

Because learning changes everything. (R) Chapter 13 Mutations and Genetic Testing Essentials of Biology SEVENTH EDITION Sylvia S. Mader Michael Windelspecht (C) McGraw Hill LLC. Rheumatoid arthritis o Immune system destroys person's own body o Inject adenoviruses that contain anti-inflammatory genes into the affected joint (C) McGraw Hill LLC 2 Figure 13.10 Use of a DNA Microarray to Test for Mutated Genes (C) McGraw Hill LLC 18 DNA Sequencing o Individual's complete genome o Important in personalized medicine o Can not only show risk for disease but also holds key to new treatment options o Based on genetic information, it is possible to improve patient outcomes and increase treatment options with increased precision o Becoming more affordable leading to popularity of personalized medicine (C) McGraw Hill LLC 19 Testing the Fetus Ultrasound--helps evaluate fetal anatomy for serious abnormalities o Uses high-frequency sound waves Testing fetal cells o Cells from amniocentesis or chorionic villus sampling o Fetal cells can also be collected from mother's blood o PCR used to amplify DNA, no risk to fetus (C) McGraw Hill LLC 20 Testing the Embryo o Testing the embryo-- following IVF, one cell can be removed from embryo without harm Access the text alternative for slide images. Inv dup 15 syndrome--inverted duplication of chromosome 15 o Inversion--segment joins in direction opposite from normal (C) McGraw Hill LLC 9 Chromosomal Mutations- Translocation Rearrangement of parts between nonhomologous chromosomes. o Alignment only possible when the inverted chromosome forms a loop (C) McGraw Hill LLC 12 Genetic Testing Genetic counseling-- potential parents are advised on their risk of inherited disorders Counselor helps couple understand the mode of inheritance, medical consequences of disorder, and decisions they might wish to make Karyotyping--visual display of chromosomes arranged by size, shape, and banding pattern. (C) McGraw Hill LLC Kateryna Kon/Shutterstock 15 Testing for a Protein Some disorders are caused by a missing enzyme o Test for quantity of enzyme produced (C) McGraw Hill LLC 16 Testing the DNA- Genetic Markers Relies on an abnormality in the DNA sequence due to presence of abnormal allele. (C) McGraw Hill LLC 25 In Vivo Gene Therapy Cystic fibrosis treatment o Gene needed is sprayed into the nose or delivered to lower respiratory tract o Use adenoviruses or liposomes to carry gene Poor coronary circulation treatment o Vascular endothelial growth factor can cause growth of new blood vessels. (C) McGraw Hill LLC 23 Ex Vivo Gene Therapy Treatment of SCID o Severe combined immunodeficiency o Lack enzyme involved in the maturation of cells producing antibodies o Bone marrow stem cells are removed and infected with RNA retrovirus carrying the gene for normal enzyme. o Williams syndrome-- chromosome 7 loses a tiny end piece o Cri du chat--chromosome 5 loses an end piece (C) McGraw Hill LLC 8 Chromosomal Mutations- Duplication Chromosome segments are repeated Individual has more than two alleles for certain traits. 5% of Down syndrome cases caused by a translocation in previous generation between chromosomes 21 and 14 o Not related to parental age but is inherited (C) McGraw Hill LLC 10 Chromosomal Mutations- Inversion A chromosome segment is turned 180° o Small portion of liver is removed and infected with retrovirus containing normal cholesterol receptor o Tissue is returned to patient (C) McGraw Hill LLC 24 Figure 13.13 Ex Vivo Gene Therapy in Humans Access the text alternative for slide images. (C) McGraw Hill LLC 2 Gene Mutations Gene: A segment of DNA that encodes for a functional product, typically a protein, crucial for cellular functions. Figure 13.9 Use of a Genetic Marker to Test for a Genetic Mutation (C) McGraw Hill LLC 17 Testing the DNA- DNA

Microarrays Mutant alleles placed onto a DNA microarray, small silicone chip which is compared to DNA of a person to indicate risk of developing disease, which can be discussed with genetic counselor. Include deletions, duplications, inversions, and translocations. All rights reserved. 2.3.4.?