**A mistake in copying DNA can result in dwarfism.**[[1]](#footnote-1)

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| This photo shows a family in which both parents have normal height, but their daughter has a type of dwarfism called achondroplasia. The average height of an adult with achondroplasia is a little over 4 feet. Achondroplasia is caused by a single dominant allele of a gene for a protein that regulates bone growth.  **1.** Suggest a genetic explanation for how these parents could have a daughter who has achondroplasia, even though neither parent had achondroplasia.  To begin our analysis, the table below shows the effects of the alleles of the gene that can cause achondroplasia. |  |

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| **Genotype** | **→** | **Protein** | **→** | **Characteristics** |
| **dd** | **→** | Normal regulator protein that inhibits bone growth enough to result in normal adult height | **→** | Person has normal height and mortality risk. |
| **Dd** | **→** | Both normal regulator protein and overactive regulator protein | **→** | Person is very short and has somewhat elevated mortality risk; person has achondroplasia. |
| **DD** | **→** | Overactive regulator protein that excessively inhibits bone growth | **→** | Abnormalities are so severe that the fetus dies before birth or the baby dies shortly after birth. |

**2a.** Which genotype results in achondroplasia? **DD** \_\_\_ **Dd** \_\_\_ **dd** \_\_\_

**2b.** Explain why each of the other two genotypes do not result in achondroplasia.

**3a.**  What were the genotypes of the parents in the family shown above?

**DD** \_\_\_ **Dd** \_\_\_ **dd** \_\_\_

**3b.**  What was the genotype of their daughter? **DD** \_\_\_ **Dd** \_\_\_ **dd** \_\_\_

Although the father did not have a **D** allele, there was a **D** allele in the sperm that fertilized the egg to produce the zygote that developed into the daughter. How did this happen?

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| This figure shows that sperm production requires both   * mitosis (to replenish the supply of sperm stem cells) and * meiosis (to produce the haploid cells that differentiate into sperm).   Before mitosis or meiosis, a cell must replicate its DNA.  A mistake in DNA replication can cause a mutation that converts a **d** allele to a **D** allele. Only a very small mistake is needed to produce this mutation, since the **d** and **D** alleles differ in only one nucleotide out of more than 1000 nucleotides in the gene.  **4a.** What is DNA replication?    **4b.** Mark each cell that carries out DNA replication with an \*.  **5.** Explain how a mistake in copying DNA can result in an offspring who has dwarfism. (A complete answer will include mitosis, meiosis, mutation, allele, sperm, fertilizes, egg, protein, and bone growth.) | Diagram  Description automatically generated |

**Challenge Question**

**6.** The risk that an offspring will have achondroplasia increases as the father’s age increases. Suggest a hypothesis that could explain why.

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| **7a.** Draw a Punnett square for two parents who both have achondroplasia.  **7b.** Circle each genotype which would result in a child with achondroplasia.  **7c.** Use an \* to mark the genotype that would result in a child with normal height.  **7d.** What would happen to a zygote with the **DD** genotype? | **A person and person posing for a picture  Description automatically generated** |

Approximately 20% of cases of achondroplasia are due to inheritance of a **D** allele from a parent with achondroplasia. Approximately 80% of cases of achondroplasia are caused by a new mutation, which changes a **d** allele to a **D** allele. Thus, achondroplasia is a genetic condition, but usually it is not hereditary.

**8.** Explain what it means to say, “Her achondroplasia is a genetic condition, but not a hereditary condition.” (This is true for the daughter described on the first page.)

**9a.** Based on the frequency of dwarfs among the people you have seen in your lifetime, what genotype do most people have? **DD** \_\_\_ **Dd** \_\_\_ **dd** \_\_\_

**9b.** Why is the **D** allele considered dominant, even though this allele is rare?

**10.** Explain how the gene that can cause achondroplasia illustrates the generalization that “A single gene often has more than one effect.” (Hint: See the table on page 1.)

1. By Dr. Ingrid Waldron, Dept. Biology, Univ. Pennsylvania, © 2023. This Student Handout and Teacher Notes (with instructional suggestions and background information) are available at <https://serendipstudio.org/exchange/bioactivities/geneticsdwarf> [↑](#footnote-ref-1)