

Human Phenotypes

Objectives

1. Determine the genotype for traits from observation of the individual.
2. Do genetics problems involving autosomal dominance, autosomal recessive, incompletely dominant, co-dominant and X-linked recessive alleles.

Introduction

A zygote receives 23 pairs of chromosomes when the gametes unite at fertilization. One of each pair is inherited from the father and the other from the mother. Thereafter, during cell division, each body cell receives copies of these chromosomes. Twenty-two pairs of the chromosomes are autosomes and one pair is the sex chromosomes.

Chromosomal inheritance has a marked effect on the general anatomy and physiology of an individual. Genes, the units of heredity that control specific characteristics, are arranged in a linear fashion along the chromosomes. Alternate forms of a gene having the same position (locus) on a pair of chromosomes and affecting the same trait are called alleles. Just as we inherit pairs of chromosomes, so we inherit pairs of alleles, alternate forms of a gene.

There are a significant number of traits that have little or no effect on our success as individuals or as a species. The traits we will be looking at today belong to this group; they have little significance in making us better or worse individuals but they to make us more interesting by making us more diverse. You will be looking at these traits in yourselves and trying to determine your genotype for each trait, generally in terms of dominant or recessive, although occasionally you will be able to determine if you are heterozygous. Work with your neighbor as your partner. For some of the traits you will need to make measurements on each other.

Determining Your Genotype and Phenotype

Read through the following descriptions of phenotypes and decide your phenotype. Suggest the possible genotype for your phenotype.

1. Shape of the face.

Most facial characteristics are affected by many different genes acting together (multiple factors or polygenic inheritance). For this trait we have simplified the nature of this trait by assuming it is the result of a single gene. Oval shape is dominant (needs only one dominant allele in a pair for the trait to be expressed). The genotype is either homozygous (both alleles are the same, either dominant or recessive) or heterozygous (one dominant allele and one recessive allele). Square shape is recessive (requires that both alleles are the recessive allele in the pair for the trait to be expressed). The genotype is homozygous.

Phenotype _____ Genotype _____

2. Cleft in the Chin

This trait has been well documented. Absence of a cleft is dominant and may be either homozygous or heterozygous. The presence of a cleft is recessive and represents a homozygous condition.

Phenotype _____ Genotype _____

3. Hair Curl

Curliness of hair is probably polygenic. To simplify we will assume it is the product of incomplete dominance of alleles of a single gene. Curly is homozygous. Wavy is heterozygous. Straight is homozygous.

Phenotype _____ Genotype _____

4. Hairline

A slight dip in the center of the hairline is called a widow's peak. A widow's peak is dominant and may be either homozygous or heterozygous. A straight hairline is recessive and homozygous.

Phenotype _____ Genotype _____

5. Eyebrow Size

Broad eyebrows are dominant and are either homozygous or heterozygous. Business increases with age. Slender eyebrows are recessive and homozygous.

Phenotype _____ Genotype _____

6. Eyebrow Shape

Eyebrows that are separated by a bare patch of skin is dominant and either homozygous or heterozygous. Eyebrows that are joined by a line of hairs is recessive and homozygous.

Phenotype _____ Genotype _____

7. Eyelash Length

Long eyelashes are dominant and homozygous or heterozygous. Short eyelashes are recessive and homozygous.

Phenotype _____ Genotype _____

8. Dimples

Dimples are dominant and homozygous or heterozygous. Absence of dimples is recessive and homozygous.

Phenotype _____ Genotype _____

9. Size of Nose

Nose size is probably polygenic. For simplicity, we will assume it is the result of incomplete dominance of two alleles of a single gene. Large is more than 38% of face length and homozygous. Medium is about 1/3 of face length and heterozygous. Small is less than 31% of face length and homozygous.

