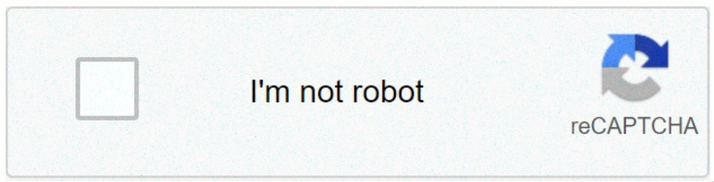

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14.2 human genetic disorders answer sheet

A genetic disorder involves a health condition caused by a genome abnormality, present from birth and out of control. Such disorders may or may not be inheritable; whether they can be transmitted from one generation to another depends on the specific nature of a gene-based health problem. In the case of non-inheritable genetic disorders, the defect in genetic makeup can be caused by new mutations occurring or changes in DNA. From a medical point of view, there are no known cures for such genetic disorders, although treatment can help relieve symptoms. This is a list of the most common genetic disorders that occur in humans. Hemophilia is a rare inherited genetic disorder that prevents blood from clotting properly. It is a bleeding disorder that disrupts the body's ability to control the clotting of blood that is needed when blood vessels rupture. Patients with hemophilia bleed longer after an injury compared to others who do not have the disorder. Smaller cuts don't really pose a problem, but deep bleeding inside the body is of greater concern. Deep internal bleeding in the ankles or knees can damage organs and endanger life. Although there is no adequate treatment for this inherited disorder, but with proper care and an active lifestyle, the condition may be improved. Family hypercholesterolemia is an inherited condition characterized by extremely high levels of (bad) cholesterol in the bloodstream. It is an important factor in emerging diseases related to a cardiovascular system, which manifests itself in the form of ischemic heart disease, hardening of the arterial circulatory system and the onset of stroke. The pathological process that leads to the onset of previous diseases is called arteriosclerosis/atherosclerosis, which occurs when cholesterol in our body is excessive, either by disorderly processes of its destruction/construction, or by its excessive intake through food. It can be treated by involving some lifestyle and diet changes, along with eliminating saturated fat intake. Sickle cell disease (SCD) is an inherited form of anemia, a condition in which there are not enough healthy red blood cells to carry adequate oxygen levels in the body. Normally, red blood cells are flexible and round and move easily through blood vessels. In sickle cell anemia, red blood cells become stiff and sticky and are sickle-shaped or crescent-shaped. They can get stuck in small blood vessels, which can or block blood flow and oxygen to parts of the body. Eventually, a patient dies due to organic insufficiency. This is the most common hereditary kidney disease that is transmitted by families as an autosomal dominant trait. Its frequency in the general population is 1 in 1,000. Polycystic kidney disease occurs in 4-10% of patients on dialysis. about patients have high blood pressure as their first symptom. The prevalence of hypertension increases with a decrease in kidney function. Pain in the backs and abdomen is found in half of patients, usually caused by cystic renal enlargement. Acute pain may indicate bleeding in the cyst, obstruction (thrombus) or infection (accompanied by fever). Brute hematuria (blood in the urine) is the first symptom of the disease and occurs due to rupture of blood vessels. Cystic fibrosis (also called mucoviscidosis), a generalized inherited disorder, causes significant damage to the lungs and digestive system. Mucus (sputum) that occurs in the lungs of healthy people is much less common compared to the dense, sticky sputum of patients suffering from cystic fibrosis. This thick mucus attaches to the lungs, causing airway obstruction. If mucus remains inside the lungs, it clogs the tiny airways and creates an environment suitable for the growth of microorganisms (bacterial infection). For this reason, it is very important to clean the airways through physical therapy. The disorder is not contagious and cannot be transferred from one child to another. Currently, there is no cure for cystic fibrosis. Tay-Sachs disease (also called GM2 gangliosidosis) is a rare hereditary autosomal disorder in which gangliosides, fat breakdown products, accumulate in nerve tissues. HEXA gene mutations cause progressive deterioration of nerve cells. At a very young age, probably at 6 months of age, children develop paralysis, dementia, blindness and spots on the retina. These children usually die at the age of 3 or 4. Tay-Sachs disease cannot be treated or cured. First, a child loses the ability to focus on objects and develop an unusually strong motor reaction to sudden sound stimulation. For example, the child is excessively