# 2 Transmission Genetics: Heritage from Mendel

## Key Concepts

- Inherited traits are determined by the genes present in the reproductive cells united in fertilization.
- Genes are usually inherited in pairs, one from the mother and one from the father.
- The genes in a pair may differ in DNA sequence and in their effect on the expression of a particular inherited trait.
- The maternally and paternally inherited genes are not changed by being together in the same organism.
- In the formation of reproductive cells, the paired genes separate again into different cells.
- Random combinations of reproductive cells containing different genes result in Mendel's ratios of traits appearing among the progeny.
- The ratios actually observed for any traits are determined by the types of dominance and gene interaction.

## Key Terms

1. allele
2. wildtype
3. genotype
4. segregation
5. testcross
6. independent assortment
7. incomplete dominance
8. pedigree
9. sibling
10. epistasis
11. complementation test
12. variable expressivity

## Concepts in Action

**2.1.** Any number of different alleles may exist at the same time in the population. A single diploid organism can have only two alleles of the same gene, one inherited from the mother and one from the father.

**2.2.**

| Parents | Progeny | | |
|---|---|---|---|
| | *AA* | *Aa* | *aa* |
| *AA* × *AA* | 1 | 0 | 0 |
| *AA* × *Aa* | 1/2 | 1/2 | 0 |
| *AA* × *aa* | 0 | 1 | 0 |
| *Aa* × *Aa* | 1/4 | 1/2 | 1/4 |
| *Aa* × *aa* | 0 | 1/2 | 1/2 |
| *aa* × *aa* | 0 | 0 | 1 |

**2.3.** *Ww*; 1/4 wrinkled seeds are expected.

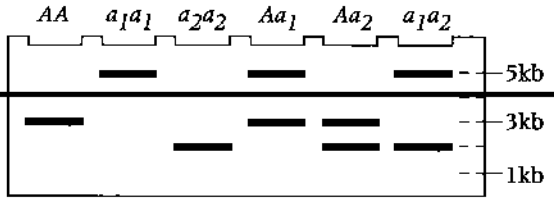
**2.4.** The birth of an affected offspring implies that parents are at risk of another affected offspring. Once the genotypes of the parents have been deduced from their own phenotypes and the fact that they have an affected offspring, the recurrence risk is calculated as the probability of an affected offspring in the next birth. **(a)** The mating must be *Aa* × *aa*, and so the risk of an *Aa* offspring in any birth (and therefore the recurrence risk) is 1/2. **(b)** The mating must be *Aa* × *Aa*, and so the risk of an *aa* offspring in any birth is 1/4. **(c)** The mating must be *Aa* × *Aa*, and so the risk of an *aa* offspring in any birth is 1/2.

**2.5.** Sixteen possible types of gametes [all possible combinations with one each of (*A*, *a*), (*B*, *b*), (*C*, *c*), and (*D*, *d*), with an expected proportion of 1/16 each.

**2.6.** **(a)** II2 is the affected person, and so the genotype must be *aa*. **(b)** I-1 and I-2 are the parents of the affected person and because neither is affected, their genotypes must both be *Aa*. **(c)** II-1 and II-3 are siblings of the affected person. Because they are not affected, and because they result from the mating *Aa* × *Aa*, their possible genotypes are *AA* and *Aa*. **(d)** From the mating *Aa* × *Aa*, the ratio of *AA*:*Aa* is 1:2; hence, the probability that II-3 is a carrier (genotype *Aa*) equals 2/3.

**2.7.** $(2/3)^2 = 4/9$; $(1/3)^2 = 1/9$; $1 - (1/3)^2 = 8/9$

**2.8.** The DNA fragment from $a_1$ with the 2-kb insertion results in a band at 3 kb + 2 kb = 5 kb, and the DNA fragment from $a_2$ with the 1-kb deletion results in a band at 3 kb-1 kb = 2 kb. DNA from each homozygous genotypes produces only one band, whereas that from each heterozygous genotype produces two bands.




**2.9.** The recessive mutations are in different genes. They are not alleles. This situation is an example of complementation in human beings.

**2.10.** The existing data enable us to group the mutants into three complementation groups as follows: {*a*, *c*, *d*}, {*b*, *f*}, and {*e*}. The missing entries are shown in the accompanying table.

| | *a* | *b* | *c* | *d* | *e* | *f* |
|---|---|---|---|---|---|---|
| *a* | ⊖ | + | − | ⊖ | + | ⊕ |
| *b* | ⊖ | ⊕ | ⊕ | ⊕ | − | |
| *c* | ⊖ | − | ⊕ | ⊕ | | |
| *d* | ⊖ | ⊕ | ⊕ | | | |
| *e* | ⊖ | + | | | | |
| *f* | ⊖ | | | | | |

**2.11.** A 15:1 ratio is characteristic of two genes with independent assortment in which the minority trait is due to a double-homozygous recessive. Suppose that the genotype *aa bb* results in ovoid-shaped fruit and that all other genotypes result in triangular fruit. The true-breeding parents would then have been *aa BB* and *AA bb*. One test of this hypothesis is that the ovoid $F_2$ plants should be true-breeding. Furthermore, crossing the $F_1$ triangular plants (*Aa Bb*) with the $F_2$ ovoid plants (*aa bb*) should yield progeny with the phenotypic ratio 3 triangular:1 ovoid.

