Year 11 Science: Where genetic variation comes from

Name: _____ Date: _____

Try these without your notes first, then check the answer key.

Practice

1. A woman who is a carrier for albinism (Aa) has children with a man who is homozygous dominant (AA). What is the chance that a child has albinism?

2. In question 1, what fraction of the children are expected to be carriers?

3. Two parents with normal pigment have a child with albinism, then a second child with normal pigment. What is the chance that the second child is a carrier?

4. A man's skin cell mutates after years of sunburn. Can the mutation be passed to his children? Explain.

5. Name the two events in meiosis that make each gamete different, and say which one swaps parts between chromosomes.

Exam-style question: drinking milk as an adult

Context: Lactase is the enzyme that breaks down lactose, the sugar in milk. Most mammals stop making it after weaning, and so do many people. In some human populations, a mutation in a stretch of DNA that switches the lactase gene on lets people keep making lactase as adults. This allele for lactase persistence (L) is dominant over the allele for lactase non-persistence (l).

6. Describe the difference between genotype and phenotype, using lactase persistence as your example.

7. Two adults who both digest milk easily have a son who, as an adult, gets sick after drinking milk. Explain how this happened, using a Punnett square, and give the chance for each of their children.

8. Discuss how the L allele first appeared and why it became common in populations with a long history of keeping dairy animals but stayed rare in others. In your answer, explain why the first mutation must have happened in a gamete-making cell, and link the nature of the allele to survival.

Fix the mistake

9. Correct this statement: "A gene and an allele are the same thing."

10. Correct this statement: "Their first child had albinism, so the next three will not."

11. Correct this statement: "Getting skin cancer from the sun is a mutation, so the person's children can inherit it."

12. Correct this statement: "Meiosis makes new alleles."

Answer key (for parents and tutors)

1. A woman who is a carrier for albinism (Aa) has children with a man who is homozygous dominant (AA). What is the chance that a child has albinism?
   0: every child gets an A from the father, so children are AA or Aa and none is aa.
2. In question 1, what fraction of the children are expected to be carriers?
   ½ (50%): the Punnett square gives AA, AA, Aa, Aa.
3. Two parents with normal pigment have a child with albinism, then a second child with normal pigment. What is the chance that the second child is a carrier?
   ⅔: both parents are Aa; a pigmented child is AA, Aa or Aa (the aa box is ruled out), and 2 of those 3 are carriers.
4. A man's skin cell mutates after years of sunburn. Can the mutation be passed to his children? Explain.
   No: it is a somatic mutation in a skin cell, and only mutations in cells that make eggs or sperm are inherited.
5. Name the two events in meiosis that make each gamete different, and say which one swaps parts between chromosomes.
   Crossing over and independent assortment; crossing over is the one where the two chromosomes of a pair swap sections.
6. Describe the difference between genotype and phenotype, using lactase persistence as your example.
   (Achieved) Genotype is the pair of alleles a person has for the gene: LL, Ll or ll. Phenotype is what shows: being able to digest milk as an adult (LL or Ll) or not (ll). Merit adds that Ll and LL give the same phenotype because L is dominant, so you cannot tell a carrier by looking.
7. Two adults who both digest milk easily have a son who, as an adult, gets sick after drinking milk. Explain how this happened, using a Punnett square, and give the chance for each of their children.
   (Merit) The son is ll, so he received an l from each parent; the parents can digest milk, so each has an L; both parents are Ll. Punnett square: L and l eggs against L and l sperm gives LL, Ll, Ll, ll: 3 persistent to 1 non-persistent, so each child has a ¼ (25%) chance of being ll. Meiosis separates each parent's L and l into different gametes, and random fertilisation joined two l gametes.
8. Discuss how the L allele first appeared and why it became common in populations with a long history of keeping dairy animals but stayed rare in others. In your answer, explain why the first mutation must have happened in a gamete-making cell, and link the nature of the allele to survival.
   (Excellence) The L allele arose as a change in the base sequence of the DNA that controls the lactase gene. It must have occurred in a cell that makes eggs or sperm, because only then can it be inherited and spread through later generations; a somatic mutation would have died with that person. In dairying populations the allele was beneficial: adults who could digest milk had an extra food and drink source, so they were more likely to survive famines and raise children, who inherited L. Over many generations differing survival raised the frequency of L. Where people did not keep dairy animals, L gave no advantage, so it stayed rare. Because L is dominant, Ll carriers of l are persistent too, so the l allele is not removed and still turns up when two carriers have children, as in part 2.
9. Correct this statement: "A gene and an allele are the same thing."
   A gene is the section of DNA for a trait; alleles are the different versions of that gene, such as A and a.
10. Correct this statement: "Their first child had albinism, so the next three will not."
    Each fertilisation is independent, so every child of two carriers has the same ¼ chance, whatever the earlier children were.
11. Correct this statement: "Getting skin cancer from the sun is a mutation, so the person's children can inherit it."
    That is a somatic mutation in skin cells; only mutations in cells that make eggs or sperm are heritable.
12. Correct this statement: "Meiosis makes new alleles."
    Only mutation makes new alleles; crossing over and independent assortment make new combinations of alleles that already exist.