frightened in every sound. At best, they learn to crawl and sit, but they can't walk, and they communicate very little with the environment. The first case of Marfan syndrome, symptoms and diagnosis was described in 1896. Marfan syndrome is a genetic disease that affects connective tissue. The main symptoms of external manifestations are disproportionately long limbs, thin fingers and very high growth. The disease is characterized by the presence of cardiovascular malformations (violation of the structure of the aorta and damage to the heart valves). This disorder mainly affects the eyes, lungs, skeletal system and circulatory system. Marfan syndrome inherit through the genes of one parent. Another reason to develop the disease is the genetic mutation due to the accumulation of excessive amounts of growth hormone, the rapid development of the lungs, and therefore rapid enfeeblement of the aorta, causing Marfan syndrome. Hereditary spherocytosis is an inherited disorder in which normal discoid red blood cells Spherical. Rigid red blood cells incorrectly capture, enlarge, and destroy the spleen, leading to anemia. Anemia is usually mild, but if the infection develops, it can become severe easily. When the disorder is severe, the patient may develop jaundice onset of gallstones. In young adults, this disorder can be replaced with hepatitis. Bone abnormalities, such as a tower-shaped skull and additional fingers and fingers, may occur. Treatment is usually not necessary, but sometimes severe anemia may require removal of the spleen. Neurofibromatosis is an inherited disorder that causes tumors to form in nerve tissues. The type 1 NF gene is on chromosome 17. This gene is also called a suppressor, as it prevents excessive cell proliferation. In the case of the mutation of this gene, its tumor suppressive function is disturbed, leading to the appearance of type 1 NNF, and the uncontrolled growth of numerous cells in the body, particularly schwann cells involved in the construction of nerve coating. Changes caused by this disease affect many organ systems, most commonly the skin, central and peripheral nervous system, bones and soft tissues. A progressive brain disorder, Huntington's disease leads to progressive degeneration of nerve cells, resulting in mental impairment. This hereditary neurodegenerative disorder is incurable and causes the breakdown of nerve cells in the brain. Huntington's disease affects a person's muscle coordination, ability to move and talk, as well as cognitive functions. Aside from this, individuals suffering from the disorder may also experience behavioral changes, difficulty swallowing, and memory loss. Each child of a parent who has Huntington's disease has a 50% chance of getting the disease. Diseases are classified as rare if fewer than 200,000 people are diagnosed with the condition. The National Institutes of Health lists 7000 rare diseases, affecting between 25 and 30 million people. People inherit most rare diseases, but there are those that appear randomly due to strange genetic mutations. Historically, pharmaceutical companies have chosen not to develop treatments for these diseases. As a result, medical professionals refer to these rare genetic disorders as orphan diseases. Some are fatal, but others have no serious complications and ultimately do not affect the individual's quality of life. When a protein called prion causes normal brain proteins to fold abnormally, individuals they can develop a prion disease such as fatal family insomnia, which involves the PRNP gene. FFI is the result of changes in the thalamus, the part of the brain that regulates sleep and consciousness, motor control, and many other bodily functions. The disease causes insomnia, weight loss, increased or decreased body temperature, and rapid dementia. Only a gene from a father to develop. Within 12 to 18 months of the first symptom, the individual dies. Ongoing research on the disease is ongoing. designer491 / Getty Images At birth, children born with progeria look healthy, but by the time they reach their second birthday, symptoms begin to appear. This genetic condition causes children to show signs of aging at an accelerated rate. They experience growth failures, body fat loss and visible changes in the skin similar to those of a much older adult. People with progeria also experience progressive hardening of the arteries, strokes and cardiovascular disease. Of the 1000 children born with the disease each year, most will only live about 14 years. Researchers identified a mutation of the LMNA gene as the cause. Thos Robinson / Getty Images Short stature with limb, facial and genital abnormalities characterize a rare genetic condition called Aarskog syndrome. The