**2.12.** We expect a 3:1 ratio of the dominant to the recessive phenotype when two heterozygous genotypes are crossed, but in this case we observe 2:1. A cross of two *Cy*/+ heterozygotes is expected to yield a progeny genotypic ratio of 1 *Cy*/*Cy*:2 *Cy* /+: 1 +/+. Because we only see two curly-winged flies for every wildtype fly, one possible explanation is that the *Cy*/*Cy* homozygotes die. In other words, *Cy* is dominant with respect to wing phenotype but recessive with respect to lethality.

**2.13.** **(a)** Two phenotypic classes are expected for each of the *Aa* and *Bb* pairs of alleles, and three are expected for the *Rr* pair, yielding a total number of 12. **(b)** 1/64. **(c)** 1/8.

**2.14.** Let *AA BB* represent the wildtype genotype with a phenotype of a disc shape. We are told that genotypes *aa BB* and *Aa bb* both have spherical fruit and that *aa bb* has elongated fruit. The $F_2$ progeny are expected in the ratio 9 *A– B–*:3 *A– bb*:3 *aa B–*: 1 *aa bb*, which implies that the expected ration of fruit-shaped phenotypes is 9 disc: 6 sphere:1 elongate.

**2.15.** $1/2 \times 1/2 \times 1/4 = 1/16$.

**2.16.** **(a)** Because the trait is rare, it is reasonable to assume that the affected father is heterozygous *HD/hd*, where *hd* represents the normal allele. Half of the father's gametes contain the mutant *HD* allele, so the probability is 1/2 that the son received the allele and will later develop the disorder. **(b)** We do not know whether the son is heterozygous *HD/hd*, but the probability is 1/2 that he is; if the son is heterozygous, half of his gametes will contain the *HD* allele. Therefore, the overall probability that the grandchild has the *HD* allele is $(1/2) \times (1/2) = 1/4$.

**2.17.** **(a)** In approaching this kind of problem, note first that the families with at least one boy include all families except those with all girls. Therefore, the simplest way to obtain the answer is to calculate the proportion of families with four girls and then subtract this from 1. The probability of having four girls is $(1/2)^4$, which equals 0.0625; the rest of the families are those with at least one boy, which account for the proportion $1 - 0.0625 = 0.9375$ of all families. **(b)** Because a particular birth order is specified, the answer is $(1/2)^4 = 1/16$. To look at the problem in another way, note that although 6/16 of all families with four children have two boys and two girls, only 1/6 of such families have the specific birth order FMFM; hence, the answer is, as before, 1/16.

**2.18.** In the functional female gametes, the ratio of *A*:*a* is 1/2:1/2 because of Mendelian segregation. In males, the products of meiosis in an *Aa* individual also consist of *A* + *A* + *a* + *a*, but as stated in the problem, half of the *A*-bearing products are nonfunctional. Hence, each male meiosis produces, on the average, three functional products, namely *A* + *a* + *a*. The ratio of *A*:*a* among functional male gametes is therefore 1:2 or, converting to proportions, 1/3 *A*:2/3 *a*. The Punnet square shown here indicates that the $F_2$ ratio of *AA*:*Aa*:*aa* is 1/6:3/6:2/6 (or, reducing the fractions, 1/6:1/2:1/3).

| | | Eggs | |
|---|---|---|---|
| | | 1/2 *A* | 1/2 *a* |
| Pollen | 1/3 *A* | 1/6 *AA* | 1/6 *Aa* |
| | 2/3 *a* | 2/6 *Aa* | 2/6 *aa* |

**2.19.** **(a)** The trait is more likely to be due to a recessive allele because there is consanguinity (mating between relatives) in the pedigree. **(b)** The double line indicates consanguineous mating. **(c)** III-1 and III-2 are first cousins. **(d)** Either I-1 or I-2 are

likely to be heterozygous *Aa* (but not both, because the trait is said to be rare), and all of II-2, II-3, III-1, and III-2 are likely to be *Aa*. On the other hand, II-1 and II-4 are most likely to be *AA*.

