No doubt, if the parental rabbits had been sacrificed, one parent (we cannot tell which) would have been predicted to have white fat and the other yellow. Luckily, the rabbits were not sacrificed, and the same animals were bred again, leading to a very interesting, different result. Often in science, an unexpected observation can lead to a novel principle, and, rather than moving on to something else, it is useful to try to explain the inconsistency. So why did the 3:1 ratio disappear? Here are some possible explanations.

First, perhaps the genotypes of the parental animals had changed. This type of spontaneous change affecting the whole animal, or at least its gonads, is very unlikely, because even common experience tells us that organisms tend to be stable to their type.

Second, in the repeat, the sample of 22 F_2 animals did not contain any yellow fat simply by chance ("bad luck"). This explanation, again, seems unlikely, because the sample was quite large, but it is a definite possibility.

A third explanation draws on the principle covered in Chapter 1 that genes do not act in a vacuum; they depend on the environment for their effects. Hence, the formula "Genotype + environment = phenotype" is a useful mnemonic. A corollary of this formula is that genes can act differently in different environments; so

genotype 1 + environment 1 = phenotype 1

and

genotype 1 + environment 2 = phenotype 2

In the present problem, the different diets constituted different environments, and so a possible explanation of the results is that the recessive allele *w* produces yellow fat only when the diet contains fresh vegetables. This explanation is testable. One way to test it is to repeat the experiment again and use vegetables as food, but the parents might be dead by this time. A more convincing way is to breed several of the white-fatted F_2 rabbits from the second experiment. According to the original interpretation, some of them should be heterozygous, and, if their progeny are raised on vegetables, yellow fat should appear in Mendelian proportions. For example, if a cross happened to be *W*/*w* and *w*/*w*, the progeny would be $\frac{1}{2}$ white fat and $\frac{1}{2}$ yellow fat.

If this outcome did not happen and no progeny having yellow fat appeared in any of the matings, we would be forced back to the first or second explanation. The second explanation can be tested by using larger numbers, and if this explanation doesn't work, we are left with the first explanation, which is difficult to test directly.

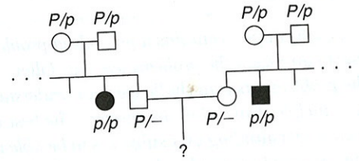
As you might have guessed, in reality, the diet was the culprit. The specific details illustrate environmental effects beautifully. Fresh vegetables contain yellow substances called xanthophylls, and the dominant allele *W* gives rabbits the ability to break down these substances to a colorless ("white") form. However, *w*/*w* animals lack this ability, and the xanthophylls are deposited in the fat, making it yellow. When no xanthophylls have been ingested, both *W*/– and *w*/*w* animals end up with white fat.

Solved problem 2. Phenylketonuria is a human hereditary disease resulting from the inability of the body to process the chemical phenylalanine, which is contained in the protein that we eat. PKU is manifested in early infancy and, if it remains untreated, generally leads to mental retardation. PKU is caused by a recessive allele with simple Mendelian inheritance.

A couple intends to have children but consults a genetic counselor because the man has a sister with PKU and the woman has a brother with PKU. There are no other known cases in their families. They ask the genetic counselor to determine the probability that their first child will have PKU. What is this probability?

SOLUTION

What can we deduce? If we let the allele causing the PKU phenotype be *p* and the respective normal allele be *P*, then the sister and brother of the man and woman, respectively, must have been *p*/*p*. To produce these affected persons, all four grandparents must have been heterozygous normal. The pedigree can be summarized as follows:



When these inferences have been made, the problem is reduced to an application of the product rule. The only way in which the man and woman can have a PKU child is if both of them are heterozygotes (it is obvious that they themselves do not have the disease). Both the grandparental matings are simple Mendelian monohybrid crosses expected to produce progeny in the following proportions:

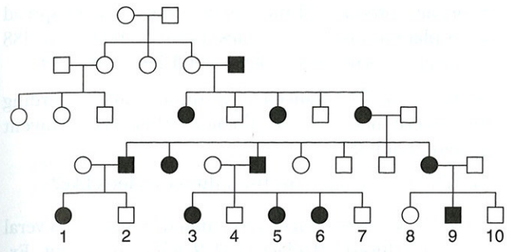
$\left.\begin{matrix}\frac{1}{4}\,P/P\\ \frac{1}{2}\,P/p\end{matrix}\right\}$ Normal $(\frac{3}{4})$

$\frac{1}{4}\,p/p$ PKU$(\frac{1}{4})$

We know that the man and the woman are normal, and so the probability of each being a heterozygote is 2/3 because, within the *P*/– class, 2/3 are *P*/*p* and 1/3 are *P*/*P*.

