Genetic transmission in populations

Introduction

Mendel's rules describe how genetic transmission happens between parents and offspring. Consider a monohybrid cross:

$$A_1A_2 \times A_1A_2$$
$$\downarrow$$
$$\tfrac{1}{4}A_1A_1 \quad \tfrac{1}{2}A_1A_2 \quad \tfrac{1}{4}A_2A_2$$

Population genetics describes how genetic transmission happens between a *population* of parents and a population of offspring. Consider, for example, the following data from the *Est*-3 locus of *Zoarces viviparus*:[1]

	Genotype of offspring		
Maternal genotype	A_1A_1	A_1A_2	A_2A_2
A_1A_1	305	516	
A_1A_2	459	1360	877
A_2A_2		877	1541

This table describes, empirically, the relationship between the genotypes of mothers and the genotypes of their offspring. We can also make some inferences about the genotypes of the fathers in this population, even though we didn't see them.

1. 305 out of 821 male gametes that fertilized eggs from A_1A_1 mothers carried the A_1 allele (37%).

2. 877 out of 2418 male gametes that fertilized eggs from A_2A_2 mothers carried the A_1 allele (36%).

Question How many of the 2,696 male gametes that fertilized eggs from A_1A_2 mothers carried the A_1 allele?

[1] Only one offspring from each mother is included in these data (from [1]).

Recall We don't know the paternal genotypes or we wouldn't be asking this question.

- There is no way to tell which of the 1360 A_1A_2 offspring received A_1 from their mother and which from their father.
- Regardless of what the genotype of the father is, half of the offspring of a heterozygous mother will be heterozygous.[2]
- Heterozygous offspring of heterozygous mothers contain no information about the frequency of A_1 among fathers, so we don't bother to include them in our calculations.

Rephrase How many of the 1336 homozygous progeny of heterozygous mothers received an A_1 allele from their father?

Answer 459 out of 1336 (34%)

New question How many of the offspring where the paternal contribution can be identified received an A_1 allele from their father?

Answer (305 + 459 + 877) out of (305 + 459 + 877 + 516 + 877 + 1541) or 1641 out of 4575 (36%)

An algebraic formulation of the problem

The above calculations tell us what's happening for this particular data set, but those of you who know me know that there has to be a little math coming so that we can describe the situation more generally. Here's the notation:

Genotype	Number	Sex
A_1A_1	F_{11}	female
A_1A_2	F_{12}	female
A_2A_2	F_{22}	female
A_1A_1	M_{11}	male
A_1A_2	M_{12}	male
A_2A_2	M_{22}	male

[2]Assuming we're looking at data from a locus that has only two alleles. If there were four alleles at a locus, for example, *all* of the offspring could be heterozygous. If you don't see that immediately, think about an A_1A_2 mother mating with an A_3A_4 father and write out the Punnett square. We are also assuming that meiosis is fair, i.e., that there's no segregation distortion, and that there's no gamete competition and no gamete-gamete assortative mating. If you don't know what any of that means don't worry about it. We'll get to (most of) it over the next few weeks.

Using that notation,

$$p_f = \frac{2F_{11}+F_{12}}{2F_{11}+2F_{12}+2F_{22}} \qquad q_f = \frac{2F_{22}+F_{12}}{2F_{11}+2F_{12}+2F_{22}}$$

$$p_m = \frac{2M_{11}+M_{12}}{2M_{11}+2M_{12}+2M_{22}} \quad q_m = \frac{2M_{22}+M_{12}}{2M_{11}+2M_{12}+2M_{22}} \quad ,$$

where p_f is the frequency of A_1 in mothers and p_m is the frequency of A_1 in fathers.[3]

Since every individual in the population must have one father and one mother, the frequency of A_1 among offspring is the same in both sexes, namely

$$p = \frac{1}{2}(p_f + p_m) \quad ,$$

assuming that all matings have the same average fecundity and that the locus we're studying is autosomal.[4]

Question: Why do those assumptions matter?

Answer: If $p_f = p_m$, then the allele frequency among offspring is equal to the allele frequency in their parents, i.e., the allele frequency doesn't change from one generation to the next. This might be considered the First Law of Population Genetics: If no forces act to change allele frequencies between zygote formation and breeding, allele frequencies will not change.

Zero force laws

This is an example of what philosophers call a **zero force law**. Zero force laws play a very important role in scientific theories, because we can't begin to understand what a force does until we understand what would happen in the absence of any forces. Consider Newton's famous dictum:

> An object in motion tends to remain in motion in a straight line. An object at rest tends to remain at rest.

or (as you may remember from introductory physics)[5]

$$F = ma \quad .$$

[3] $q_f = 1 - p_f$ and $q_m = 1 - p_m$ as usual.

[4] And that there are enough offspring produced that we can ignore genetic drift. Have you noticed that I have a fondness for footnotes? You'll see a lot more before the semester is through, and you'll soon discover that most of my weak attempts at humor are buried in them.

[5] Don't worry if you're not good at physics. I'm probably worse. What I'm about to tell you is almost the only thing about physics I can remember. I graduated from college with only one semester of college physics, and it didn't even require calculus as a pre-requisite.

If we observe an object accelerating, we can immediately infer that a force is acting on it. Not only that, we can also infer something about the magnitude of the force. **However**, if an object is not accelerating we cannot conclude that no forces are acting. It might be that opposing forces act on the object in such a way that the resultant is no *net* force. Acceleration is a *sufficient* condition to infer that force is operating on an object, but it is not *necessary*.

What we might call the "First Law of Population Genetics" is analogous to Newton's First Law of Motion:

> If all genotypes at a particular locus have the same average fecundity and the same average chance of being included in the breeding population, allele frequencies in the population will remain constant from one generation to the next.

For the rest of the semester we'll be learning about the forces that cause allele frequencies to change and learning how to infer the properties of those forces from the changes that they induce. But you must always remember that while we can infer that some evolutionary force is present if allele frequencies change from one generation to the next, we *cannot* infer the absence of a force from a lack of allele frequency change.

References

[1] F B Christiansen. Studies on selection components in natural populations using population samples of mother-offspring combinations. *Hereditas*, 92:199–203, 1980.

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