**2.20.** The accompanying Punnet squares indicate that the expected ratios of genotypes in **(a)** are 9/16 DD: 6/16 *Dd*: 1/16 *dd*, an in **(b)** are 3/8 *DD*: 4/8 *Dd*: 1/8 *dd*.

**(a)**

| Sperm \ Eggs | 3/4 *D* | 1/4 *d* |
|---|---|---|
| 3/4 *D* | 9/16 *DD* | 3/16 *Dd* |
| 1/4 *d* | 3/16 *Dd* | 1/16 *dd* |

**(b)**

| Sperm \ Eggs | 3/4 *D* | 1/4 *d* |
|---|---|---|
| 1/2 *D* | 3/8 *DD* | 1/8 *Dd* |
| 1/2 *d* | 3/8 *Dd* | 1/8 *dd* |

## Study Questions

**2.S1.** Two parents with blood types A and B have a child who has the O blood type. What is the chance that their next child will be O?

**A)** 0
**B)** 1/2
**C)** 1/4
**D)** 1

**2.S2.** In the cross Aa Bb Cc Dd Ee × Aa Bb Cc Dd Ee, in which all genes undergo independent assortment, what proportion of offspring are expected to be homozygous dominant for all four genes?

**A)** 1/2
**B)** 1/5
**C)** $(1/2)^5$
**D)** $(1/4)^4$
**E)** $(1/4)^5$

**2.S3.** Among sibships consisting of five children, and assuming a sex ratio of 1:1, what is the proportion with no girls?

**A)** 1/2
**B)** $(1/2)^5$
**C)** $(1/5)^5$
**D)** $1 - (1/2)^5$

**2.S4.** Assuming equal sex ratios, if a mating has already produced 5 boys, what is the probability that the next child will be a boy?

**A)** 0
**B)** 1
**C)** 1/2
**D)** $(1/2)^3$
**E)** $1 - (1/2)^3$

**2.S5.** Assuming independent assortment, how many different gametes can be formed by an organism that is homozygous for 7 and heterozygous for 2 genes?

**A)** 2
**B)** 4
**C)** $3^2$
**D)** 5
**E)** 6

**2.S6.** In genetic analysis, the complementation test is used to determine whether two recessive mutations that cause similar phenotype are _____ of the same gene.

**2.S7.** Consider a family with six children, and remember that each birth is equally likely to result in a boy or a girl. What proportion of sibships will include at least one girl?

**2.S8.** A normal woman has a brother, who is albino (a trait determined by a rare recessive autosomal allele). What is the probability that her phenotypically normal son is heterozygous for the gene?

**2.S9.** In a testcross of Aa BB Cc Dd Ee FF, where the genes show independent assortment:

**a.** What is the expected frequency of aa bb Cc dd ee Ff progeny?

**b.** What is the expected frequency of progeny that are heterozygous for all six genes?

**2.S10.** Assuming sex ratio of 1:1,

**a.** What is the probability that a couple will have eight boys?

**b.** If they already have seven boys, what is the probability that the eighth child will be a boy?

# The Chromosomal Basis of Heredity

# 3

## Key Concepts

- Chromosomes in eukaryotic cells are usually present in pairs.
- The chromosomes of each pair separate in meiosis, one going to each gamete.
- In meiosis, the chromosomes of different pairs undergo independent assortment.
- Chromosomes consist largely of DNA combined with histone proteins.
- In many animals, sex is determined by a special pair of chromosomes, the X and Y.
- Irregularities in the inheritance of an X-linked gene in *Drosophila* gave experimental proof of the chromosomal theory of heredity.
- The progeny of genetic crosses follow the binomial probability formula.
- The chi-square statistical test is used to determine how well observed genetic data agree with expectations from a hypothesis.

## Key Terms

1. synapsis
2. kinetochore
3. diplotete
4. chiasma
5. telomerase
6. anaphase II
7. histone
8. chromosome territory
9. heterochromatin
10. nondisjunction
11. chi-square
12. significant

## Concepts in Action

**3.1.** It means that, if there is no crossing over between a gene and the centromere, homologous alleles are separated from one another at anaphase I; if there is crossing over between a gene and the centromere, they are separated at anaphase II. In either case, they undergo segregation (separation) at one of the anaphase divisions.

**3.2.** After one cell cycle carried out in the presence of colchicine, a human cell would be expected to have $46 \times 2 = 92$ chromosomes.

**3.3.** It means that nonhomologous chromosomes have no influence on each other's orientation as they align on the metaphase plate at metaphase I. Hence, genes on nonhomologous chromosomes are independent in whether or not they proceed to the same anaphase pole, which is equivalent to independent assortment.

**3.4.** After the centromeres have split, each former sister chromatid is counted as a chromosome in its own right, so at anaphase there are 48 chromosomes.