The probability of *both* the man and the woman being heterozygotes is 2/3 × 2/3 = 4/9. If both are heterozygous, then one-quarter of their children would have PKU, and so the probability that their first child will have PKU is 1/4 and the probability of their being heterozygous *and* of their first child's having PKU is 4/9 × 1/4 = 4/36 = 1/9, which is the answer.

Solved problem 3. A rare human disease afflicted a family as shown in the accompanying pedigree.



a. Deduce the most likely mode of inheritance.

b. What would be the outcomes of the cousin marriages 1 × 9, 1 × 4, 2 × 3, and 2 × 8?

SOLUTION

a. The most likely mode of inheritance is X-linked dominant. We assume that the disease phenotype is dominant because, after it has been introduced into the pedigree by the male in generation II, it appears in every generation. We assume that the phenotype is X linked because fathers do not transmit it to their sons. If it were autosomal dominant, father-to-son transmission would be common.

In theory, autosomal recessive could work, but it is improbable. In particular, note the marriages between affected members of the family and unaffected outsiders. If the condition were autosomal recessive, the only way in which these marriages could have affected offspring is if each person marrying into the family were a heterozygote; then the matings would be *a*/*a* (affected) × *A*/*a* (unaffected). However, we are told that the disease is rare; in such a case, heterozygotes are highly unlikely to be so common. X-linked recessive inheritance is impossible, because a mating of an affected woman with a normal man could not produce affected daughters. So we can let *A* represent the disease-causing allele and *a* represent the normal allele.

b. 1 × 9: Number 1 must be heterozygous *A*/*a* because she must have obtained *a* from her normal mother. Number 9 must be *A*/Y. Hence, the cross is *A*/*a* ♀ × *A*/Y ♂.

Female gametes	Male gametes	Progeny
$\frac{1}{2}\,A$	$\frac{1}{2}\,A$	$\frac{1}{4}\,A/A$ ♀
	$\frac{1}{2}\,Y$	$\frac{1}{4}\,A/Y$ ♂
$\frac{1}{2}\,a$	$\frac{1}{2}\,A$	$\frac{1}{4}\,A/a$ ♀
	$\frac{1}{2}\,Y$	$\frac{1}{4}\,a/Y$ ♂

1 × 4: Must be *A*/*a* ♀ × *a*/Y ♂.

Female gametes	Male gametes	Progeny
$\frac{1}{2}\,A$	$\frac{1}{2}\,a$	$\frac{1}{4}\,A/a$ ♀
	$\frac{1}{2}\,Y$	$\frac{1}{4}\,A/Y$ ♂
$\frac{1}{2}\,a$	$\frac{1}{2}\,a$	$\frac{1}{4}\,a/a$ ♀
	$\frac{1}{2}\,Y$	$\frac{1}{4}\,a/Y$ ♂

2 × 3: Must be *a*/Y ♂ × *A*/*a* ♀ (same as 1 × 4).

2 × 8: Must be *a*/Y ♂ × *a*/*a* ♀ (all progeny normal).

Problems

BASIC PROBLEMS

1. Make up a sentence including the words *chromosome*, *genes*, and *genome*.
2. Peas are diploid and $2n = 14$. *Neurospora* is haploid fungus and $n = 7$. If it were possible to fractionate genomic DNA from both by using pulsed field electrophoresis, how many distinct DNA bands would be visible in each species?
3. The broad bean (*Vicia faba*) is diploid and $2n = 18$. Each haploid chromosome set contains approximately 4 m of DNA. The average size of each chromosome during metaphase of mitosis is 13 μm. What is the average packing ratio of DNA at metaphase? (Packing ratio = length of chromosome/length of DNA molecule therein.) How is this packing achieved?
4. If we call the amount of DNA per genome "*x*," name a situation or situations in diploid organisms in which the amount of DNA per cell is
   a. x **b.** $2x$ **c.** $4x$
5. Name the key function of mitosis.
6. Name two key functions of meiosis.
7. Can you design a different nuclear-division system that would achieve the same outcome as that of meiosis?
8. In a possible future scenario, male fertility drops to zero, but, luckily, scientists develop a way for women to produce babies by virgin birth. Meiocytes are converted directly (without undergoing meiosis) into zygotes, which implant in the usual way. What would be the short- and long-term effects in such a society?