Phenotype _____ Genotype _____

10. Size of Ears

Ear size is probably polygenic. We will assume it is the result of incomplete dominance of one pair of alleles of a single gene. Large ears are 38% or more of the face length and homozygous. Medium sized ears are about 1/3 of the face length and heterozygous. Small ears are 31% or less of face length and homozygous.

Phenotype _____ Genotype _____

11. Earlobes

Definite free earlobes are dominant and homozygous or heterozygous. Attached earlobes are recessive and homozygous.

Phenotype _____ Genotype _____

12. Width of Mouth

Several factors govern mouth width, but we will assume it is the product of the incomplete dominance of a pair of alleles of a single gene. A wide mouth is 95% or more of interpupillary distance and is homozygous. A medium mouth is 70%-90% of interpupillary distance and is heterozygous. A small mouth is 65% or less of interpupillary distance and is homozygous. Try not to smile when being measured.

Phenotype _____ Genotype _____

13. Eye Shape

For the purposes of the lab assume that almond eye shape is dominant and homozygous or heterozygous. Round eyelid openings are recessive and homozygous. Remember, we are considering the openings only, not the eyeballs themselves, which are round.

Phenotype _____ Genotype _____

14. Eye Spacing

Eyes are considered close together if the distance between them is less than the eye width, corner to corner. We will assume that close eye spacing is homozygous. Medium spacing is defined as being equal to the eye width and is heterozygous. Wide eye spacing is defined as being greater than the eye width and is homozygous.

Phenotype _____ Genotype _____

15. Eye Size

We will assume that eye size is the result of incomplete dominance of a pair of alleles of a single gene. Large eyes (>30mm, corner to corner) are homozygous. Medium eyes (26-30mm, corner to corner) are heterozygous. Small eyes (<26mm, corner to corner) are homozygous.

Phenotype _____ Genotype _____

16. Eye Slant

Horizontal eyes, corner to corner, is dominant and either homozygous or heterozygous. Slanted eyes, outer corners up, is recessive and homozygous.

Phenotype _____ Genotype _____

17. Freckles on Cheeks

Freckles are dominant and homozygous or heterozygous. Lack of freckles is recessive and homozygous.

Phenotype _____ Genotype _____

18. Tongue Rolling

The ability to roll the tongue, side edges up, is a dominant trait and is either homozygous or heterozygous. The inability to roll the tongue is recessive and homozygous.

Phenotype _____ Genotype _____

19. Tongue Folding

The ability to fold the tip of the tongue backwards, without touching the roof of the mouth, is recessive and homozygous. The inability to fold the tongue as described is dominant and is either homozygous or heterozygous.

Phenotype _____ Genotype _____

20. Mid-digital Hair

The presence of hair on the back of the middle joint of the fingers is dominant and is either homozygous or heterozygous. The absence of hair on the back of the middle joint of the fingers is recessive and homozygous. You will need to look carefully as the hair can be very fine, particularly in women.

Phenotype _____ Genotype _____

21. Hitch-hiker's Thumb

A thumb that is straight when fully bent back is dominant and is either homozygous or heterozygous. A thumb in which the distal segment (furthest from the hand) forms an angle with the proximal segment is recessive and homozygous.

Phenotype _____ Genotype _____

22. Bent Little Finger

In the dominant condition the center line of the distal segment of the little finger bends slightly towards the ring finger. It is either homozygous or heterozygous. A perfectly straight little finger is recessive and homozygous.

Phenotype _____ Genotype _____

23. Interlaced Fingers

Fold your hands together, interlacing the fingers. Now look at your thumbs. Left over right is dominant and either homozygous or heterozygous. Right thumb over left is recessive and homozygous.

Phenotype _____ Genotype _____

24. Hair on the Back of the Hand

Hair on the back of the hand is dominant and either homozygous or heterozygous. Hairless hands are recessive and homozygous.

Phenotype _____ Genotype _____

25. Tendons of the Palmar Tendons

Clench the fist tightly and flex the hand. Feel the tendons in the wrist. You should be able to distinguish two, perhaps even three. Do this for both wrists. One wrist might have two tendons, the other three. Two tendons in both wrists is dominant. It is either homozygous or heterozygous. The appearance of a third tendon, even in just one wrist, is recessive and homozygous.

