

## Chromosomes and human heredity study guide

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# KARYOTYPES AND PEDIGREES

## Inheritance of Genetic Traits

Name: \_\_\_\_\_ ANSWER KEY TEST Mon



### Diseases Unit Review Guide

1. There are 5 major types of asexual reproduction. Name all 5 and give an example of each.

- Binary Fission - example: bacteria
- Budding - example: yeast
- Cutting - example: spider plant
- Runners - example: strawberries
- Regeneration - example: starfish

2. Comparison of Asexual versus Sexual Reproduction

	Number of Parents (cells)	What type of organisms	# of Chromosomes parents-vs-offspring	Variety of DNA (more or less)	Examples
Asexual	1	unicellular	same	less	bacteria
Sexual	2	multicellular	half	more	human

3. Compare the process of Mitosis and Meiosis. Use your meiosis notes to help you.

Mitosis	Meiosis
Circle the type of reproduction: <u>asexual</u> or sexual	Circle the type of reproduction: asexual or <u>sexual</u>
What type of cells are produced: <u>Non-sex cells</u>	What type of cells are produced: <u>Sex cells</u>
How many cells are produced: <u>1</u>	How many cells are produced: <u>4</u>
How does the DNA compare to the original parent cell? <u>identical</u>	How does the DNA compare to the original parent cell? <u>mixed</u>

## Chromosomes and Heredity

modified from: [http://brookings.k12.sd.us/biology/other\\_units.htm](http://brookings.k12.sd.us/biology/other_units.htm)

A family record that shows how a trait is inherited over several generations is called a \_\_\_\_\_

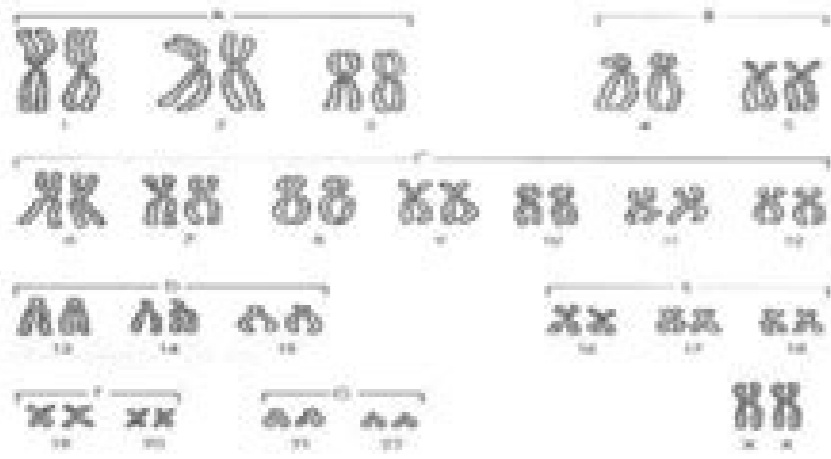
- A. karyotype
- B. Punnett square
- C. pedigree
- D. periodic table

A person that has ONE copy of an AUTOSOMAL RECESSIVE allele and does not express the trait, but can pass it along to his/her offspring is called a \_\_\_\_\_.

- A. mutant
- B. carrier
- C. hemophiliac
- D. gene marker

The failure of homologous chromosomes to separate during meiosis is called \_\_\_\_\_

- A. segregation
- B. codominance
- C. sex-linkage
- D. nondisjunction



This picture of an organism's chromosomes is called a \_\_\_\_\_

- A. pedigree
- B. Punnett square
- C. karyotype

The person shown at the left is a \_\_\_\_\_ because they have two X chromosomes.

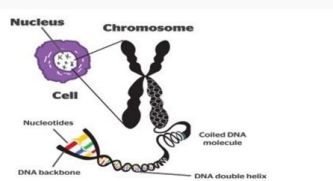
- A. male
- B. female

The chromosomes that DO NOT determine sex are called \_\_\_\_\_

- A. sex chromosomes
- B. autosomes
- C. gene markers
- D. pedigree partners

Which parent determines the sex of the baby?

