

205. POPULATION GENETICS

Evolution can be defined as the change in allele frequencies in a population. For this definition to be useful, we must also define the terms "allele frequency" and "population". A population is a group of freely interbreeding individuals. Alleles are different forms of the same gene. For example, different alleles of a gene that controls the composition of a sugar chain on the surface of red blood cells determine an individual's ABO blood type (see Box 1).

Box 1. Genetics of Human Blood Groups

Human ABO blood types depend on which of three alleles (I^A , I^B , or I^O) of a particular gene are present. This gene codes determines which of two sugars will be found in a chain of sugars on the cell surface (including red blood cells). The I^A allele results in one sugar being present, the I^B allele results in another sugar. The I^O allele produces neither sugar. Therefore, individuals with the genotypes $I^A I^A$ or $I^A I^O$ are blood group A, with type A sugar chains on their cells. Individuals with the genotypes $I^B I^B$ or $I^B I^O$ are blood group B. Individuals with the genotype $I^A I^B$ are blood group AB (both sugar chains are present), and those with the genotype $I^O I^O$ are blood O (neither sugar chain is present).

The ABO blood group needs to be matched for blood transfusions. The mammalian immune system produces antibodies (defensive proteins) against foreign macromolecules, including the blood group sugar chains on red blood cells. An individual with type A blood will form antibodies against the foreign B sugar chains, rejecting a transfusion of type B blood. On the other hand, an individual with type B blood will form antibodies against type A sugar chains in blood from an $I^A I^A$ or $I^A I^O$ individual. Blood group O individuals have been termed universal donors; because they lack the sugar chains in question, they can donate blood to types A, B, AB and O. Blood group AB individuals, "universal recipients", recognize both the A and B forms of the sugar chains and can safely accept blood from types A, B and O donors.

Additional cell surface molecules, including the Rh system, are also involved in recognition of self and rejection of foreign cells and tissues.

How do we expect allele frequencies to behave from generation to generation? Imagine that you have a large box of beads of different colors, 75% red, 15% blue and 10% white. You reach into the box and draw a single bead. What is the likelihood that that bead will be white? One in ten, or 10%. If you drew 200 beads from the box, you would expect about 20 of them (10%) to be white. Likewise, we expect the frequency of alleles in the gametes produced by a parental generation to be equal to the frequency of those alleles in the parental generation. This expectation was expressed mathematically by Hardy and Weinberg; populations in which allele frequencies are constant are said to be in Hardy-Weinberg equilibrium.

Population biologists begin with the assumption that populations are in Hardy-Weinberg equilibrium. They then check allele frequencies. If allele frequencies are changing, the biologist has an interesting puzzle: What is causing the changes in allele frequencies?

Hardy-Weinberg equilibrium depends on a large interbreeding population with random mating and no factor tending to change gene frequencies. The smaller the population size, the more quickly sheer chance will change gene frequencies. This is called genetic drift. If you draw 20 beads from the box, it is unlikely that exactly 15 of them will be red, three blue and two white. If

these 20 beads form the next generation, then allele frequencies have shifted. Extreme examples of chance influencing allele frequency are population bottlenecks and the founder effect. In a population bottleneck, some disaster nearly destroys a population, leaving a few individuals to regenerate the population. The genetic homogeneity of cheetahs indicates that they went through such a population bottleneck. The founder effect refers to colonization of new habitats, such as islands, by a few individuals from a larger population. A founder effect can be seen in the allele frequencies in Amish communities.

Allele frequencies can be changed by mutation, although usually the mutation frequency is too small to have much effect. Immigration can bring new alleles into the population. But the most significant effects on allele frequencies result from natural selection. Breeding is not random, and some individuals are more likely to survive to produce offspring than others.

Text References

The Mechanisms of Evolution, POH Chapter 18

Purpose

To compare phenotype frequencies in two human generations.

Materials: PTC tasting paper

Participation in this activity is voluntary, but the broader the participation, the larger will be our database and the more interesting our conclusions. Note that the record sheet does not identify individuals.

Record as much information as possible for your own generation (you and your siblings) and for your parents' generation (your parents, aunts and uncles). Since we are using these individuals only as samples of their generations, it does not matter whether they are biologically related to you.

The genes we will examine, and the phenotypes associated with their genotypes, are described below. Test each phenotype in class, observing yourself and your classmates until you feel comfortable recognizing the different types.

