*13.2: Human Genetic Disorders*

A. How Are Genetic Disorders Inherited in Humans?

**\*Some genetic disorders are caused by mutations in the DNA of genes. Other disorders are caused by changes in the overall structure or number of chromosomes.**

1. Cystic Fibrosis – occurs in the \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ & \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ where the body produces abnormally thick mucus making it hard for the person to breathe.

 \*Occurs when two mutated alleles are inherited, one from each parent

2. Sickle-Cell Disease – caused by a mutation that affects \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ - a protein in red blood cells that carries oxygen.

\*Red blood cells have a sickle (crescent) shape and can’t carry as much oxygen as normal cells and also clog blood vessels.

 3. Hemophilia – person’s blood clots very slowly or not at all

 \*Don’t produce enough of a protein needed for normal \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_.

 4. Down Syndrome – have an extra copy of chromosome \_\_\_\_\_\_\_\_\_\_\_\_\_\_ in every cell.

\*Three copies instead of two, most often occurs when chromosomes fail to separate properly during meiosis.

B. How Are Genetic Disorders Traced, Diagnosed, and Treated?

**\*Today, doctors use tools such as pedigrees, karyotypes, and genetic testing to trace and diagnose genetic disorders. People with genetic disorders are helped through medical care, education, and job training.**

1. Pedigrees – a \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ or “family tree” that tracks which members of a family have a particular trait.

 \*The trait in a pedigree can be ordinary, such as eye color, or a genetic disorder

2. Karyotypes - \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ of all the chromosomes in a person’s cell. (See page 454).

\*Arranged in pairs and can show if a person has the correct number of chromosomes in their cells.