- A. father
- B. mother



COMPLETE ANDROGEN INSENSITIVITY SYNDROME IN THREE SISTERS: A CASE REPORT

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SUMMARY

Complete androgen insensitivity syndrome (CAIS) is a rare genetic disorder characterized by the absence of androgen receptors in the target tissues... The underlying mechanism is an autosomal recessive mutation in the androgen receptor gene... CAIS may be observed in several members of a family. For this reason, the family members of the affected individual should be screened. In this article, we present a case of three sisters diagnosed with CAIS after investigating the phenotype of one of them during an infertility workup.

Keywords: complete androgen insensitivity syndrome, androgen receptor mutation, infertility, androgen insensitivity syndrome

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KOMPLET ANDROGEN HUSARSIZLIK SENDROMU ÜÇ KIZ KARDEŞ

ÖZET

Komplekt androgen insensiviteci sendromu (KAIS) nadir genetik bir durumdur. Karakteristik olarak androgen reseptörlerinin hedef dokularda bulunmamasıdır. Bu durum otozomal resesif bir mutasyonun sonucudur. KAIS bir ailede birden fazla üyede gözlemlenebilir. Bu nedenle aile üyelerinin de taraması önerilmektedir. Bu çalışmada üç kız kardeşin KAIS ile tanındığını ve aile üyelerinin de taramasını yaptığımızı sunuyoruz. Bu çalışmada üç kız kardeşin KAIS ile tanındığını ve aile üyelerinin de taramasını yaptığımızı sunuyoruz.

Anahtar kelimeler: complete androgen insensitivity syndrome, androgen receptor mutation, infertility, androgen insensitivity syndrome

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Heredity and genetics answer key. Chapter 11 section 3 chromosomes and human heredity study guide answers. Human heredity answer key.

