysis in human genetics

Cystic fibrosisA fatal recessive genetic disorder associated with abnormal secretions of the exocrine glands. Sickle cell anemiaA recessive genetic disorder associated with an abnormal type of hemoglobin, a blood transport protein. ONHUMAN GENETICS: PEDIGREE ANALYSIS IN HUMAN GENETICS SPECIFICALLY FOR YOUFOR ONLY$13. 90/PAGEOrder NowManfan syndromeAn autosomal dominant genetic disorder that affects the skeletal system, the cardiovascular system, and the eyes. X-linkedThe pattern of inheritance that results from genes located on the X chromosome. Y-linkedThe pattern of inheritance that results from genes located only on the Y chromosome. HemizygousA gene present on the X chromosome that is expressed in males in both the recessive and the dominant condition. HypophosphatemiaAn X-linked dominant disorder. Those affected have low phosphate levels in the blood and skeletal deformities. Color blindnessDefective color vision caused by reduction or absence of visual pigments. There are three forms: red, green, and blue blindness. Muscular dystrophyA group of genetic diseases associated with progressive degeneration of muscles. Two of these, Duchenne and Becker muscular dystrophy, are inherited as X-linked, recessive traits. Huntington diseaseAn autosomal dominant disorder associated with progressive neural degeneration and dementia. Adult onset is followed by death 10 to 15 years after symptoms appear. PorphyriaA genetic disorder inherited as a dominant trait that leads to intermittent attack of pain and dementia. Symptoms first appear in adulthood. PenetranceThe probability that a disease phenotype will appear when a disease-related genotype is present. ExpressivityThe range of phenotypes resulting from a given genotype. CamptodactylyA dominant human genetic trait that is expressed as immobile, bent little

fingers. XXXXXXXXXX End of Vocab: Start of READINGS/FACTS on Chapter 4: Pages 70-99. XXXXXXXXXX End of Vocab: Start of READINGS/FACTS on Chapter 4: Pages 70-99. Pedigree construction and analysis are basic methods in human genetics. Pedigree construction and analysis are basic methods in human genetics. A pedigree is an orderly presentation of family information, using standardized symbols. A pedigree is an orderly presentation of family information, using standardized symbols. Analysis of the pedigree using knowledge of Mendelian principles can determine whether the trait has a dominant or a recessive pattern of inheritance and whether the gene in question is located on an X or a Y chromosome or on one of the other 22 chromosomes (the autosomes). Analysis of the pedigree using knowledge of Mendelian principles can determine whether the trait has a dominant or a recessive pattern of inheritance and whether the gene in question is located on an X or a Y chromosome or on one of the other 22 chromosomes (the autosomes). Genetic disorders can be inherited in a number of different ways. We will consider six patterns of inheritance. Genetic disorders can be inherited in a number of different ways. We will consider six patterns of inheritance. Only males carry a Y chromosome. Only males carry a Y chromosome. Ehlers-Danlos syndrome, which is characterized by loose joints and easily stretched skin, can be inherited as an autosomal dominant, autosomal recessive, or X-linked recessive trait. Ehlers-Danlos syndrome, which is characterized by loose joints and easily stretched skin, can be inherited as an autosomal dominant, autosomal recessive, or X-linked recessive trait. A trait can have a single pattern of inheritance but be caused by mutation in any of several genes. A trait can have a single pattern of inheritance but be caused by mutation in any of

several genes. Porphyria, a metabolic disorder associated with abnormal behavior, is inherited as an autosomal dominant trait. It can also be caused by mutation in genes on chromosomes 1, 9, 11, and 14. Porphyria, a metabolic disorder associated with abnormal behavior, is inherited as an autosomal dominant trait. It can also be caused by mutation in genes on chromosomes 1, 9, 11, and 14. If the pattern of inheritance can be established, it can be used to predict genetic risk in several situations, including: pregnancy outcome, adult-onset disorders, and recurrence risks in future offspring. If the pattern of inheritance can be established, it can be used to predict genetic risk in several situations, including: pregnancy outcome, adult-onset disorders, and recurrence risks in future offspring. Recessive traits carried on autosomes have several distinguishing characteristics: Look on page 75. Recessive traits carried on autosomes have several distinguishing characteristics: Look on page 75. CF (Cystic fibrosis), affects the glands that produce mucus, digestive enzymes, and sweat. CF (Cystic fibrosis), affects the glands that produce mucus, digestive enzymes, and sweat. Almost all children with CF have phenotypically normal, heterozygous parents. Almost all children with CF have phenotypically normal, heterozygous parents. The CF gene encodes a protein called the cystic fibrosis transmembrane conductance regulator (CFTR), which is inserted in the plasma membrane of specific gland cells. The CF gene encodes a protein called the cystic fibrosis transmembrane conductance regulator (CFTR), which is inserted in the plasma membrane of specific gland cells. CFTR, regulates the flow of chloride ions across the plasma membrane. CFTR, regulates the flow of chloride ions across the plasma membrane.