Phenotype _____ Genotype _____

Table 1

Characteristics	% of Class with trait
1. Shape of Face	
Oval, Dominant	
Square, Recessive	
2. Cleft in Chin	
Cleft, Recessive	
No Cleft, Dominant	
3. Hair Curl	
Curly, Homozygous	
Wavy, Heterozygous	
Straight, Homozygous	
4. Hairline	
Widow's Peak, Dominant	
Straight Line, Recessive	
5. Eyebrow Size,	
Broad, Dominant	
Slender, Recessive	
6. Eyebrow Shape	
Separate, Dominant	
Joined, Recessive	
7. Eyelash Length	
Long, Dominant	
Short, Recessive	
8. Dimples	
Dimples, Dominant	
No dimples, Recessive	
9. Size of Nose	
Large, Homozygous	
Medium, Heterozygous	
Small, Homozygous	
10. Size of Ears	
Large, Homozygous	
Medium, Heterozygous	
Small, Homozygous	
11. Earlobes	
Free, Dominant	
Attached, Recessive	
12. Mouth Width	
Wide, Homozygous	
Medium, Heterozygous	
Small, Homozygous	
13. Eye Shape	
Almond, Dominant	
Round, Recessive	
14. Eye Spacing	
Close, Homozygous	
Medium, Heterozygous	
Wide, Homozygous	
15. Eye Size	
Large, Homozygous	
Medium, Heterozygous	
Small, Homozygous	

16. Eye Slant	
Horizontal, Dominant	
Slant, Recessive	
17. Freckles on Cheeks	
Present, Dominant	
Absent, recessive	
18. Tongue Rolling	
Not able, Dominant	
Able, Recessive	
19. Tongue Folding	
Not able, Dominant	
Able, Recessive	
20. Mid-digital Hair	
Present, Dominant	
Absent, Recessive	
21. Hitch-hiker's Thumb	
Straight, Dominant	
Bent, recessive	
22. Bent Little Finger	
Bent, Dominant	
Straight, Recessive	
23. Interlaced Fingers	
Left over, Dominant	
Right over, Recessive	
24. Hair on back of Hand	
Present, Dominant	
Absent, recessive	
25. Tendons of Palmar Muscle	
Two, Dominant	
Three, Recessive	
26. Taste Test	
P.T.C.	
Sodium benzoate	
Thiourea	

Genetics Problems

The following set of genetics problems cover the full spectrum of different types of genetics problems you should be able to solve. Your professor may choose to have you solve these problems as a class exercise or a homework assignment. In addition, your professor may choose to have you answer all or some of these problems or include other problems at his or her discretion. As you work on these problems, check tables one and two at the end of the problems for some suggested symbols to use to represent the different alleles referred to in the questions.