Bent little finger (*B* for bent is dominant over *b* for straight)

Lay your hands flat and relaxed on the table. If the last joint of the little finger bends inward, you have at least one *B* allele

Hitchhiker's thumb (*H* for straight is dominant over *h* for bent)

Hold your hand as though you were hitch-hiking. If the last joint of the thumb will bend back more than 60°, you are *hh*.

Mid-digital hair (*M*, allowing hair to be present, is dominant over *m* (no hair))

Use a hand lens or dissecting microscope to look for any hair on the middle segment of any finger. If any hair, no matter how fine, is present on any finger you are *M*.

Long palmar muscle (*L*, long palmar muscle absent, is dominant over *l*, long palmar muscle present)

The long palmar muscle can be detected by observing its tendon in the wrist. Turn your hand palm up, clench your fist, and flex your wrist so your clenched fingers move away from your

face. If you have a long palmar muscle, you will detect three tendons, otherwise only two will be present.

Interlacing fingers (C , left thumb over right thumb, is dominant over c , right thumb over left thumb)

Fold your hands together with your fingers comfortably interlaced. Check to see which thumb is on top.

Free or attached earlobes (Free earlobes, A , is dominant over attached earlobes, a)

If there is any sign of an earlobe hanging free, the individual is A __.

Widow's peak (Widow's peak, W , is dominant over smooth hairline, (w))

If the hairline dips to a point in the middle of the forehead, a widow's peak is present. This characteristic cannot be determined if male pattern baldness has changed the hairline.

Tongue rolling (Tongue-rolling, R , is dominant over inability to roll tongue, r)

Stick out your tongue and try to roll it into a tube.

PTC tasting (Tasting PTC, T , is dominant over the inability to taste it, t)

Chew a piece of PTC paper for a few seconds. If you can detect the bitter taste of PTC, you are T __.

Pigmented iris (Pigmented iris, P , is dominant over non-pigmented iris, p .)

Blue eyes have unpigmented irises and are blue because blue light is more readily reflected from the deeper regions of the iris than are longer wavelengths. Pigmentation results in green, hazel, or brown eyes, any shade other than blue.

ABO blood group (I^A and I^B are both dominant over I^O . I^A and I^B are codominant with each other.)

Record your blood ABO blood group from your medical history.

Red-green color-blindness (Color vision, C , is dominant over the inability to distinguish red from green, c , but this gene is sex-linked.)

Homozygous dominant (CC) or heterozygous (Cc) females have normal color vision. Only homozygous recessive (cc) females are color-blind. Since the gene is on the X chromosome, males have only one copy of this gene, obtained from their mother, and the genotype of this copy determines their phenotype.

Additional Activities

Determine the characteristics of your grandparents and/or children. Use the information to diagram a family tree show inheritance of the various traits.

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PRELAB PREPARATION:

1. The Hardy-Weinberg equilibrium states that, under certain conditions, the frequency of alleles and the frequency of genotypes remains constant. When there are two alleles at the same locus, the equation is:

$$p^2 + 2pq + q^2 = 1$$

Explain what the "*p*"s and "*q*"s stand for in the equation. How does the equation relate allele frequencies to genotype frequencies?

2. What factors might change allele frequencies?

RESULTS:

3. Fill out the data table as completely as possible for siblings in your generation and in your parents generation.

Directions: Record the generation (1 for parental, 2 for your own) in the first column. For each individual, record the sex (M or F) and the genotype as indicated below.

- a. Bent little finger
*B*_ bent, *bb* straight
- b. Hitchhiker's thumb
*H*_ straight, *hh* bent
- c. Mid-digital hair
*M*_ hair, *mm* no hair
- d. Long palmar muscle
*L*_ muscle absent, *ll* muscle present
- e. Interlacing fingers
*C*_ left thumb on top, *cc*, right thumb
on top
- f. Free or attached earlobes
*A*_ free earlobes, *aa* attached
earlobes
- g. Widow's peak
*W*_ widow's peak, *ww* smooth
hairline
- h. Tongue-rolling
*R*_ tongue-rolling, *rr* cannot roll
tongue
- i. PTC tasting
*T*_ PTC tasting, *tt* PTC non-tasting
- j. Pigmented iris
*P*_ pigmented iris, *pp* non-pigmented
iris
- k. ABO blood group
*I^a*_ Type A, *I^b*_, type B, *I^AI^B* Type
AB, *IOIO*, Type O
- l. Red-green color-blindness
In females, *C*_ normal color vision,
cc color blind. In males, *C*
normal color vision, *c* color
blind.

4. Tally the class results for each genotype for each generation. Did the frequency of homozygous recessives change for any of the traits? What would you have to do to determine if the population was in Hardy-Weinberg equilibrium?

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