Sickle cell anemia in certain populations is related to the frequency of

malaria. Sickle cell heterozygotes are more resistant to malaria than are homozygous normal individuals. Sickle cell anemia in certain populations is related to the frequency of malaria. Sickle cell heterozygotes are more resistant to malaria than are homozygous normal individuals. In autosomal dominant disorders, heterozygotes and those with homozygous dominant genotype have an abnormal phenotype. In autosomal dominant disorders, heterozygotes and those with homozygous dominant genotype have an abnormal phenotype. Dominant traits have a distinctive pattern of inheritance: Look on page 81. Dominant traits have a distinctive pattern of inheritance: Look on page 81. The most dangerous effects of Marfan syndrome are on the cardiovascular system, especially the aorta. The aorta is the main blood-carrying vessel in the body. The most dangerous effects of Marfan syndrome are on the cardiovascular system, especially the aorta. The aorta is the main blood-carrying vessel in the body. The gene responsible for Marfan syndrome is located on chromosome 15. The normal product of the gene is a protein called fibrillin, which is part of connective tissue. The gene responsible for Marfan syndrome is located on chromosome 15. The normal product of the gene is a protein called fibrillin, which is part of connective tissue. Human females have two X chromosomes, and males have an X chromosome and a Y chromosome. These chromosomes are called sex chromosomes because they play major roles in determining the sex of an individual. Human females have two X chromosomes, and males have an X chromosome and a Y chromosome. These chromosomes are called sex chromosomes because they play major roles in determining the sex of an individual. The X and Y chromosomes are very different in size and appearance. The X chromosome is medium-sized with a centromere offset

from the middle of the chromosome, whereas the Y chromosome is much smaller (about 25% as large as the X) and has its centromere very close to one end. The X and Y chromosomes are very different in size and appearance. The X chromosome is medium-sized with a centromere offset from the middle of the chromosome, whereas the Y chromosome is much smaller (about 25% as large as the X) and has its centromere very close to one end. At meiosis, the X and Y chromosomes pair only at a small region at the tip of the short arms, indicating that most genes on the X chromosome are not present on the Y. At meiosis, the X and Y chromosomes pair only at a small region at the tip of the short arms, indicating that most genes on the X chromosome are not present on the Y. Female humans have two copies of all X-linked genes and can be heterozygous or homozygous for any of them. Males, in contrast, carry only one copy of the X chromosome. Female humans have two copies of all X-linked genes and can be heterozygous or homozygous for any of them. Males, in contrast, carry only one copy of the X chromosome. Males are affected by X-linked recessive genetic disorders far more often than females are. Males are affected by X-linked recessive genetic disorders far more often than females are. Dominant X-linked traits have a distinctive pattern of transmission with three characteristics: Look on page 83. Dominant X-linked traits have a distinctive pattern of transmission with three characteristics: Look on page 83. Affected males transmit the trait only to daughters, never to sons. Affected males transmit the trait only to daughters, never to sons. The most common form of muscular dystrophy is an X-linked disorder, Duchenne muscular dystrophy (DMD), which affects 1 in 3, 500 males in the United States. The most common form of muscular dystrophy is an X-linked disorder, Duchenne muscular dystrophy (DMD),

which affects 1 in 3, 500 males in the United States. The DMD gene is located near one end of the X chromosome and encodes a protein called dystrophin. The DMD gene is located near one end of the X chromosome and encodes a protein called dystrophin. Becker muscular dystrophy (BMD), make a shortened form of dystrophin that is partially functional. Becker muscular dystrophy (BMD), make a shortened form of dystrophin that is partially functional. BMD and DMD, are two diseases that are caused by different mutations in the same gene. BMD and DMD, are two diseases that are caused by different mutations in the same gene. Genes carried on the Y chromosome are called Y-linked. Genes carried on the Y chromosome are called Y-linked. Mitochondria are cytoplasmic organelles that convert energy from food molecules into ATP, a molecule that powers many cellular functions. Mitochondria are cytoplasmic organelles that convert energy from food molecules into ATP, a molecule that powers many cellular functions. Patterns of gene expression are influenced by many different environmental factors. Patterns of gene expression are influenced by many different environmental factors.