disorder affects more men than women and sometimes affects intellectual abilities. About 20 percent of people with Aarskog syndrome have a mutation of the FGDI gene, but the cause is unknown in others. Symptoms vary from person to person. Dropped eyelids, an underdeveloped upper jaw, fleshy ear lobes, prominent widow's beak, and delayed dental rash further identify those with the condition. The disorder occurs in 1 in 25,000 children. stevanovicigor/Getty Images Unlike many other rare genetic disorders, Gitelman syndrome may not appear until the second decade of life. Two mutated genes, one from each parent, cause the disorder. Gitelman syndrome, often compared to Bartter syndrome, rarely damages the kidneys leading to a progressive kidney condition. The disorder causes an inability to reabsorb salt, forcing changes in electrolytes and fluid out of cells, and leads to dehydration. People may experience frequent urination as well as pass large amounts of urine. In addition, some have heart palpitations, fainting and salt cravings. Most people with Gitelman syndrome can expect a good prognosis, although there is no cure. cdascher / Getty Images Appearing between the ages of 1 and 4, Angelman syndrome used to be called happy puppet syndrome. Symptoms vary greatly. Severe developmental delays, learning disabilities, tremors, sudden movements in the arms and legs, hand flapping, sleep disorders, and seizures characterize the disorder. Some people can't speak at all, while others learn to communicate through Diagnosed children often have a happy disposition and burst into uncontrollable laughter for no apparent reason. About ten percent are unable to walk. Cases may not be diagnosed and confused with other disorders, but researchers estimate that the syndrome affects 1 in 20,000 children. Most people have one percent methemoglobin in their blood. Methemoglobinemia, or blood disorder, occurs when that level increases to 20 percent or more. The disease can be genetic or acquired. There are two genetic forms of the disease. Type 1 requires genes transmitted by both parents and affects only red blood cells. The skin turns bluish. People with type 2 methemoglobinemia require a single-parent gene; this is the rarest form and affects all cells. Babies born with this type usually do not live beyond their first year. The main symptom of xeroderma pigmentosa is increased sensitivity to ultraviolet radiation. Damage is most common to the skin, eyelids and eye surfaces, however, the tip of the tongue can also be damaged due to sun exposure. People with XP may also have poor balance and speech issues. Risks include a 10,000-times increased chance of developing skin cancer, melanomas and squamous cell carcinomas, and many die of cancer at an early age. Only one in a million children inherit XP. However, the most severe form of the disease occurs 30 times more often in Navajo children. Sohel Parvez Haque / Getty Images A rare multisistmic genetic disorder, hypohydrotic ectodermal dysplasia occurs in men, but decreased symptoms can also occur in girls carrying a single copy of the gene. Individuals with HED do not have sweat glands or only a small number of them, which prevents sweating. They may also have abnormally dispersed hair, thin skin, and missing teeth. Wheezing, shortness of breath, recurrent respiratory infections, and itchy rashes can occur frequently. Although there is no specific treatment, the prognosis is generally positive. Paladjai / Getty Images Babies born with lamellar ichthyosis have a layer of shiny, zeroy skin at birth that disappears within the first two weeks. Once shed, reveal a layer of red, scaly skin underneath. People with this condition have a high risk of skin infection, chronic skin blisters and dehydration. Signs of lamellar ichthyosis include thick skin on the palms of the hands and lower feet, breathing problems, nail abnormalities, and ectropion, an eyelid that rotates outward. This rare condition only occurs in one of 200,000 to 600,000 individuals and can affect both men and women. Prognosis depends on the severity of symptoms, but is generally poor. Castillof / Getty Images Children born with a partial or complete fusion of the legs have an

extremely rare disorder called sirenomelia syndrome or mermaid. Researchers have not discovered reasons for the condition, but believe genetic and environmental factors play a role. In some cases, the spine and pelvis are malformed; individuals may have no feet, and often have no tailings. A variety of serious complications often prove fatal in the first few years of life. However, a small number of children survive into childhood and live in young adulthood. NanoStockk / Getty Images Images Images