I. Monohybrid Crosses

1. In sweet peas the gene for the red color in flowers (R) is dominant over the gene for the white flowers (r). If a homozygous red-flowered plant is crossed with a white-flowered plant, what will be the genotype of the offspring? What will be the result of the next generation if the first offspring (F₁) are allowed to self-pollinate?
2. A blue-eyed man, both of whose parents are brown-eyed, marries a brown-eyed woman whose father was brown-eyed and whose mother was blue-eyed. They had one child who is blue-eyed. What are the genotypes of all the members mentioned?
3. The ability to taste phenylthiocarbamide (PTC) in humans appears to be due to a dominant factor. These persons are called tasters. A man who is a taster, and who has a non-taster mother, marries a woman who is a taster. She has 3 brothers and 2 sisters, three of whom are non-tasters. What chances will the children of this marriage have of being non-tasters?
4. Twenty-five percent of the offspring of a certain pair of white sheep are black. What was the genotype of the parents?
5. A woman has a rare abnormality of the eyelids called ptosis, which makes it impossible for her to open her eyes completely. The condition is due to a dominant gene (P). The woman's father had ptosis, but her mother had normal eyelids. What are the genotypes of the woman, her father, and her mother? If she married a man with normal eyelids, what proportion of the children would be expected to have ptosis and what proportion would be normal?
6. Two dark-haired parents have a red-haired child. What genotype must the two parents have to permit this?
7. Assume that the dimple is inherited as a simple dominant gene. A dimpled man whose mother had no dimple married a woman with no dimple. What are the genotypes of the father, mother, F₁ generation, and F₂ generation?
8. In mice, yellow coat color (Z) is a dominant factor and black (z) is its recessive. The homozygous condition (ZZ) is lethal and kills the embryo within a few days after conception. If two yellow mice bred, what genotypes would you expect and in what ratio? What will be the phenotypes and in what ratio?
9. One variety of snapdragon with dominant yellow leaves always produce normal green leaf plants as well as yellow leaf heterozygous plants. The homozygous yellow leaf lack the ability to make chlorophyll and die as seedlings. Show the result of the cross of two yellow leaf plants.
10. Can wavy haired parents have a curly haired child?
11. Yellow guinea pigs crossed with white ones always produce cream-colored offspring. Matings between the two cream colored guinea pigs produced, in one case, 22 yellow, 47 cream, and 19 white. What type of inheritance is this?

II. Sex-linked Characteristics

12. In man, colorblindness, an inability to distinguish between red and green, is inherited as a sex-linked character on the X chromosome. It is recessive. If a colorblind man marries a homozygous normal woman, what chance is there for the children of being colorblind? If a normal man marries a colorblind woman, where will colorblindness be found in the family? Why is it so commonly believed that women cannot be colorblind?

13. Let us assume that a colorblind man has a colorblind sister and his mother has normal vision. What is the genotype, as to color vision, of this man's mother and of his father? What kind of color vision will his sons and his daughters have, and in what ratios, if he marries a woman identical in genotype to his mother?

14. In man, hemophilia (failure of blood to clot) is a recessive and sex-linked character. A man and a woman, both normal in respect to blood-clotting, have (a) a hemophilic son who has a daughter who is normal, (b) a daughter who has normal blood and who has one hemophilic and one normal son, and (c) another daughter with normal blood who has five sons, all normal. What are the probable genotypes of grandparents, children, and grandchildren?

15. In the fruit fly, red eye is dominant to white eye, which is sex-linked. Specify the distribution of eye color (a) among the males and (b) among the females, derived from each of the following crosses:

- a. white-eyed female x red-eyed male
- b. heterozygous red-eyed female x white-eyed male
- c. heterozygous red-eyed female x red-eyed male

III. Multiple Alleles

16. In man, blood groups are inherited on the basis of multiple alleles. Determine the genotypes of the following parents.

- a. Father group A, mother group B. All four groups are represented among the children.
- b. Father group AB, mother group B, 1/4 of children A, 1/4 AB, and 1/2 B.
- c. Both parents are A, 3/4 of the children are A, 1/4 type O.

17. Suppose a father and a mother claim that they have been given the wrong baby at the hospital. The mother belongs to group B, the child to AB, one possible father to B, the other to A. Which man is the most probable father? Who would be the most probable father if the mother belongs to group B, the child to O, the possible father to A, and the other to AB?

18. A man with normal vision and blood type B marries a woman with normal vision and blood type A. They have two children, a color-blind boy with blood type A and a girl with normal vision and blood type O. What are the genotypes of the parents?

19. An Rh⁺ man, type A blood, marries an Rh⁻ woman, type B blood. Their first child is Rh⁺ type A. Could the second child be Rh⁻, type O?

IV. Dihybrid Crosses

20. If a pure tall, red-flowered plant is crossed with a dwarf white-flowered plant, what will be the appearance of the members of the F₁ generation? What will the F₂ generation be like in appearance?