Parents pass on traits to their children through their genes. Genes are the blueprint for our bodies. Almost every cell in the human body contains a copy of this blueprint, mostly stored inside a special sac within the cell called the nucleus. Genes are part of chromosomes, which are long strands of a chemical substance called deoxyribonucleic acid (DNA). Therefore, genes are made up of DNA. A DNA strand looks like a twisted ladder. The genes are like a series of letters strung along each rung. These letters are used like an instruction book. The letter sequence of each gene contains information on building specific molecules (such as proteins or hormones, both essential to the growth and maintenance of the human body). The genes are copied 'letter for letter' to a similar substance called ribonucleic acid (RNA). The working parts of the cell read the RNA to create the protein or hormone according to the instructions. Each gene codes the instruction for a single protein only, but one protein may have many different roles in the human body. Also, one characteristic, such as eye colour, may be influenced by many genes. Recall that the nucleus of a cell contains chromosomes on which genes are located. The nucleus of a eukaryotic cell contains the DNA, the genetic material of the cell. The DNA contains the information necessary for constructing the cell and directing the multitude of synthesis tasks performed by the cell in the process of life and reproduction. Inside the nuclear envelope is the chromatin, meaning "colored substance" after the early experiments in which that material was highly colored by the staining techniques used. Chromatin consists of DNA associated with proteins which forms long strands called chromosomes. State that a gene is a section of a molecule of DNA and that a gene codes for a specific protein. A gene is the basic physical and functional unit of heredity. Genes, which are made up of DNA, act as instructions to make molecules called proteins. In humans, genes vary in size from a few hundred DNA bases to more than 2 million bases. The Human Genome Project has estimated that humans have between 20,000 and 25,000 genes. Every person has two copies of each gene, one inherited from each parent. Define gene, allele and genome. Define: Give the precise meaning of a word, phrase or physical quantity. Gene - A gene is the molecular unit of heredity of a living organism. Allele - An allele is one of two or more versions of a gene. Genome - A genome is an organism's complete set of DNA, including all of its genes. Describe a DNA molecule. Describe: Give a detailed account. Image from www.britannica.com. DNA, or deoxyribonucleic acid, is the hereditary material in almost all living organisms. Nearly every cell has the same DNA. Most DNA is located in the cell nucleus, but a small amount of DNA can also be found in the mitochondria. DNA is made up of molecules called nucleotides. Each nucleotide contains a phosphate group, a sugar group and a nitrogen base. The four types of nitrogen bases are adenine (A), thymine (T), guanine (G) and cytosine (C). The order of these bases is what determines DNA's instructions, or genetic code. Nucleotides are attached together to form two long strands that spiral to create a structure called a double helix. Explain DNA replication. Explain: Give a detailed account of causes, reasons or mechanisms. Image from 2012books.lardbucket.org. All organisms must duplicate their DNA with extraordinary accuracy before each cell division. DNA replication begins with the "unzipping" of the parent molecule by the enzyme helicase as the hydrogen bonds between the base pairs are broken. Once exposed, the sequence of bases on each of the separated strands serves as a template to form a complementary set of bases on the strand being synthesized. The new complementary strands are put back together with the enzyme DNA polymerase. Some DNA polymerases also have proofreading ability; they can remove nucleotides from the end of a growing strand in order to correct mismatches. When the process is complete, two DNA molecules have been formed identical to each other and to the parent molecule. Outline DNA transcription in terms of the formation of an RNA strand complementary to the DNA strand by RNA polymerase. Outline: Give a brief account or summary. Image from www.contexto.info. Protein synthesis requires two steps: transcription and translation. Transcription is the process of making an RNA copy of a gene sequence. This copy, called a messenger RNA (mRNA) molecule, leaves the cell nucleus and enters the cytoplasm, where it directs the synthesis of the protein, which it encodes. Translation is the process of translating the sequence of a messenger RNA (mRNA) molecule to a sequence of amino acids during protein synthesis. The genetic code describes the relationship between the sequence of base pairs in a gene and the corresponding amino acid sequence that it encodes. In the cell cytoplasm, the ribosome reads the sequence of the mRNA in groups of three bases to assemble the protein. Define mutation as a change in a gene or chromosome. Define: Give the precise meaning of a word, phrase or physical quantity. A mutation is when a gene is copied incorrectly, this gene can be passed down. It doesn't happen very often or on purpose. Some variations are repaired by the cell's DNA error correction mechanisms. Other variations are not and can be passed on to subsequently replicated cells; in such cases, the variation is termed a mutation. If the mutation occurs in a gamete (sex cell), then the mutation can be passed on to the next generation. Outline the effects of ionising radiation (for example gamma rays, X-rays and ultraviolet rays) and some chemical mutagens (for example chemicals in tobacco) on the rate of mutations. Outline: Give a brief account or summary. Image from www.cancer.gov. Radioactive materials that decay spontaneously produce ionizing radiation. Ionizing radiation has sufficient energy to break some chemical bonds. Any living tissue in the human body can be damaged by ionizing radiation. The body attempts to repair the damage, but sometimes the damage is of a nature that cannot be repaired or it is too severe or widespread to be repaired. Also mistakes made in the natural repair process can lead to cancerous cells. The most common forms of ionizing radiation are alpha and beta particles, or gamma and X-rays. Research studies have also shown that cigarette smoke and many air pollutants are strongly linked to chromosome damage in cells. Describe that many mutations are harmful but some are neutral and a few are beneficial. Describe: Give a detailed account. Image from www.smithsonianmag.com. Many people consider mutations in the human genome to be a negative event as they associate mutations with cell damage, cancer or genetic diseases. However, when proper research is done, we can see that this is in fact incorrect: few human mutations are actually bad, most are completely neutral, and in fact, many are beneficial. Without mutations, we could not exist as we do and there would never have been evolution and natural selection as we know it. Mutations allow genetic diversity to exist within a population, increasing the range of alleles and keeping the human species alive. 