Dna abnormalities that manifest as disorders

1. Using our knowledge of the genetic code, transcription and translation, explain how a single gene disorder may arise from a mutation. Cite an example of a single gene disorder, explaining what gene has been altered, how this affects the product of this gene, and how this may affect the health of the individual carrying the mutation. If possible, state what the specific mutation is. 2. Using your knowledge of replication, meiosis, and fertilization, explain how a chromosome abnormality may arise in the offspring of two healthy individuals.

Cite an example of a chromosome abnormality, explaining what chromosome or chromosomal segment is missing or duplicated, and how this may affect the health of the individual carrying the abnormality. 3. Not all disorders are easy to explain, but some (such as breast cancer, colon cancer, type 2 diabetes) tend to happen more often within certain families. Using your knowledge of biotechnology, explain how specific DNA abnormalities can be used to determine a person`s chances of developing some diseases, and how this knowledge may prevent the development of the disease.

ANSWERSSingle gene disorder may arise from a mutationSingle gene disorder may arise from mutation. The process of transcription and translation can be disrupted by these forms of mutations. These mutations include single base substitution that suppress termination of protein translation, alter patterns of mRNA splicing during transcription (Garden 2002). Another mutation is single base deletion that definitely changes the outcome of mRNA transcription or the protein synthesis of translation.

One example of a single cell disorder is the sickle cell disease. In people with sickle cell disease, at least one of the beta hemoglobin subunits in hemoglobin is replaced with hemoglobin S (Genetic Home Reference, 2007). Sickle red blood cells become distorted forming a cell that looks like a sickle. This distorted red blood cell is unable to fully transfer oxygen and may sometimes clog up in the blood vessels. The low oxygen capacity and short life span of sickled red blood cells causes anemia to the person afflicted with these genetic disorder. This is a form of single base substitution where a normal amino acid (valine) is replaced by another one (glutamine).

Chromosome abnormality may arise in the offspring of two healthy individualsThree mechanisms contribute to genetic variation during meiosis and fertilization. These are independent assortment of chromosomes, crossing over and random fertilization. Within these three processes is window for chromosome disorder to occur. Meiotic nondisjunction, the failure of the chromosomes to disjoin and pass to opposite poles, in either the first or second meiotic division is the major cause of chromosomal abnormalities (Tissot & Kaufman, 2007).

This includes trisomy, which is the presence of 3 chromosomes instead of 2. Down syndrome, or trisomy 21, is the classic example of a human disease caused by autosomal nondisjunction where some, but not all, affected individuals do survive (Tissot & Kaufman, 2007). Trisomy 21 is characterized by an extra chromosome 21. Individuals with Down syndrome may are most commonly characterized by oblique eye fissures with epicanthic skin folds on the inner corner of the eyes, poor muscle tone, a flat nasal bridge, a single palmar fold (also known as a simian crease), a protruding tongue (due to

small oral cavity, and an enlarged tongue near the tonsils), a short neck, white spots on the iris known as Brushfield spots, excessive flexibility in joints, congenital heart defects, excessive space between large toe and second toe, a single flexion furrow of the fifth finger, and a higher number of ulnar loop dermatoglyphs (Wikipedia, 2007). In most cases, individuals with down syndrome have mental retardation.

Specific DNA abnormalities can be used to determine a person`s chances of developing some diseasesSome minor DNA abnormalities do not cause immediate problems but may cause an individual to have higher chances of getting a certain type of disease. Since these abnormal genes have slight differences than normal, it is possible to identify these differences by comparing them to normal genes. Recent developments in technology like the southern blotting and the use of polymerase chain reaction allowed scientist to detect genetic disorder before they can cause a disease. For example, scientists were able to search for the gene that causes an important form of colon cancer known as hereditary nonpolyposis colorectal cancer (HNPCC).

People who inherit the HNPCC gene have an 80% or greater chance of developing colon cancer and other cancers, usually at an early age (Patlak, 2003). Women with the gene also face a markedly increased risk of uterine and ovarian cancer (Patlak, 2003). Applying the same principle, scientist can isolate certain genetic traits common to people afflicted by a certain disease. Analysis can then be focused on the susceptibility of this gene to the disease. It can then serve as an early warning device where people can

prepare to prevent the disease from occurring or produce a genetic solution to the disease.

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