21. A man with short eyelashes and a Roman nose marries a woman with long eyelashes and a Roman nose. They have two children, one with long eyelashes and a straight nose and the other with short eyelashes and a Roman nose. By a later marriage with another woman, who also has long eyelashes and a Roman nose, the man has 9 children, all with long eyelashes and Roman noses. What are the genotypes of the man and his two wives?

22. It has been reported that the purple flower color in one species of the Japanese morning glory may be caused by dominant alleles at either one of two separate gene pairs, e.g., A-bb or aaB-. When dominant alleles are present at both gene pairs (A-B-), the color of the flower is blue, and when both are homozygous recessive (aabb), the color is scarlet.

If AA^{bb} was crossed with aaBB the flower color would be blue. If these blue flowers (F₁) were crossed, what would be the phenotype and genotype of the F₂? If the F₁ were crossed to either of the parents, what would be the phenotype and genotype of the F₂?

23. If a male white coffee fruit fly were crossed with a female with scalloped wings, what would be the phenotype and genotype of the F₁ and F₂?

V. Trihybrid Crosses

24. A woman has migraine headaches (homozygous condition), is left-handed, and carries the trait for Tay-Sachs disease. She marries a man who has no headaches, has no trait for Tay-Sachs disease, and is right-handed (however, his mother is left-handed). What are the possible phenotypes of their children?

Appendix

Table of genotypes for monohybrid problems.

Trait Phenotype	Dominant Condition	Recessive Condition	Possible Dominant	Possible Recessive
Flower Color (Sweet Peas)	Red	White	R?	rr
Eye color (Human)	Brown	Blue	B?	bb
Ability to taste (PTC)	Taster	Non-taster	T?	tt
Sheep coat color	White	Black	W?	ww
Ptosis (inability to open eyelid completely)	Condition present	Condition absent	P?	pp
Hair color (human)	Non-red	Red	H?	hh
Dimple	Present	Absent	D?	dd
Coat color (mouse)	Yellow	Black	Zz	zz
Leaf color (Snapdragon-mature)	Yellow	Green	Gg	gg
Curly Hair (human)	Curly Wavy Straight		CC CS SS	
Coat color Guinea pig	Yellow Cream White		YY WY WW	

Table of genotypes for multiple allele, sex-linked, dihybrid, and trihybrid genetics problems.

Trait Phenotype	Dominant Condition	Recessive Condition	Possible Dominant	Genotype Recessive
Color blindness	Normal	Inability to distinguish between red and green	XY XX $X^C X$	$X^C Y$ $X^C X^C$
Hemophilia	Normal blood	failure of blood to clot	XY XX $X^h X$	$X^h Y$ $X^h X^h$
Eye color	Red eyes	White eyes	XY XX $X^w X$	$X^w Y$ $X^w X^w$
Blood groups	Type A Type B Type AB	Type O	AA AO BB BO AB	O
Rh factor in blood	Present	Absent	R?	rr
Flower height	Tall	Short	T?	tt
Eyelashes	Long	Short	E?	ee
Nose	Roman	Straight	N?	nn
Freckles	Present	Absent	F?	ff
Double-jointed thumb	Present	Absent	J?	jj
Eye color (fruit fly)	Red	White coffee	XY $X^f X$	$X^f Y$ $X^f X^f$
Wing appearance (fruit fly)	Normal	Scalloped margins	XY $X^S X$	$X^S Y$ $X^S X^S$
Wing appearance (fruit fly)	Normal	Vestigial	V?	vv
Wing appearance (fruit fly)	Normal	Dumpy	D?	dd
Migraine Headaches	Present	Absent	M?	mm
Right handed	Right handed	Left handed	L?	ll
Tay-Sachs disease	Absent	Present	T?	tt
Bristles (fruit fly)	Normal	Shaven	H?	hh
Eye color (fruit fly)	Red	Brown	B?	bb