MATH 448 — Lecture 14

§6.1: Mendelian genetics

Mendel's experiment. A strain of pea plants is *true-breeding* for a trait if every offspring of two plants of the strain shows the trait. Cross a true-breeding tall plant with a true-breeding dwarf plant. The offspring, called the F_1 *generation*, are all tall. Cross F_1 plants with each other. The offspring, the F_2 *generation*, contain dwarf plants again: Mendel counted 787 tall and 277 dwarf, close to the ratio $3:1$. For each of seven traits (for example round versus wrinkled seeds, with counts $5474 : 1850$), one form appeared in all of F_1 and in about $\frac{3}{4}$ of F_2. Mendel proposed a model that produces these numbers.

Mendel's model for one gene

A *gene* controls a trait, and its versions are its *alleles*: D (tall) and d (dwarf).

(M1) A plant has two alleles of the gene, its *genotype*: DD, Dd, or dd. A genotype with two equal alleles is *homozygous*, with two different alleles *heterozygous*. The observable trait is the *phenotype*. The allele D is *dominant* and d is *recessive*: DD and Dd are tall, dd is dwarf.

(M2) *Segregation.* A plant forms *gametes* (egg and pollen cells), each carrying exactly one of its two alleles, each with probability $\frac{1}{2}$.

(M3) *Random union.* An offspring is formed from one gamete of each parent, and the two gametes are independent.

One-gene crosses. A DD plant forms only D gametes and a dd plant only d gametes, so every offspring of $DD \times dd$ is Dd, hence tall. These are the F_1 plants. For the cross $Dd \times Dd$ of two F_1 plants, each parent forms D and d gametes with probability $\frac{1}{2}$ each. By (M3), $P(dd) = P(d \text{ from mother})\, P(d \text{ from father}) = \frac{1}{2} \cdot \frac{1}{2} = \frac{1}{4}$, and similarly $P(DD) = \frac{1}{4}$. A Dd offspring arises from D and d in either order, so $P(Dd) = \frac{1}{4} + \frac{1}{4} = \frac{1}{2}$. A *Punnett square* displays this: the possible gametes of the two parents label the rows and columns, and each cell holds the genotype formed by its row and column gametes.

	D	d
D	DD	Dd
d	Dd	dd

The four cells are equally likely, so the genotypes $DD : Dd : dd$ occur in the ratio $1:2:1$. The phenotype tall has probability $P(DD) + P(Dd) = \frac{3}{4}$, and dwarf has probability $\frac{1}{4}$. This is the observed $3:1$ ratio.

Two genes. Let W (round) be dominant to w (wrinkled). Mendel assumed *independent assortment*: the allele a plant passes on for one gene is independent of the allele it passes on for the other gene. A $DdWw$ plant then forms the four gametes DW, Dw, dW, dw with probability $\frac{1}{4}$ each. If A is an event that depends only on the first gene of the offspring and B an event that depends only on the second, independent assortment together with (M3) gives

$$P(A \cap B) = P(A)\, P(B). \tag{14.1}$$

A cross of n genes can therefore be computed as n one-gene crosses. For parents heterozygous for all n genes, the Punnett square has $2^n \times 2^n$ cells.

In the cross $DdWw \times ddWw$, the first parent forms the four gametes above. The second parent forms dW and dw with probability $\frac{1}{2}$ each, and these are listed twice so that the four columns are equally likely.

	dW	dw	dW	dw
DW	$DdWW$	$DdWw$	$DdWW$	$DdWw$
Dw	$DdWw$	$Ddww$	$DdWw$	$Ddww$
dW	$ddWW$	$ddWw$	$ddWW$	$ddWw$
dw	$ddWw$	$ddww$	$ddWw$	$ddww$

Counting cells, tall round occurs in 6, tall wrinkled in 2, dwarf round in 6, and dwarf wrinkled in 2 of the 16 cells.

Problem 14.1. If offspring received all the genes of both parents instead of one allele from each, and each plant of the founding generation F_0 has two copies of a gene, how many copies does a plant of F_n have?

Problem 14.2. A *testcross* crosses a plant of unknown genotype with a dd plant. (a) What offspring phenotypes can a testcross of a DD plant produce, and of a Dd plant? (b) A tall plant is chosen at random from the F_2 generation of $Dd \times Dd$. What is the probability that it is Dd? (c) This plant is testcrossed. What is the probability that the first offspring is dwarf?

Problem 14.3. Find the probability of each of the four phenotypes in the cross $DdWw \times ddWw$ using (14.1), and compare with the table.

Problem 14.4. Mendel followed seven traits. Two plants are heterozygous for all seven genes, and the genes assort independently. (a) What is the probability that an offspring shows the dominant phenotype for all seven traits? The recessive phenotype for all seven? (b) How many cells does the Punnett square have? (c) For $AaBbCC \times AaBbCC$, how many different gametes can a parent form, and how many offspring genotypes and phenotypes are possible?

Problem 14.5. About 1 in 31 adults in a certain population is heterozygous for the recessive Tay–Sachs allele. A child with two copies develops the disease. (a) Two adults are drawn at random from this population. What is the probability that their child has the disease? (b) A couple's first child has the disease. What is the probability that their second child has it?

Answers. 14.1: 2^{n+1}. 14.2: (a) DD: all tall, Dd: half tall and half dwarf; (b) $\frac{2}{3}$; (c) $\frac{1}{3}$. 14.3: tall round $\frac{3}{8}$, tall wrinkled $\frac{1}{8}$, dwarf round $\frac{3}{8}$, dwarf wrinkled $\frac{1}{8}$, in agreement with the table. 14.4: (a) $(\frac{3}{4})^7 = \frac{2187}{16384} \approx .13$ and $(\frac{1}{4})^7 = \frac{1}{16384}$; (b) $128^2 = 16384$; (c) 4 gametes, 9 genotypes, 4 phenotypes. 14.5: (a) $\frac{1}{3844}$; (b) $\frac{1}{4}$.