1. Malaria resistance. Individuals with sickle cell have been observed to portray malaria resistance. Generally, this is a harmful mutation since sickle blood cells are not as effective as normal cells in their functioning. In regions where there are increased cases of malaria, this mutation is favorable as individuals with blood cells that are sickle shaped are not likely to contract malaria. 2. Lactose tolerance. Lactose intolerance has made it simpler to wean young children. Nevertheless, human beings still like drinking milk. People found out they could use milk products as they had developed lactose tolerance due to genetic mutation. Lactose tolerance even has a strong relationship with being descendants of cultures that used milk like a food. Identify the karyotype of an individual. In every cell in the human body there is a nucleus, where genetic material is stored in genes. Genes carry the codes responsible for all of our inherited traits and are grouped along rod-like structures called chromosomes. Typically, the nucleus of each cell contains 23 pairs of chromosomes, half of which are inherited from each parent. Down syndrome occurs when an individual has a full or partial extra copy of chromosome 21. Key Terms: chromosome, genes, DNA, genetic material, Crick, structural proteins, hormones, substitution, inversion, mRNA, translation, Watson, Franklin, genetically identical, nucleotides, helicase, DNA polymerase, homologous, parallel, RNA, transcription, polynucleotide, phosphate group, nitrogenous base, ribosomes, complementary base pairing, mutation, duplication, deletion, RNA, polypeptide, base-pairing rule, sense strand, anti-sense strand, triple codon, RNA, natural selection, mutagen, histone, uracil, amino acid, adenine, thymine, guanine, cytosine, intracellular enzymes, universal code, protein synthesis, sex chromosome, karyotype, Down's syndrome, Sickle Cell Anemia, Class Material Part 1 DNA Structure Part 2 DNA Replication Part 3 Transcription Part 4 Translation Part 5 Chromosomes Part 6 Mutations What do a man, a mushroom, and an elephant have in common? A very long and simple double helix molecule makes us more similar and much more different than any other living thing. But, how does a simple molecule determine the form and function of so many different living things? The discovery of double-helix structure of DNA is to science what Mona Lisa is to painting. It has been called the single biggest discovery of all times. But it was not just stumbled upon - it was a race. Rosalind Franklin was a British scientist who helped discover the structure of DNA, but you most likely have not heard of her. Hank will attempt to fix this gap in your knowledge on today's SciShow. In 1990, The Human Genome Project proposed to sequence the entire human genome over 15 years with \$3 billion of public funds. Then, seven years before its scheduled completion, a private company called Celera announced that they could accomplish the same goal in just three years at a fraction of the cost. Tien Nguyen details the history of this race to sequence the human genome. Your genome, every human's genome, consists of a unique DNA sequence of A's, T's, C's and G's that tell your cells how to operate. Thanks to technological advances, scientists are now able to know the sequence of letters that makes up an individual genome relatively quickly and inexpensively. Mark J. Kiel takes an in-depth look at the science behind the sequence. Paul Andersen describes the molecular structure of DNA. He describes the major parts of a nucleotide and explains how they are assembled into a nucleic acid. The nitrogenous base, deoxyribose sugar and phosphate group make up a single nucleotide. The 5' and 3' end of DNA is described. The importance of hydrogen bonds in the 3-dimensional shape is also included. These DNA molecular visualizations were created for the multifaceted 'DNA' project. Paul Andersen explains how DNA replication ensures that each cell formed during the cell cycle has an exact copy of the DNA. He describes the Meselson-Stahl experiment and how it showed that DNA copies itself through a semi-conservative process. He then explains how multiple enzymes, like DNA polymerase, helicase, primase, ligase, and single strand binding proteins copy DNA. He also differentiates between the leading and the lagging strand. He explains how DNA is anti-parallel in nature and how eukaryotic cells have multiple origins of replication. Hank introduces us to that wondrous molecule deoxyribonucleic acid - also known as DNA - and explains how it replicates itself in our cells. Paul Andersen explains the central dogma of biology. He explains how genes in the DNA are converted to mRNA through the process of transcription. He then explains how ribosomes use this message to convert the mRNA to a functioning protein. He also shows you how to decode a gene by converting the DNA to complementary mRNA and then to the specific amino acids in a protein. Hank imagines himself breaking into the Hot Pockets factory to steal their secret recipes and instruction manuals in order to help us understand how the processes known as DNA transcription and translation allow our cells to build proteins. RNA is synthesized from DNA, and enters the ribosome where protein translation and synthesis occurs. This video looks first at the structure of DNA before going on to describe how DNA carries out its four major functions: the storage of information; the replication of information; creating slight changes in the information through mutations that forces of natural selection act upon; and the translating of information into the proteins that define an individual. The DNA in just one of your cells gets damaged tens of thousands of times per day. Because DNA provides the blueprint for the proteins your cells need to function, this damage can cause serious issues—including cancer. Fortunately, your cells have ways of fixing most of these problems, most of the time. Monica Mensini details the processes of DNA damage and repair. Try these clips on Mutations. A gene known as HbS was the center of a medical and evolutionary detective story that began in the middle 1940s in Africa. Doctors noticed that patients who had sickle cell anemia, a serious hereditary blood disease, were more likely to survive malaria, a disease which kills some 1.2 million people every year. What was puzzling was why sickle cell anemia was so prevalent in some African populations.

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