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MENDELIAN INHERITANCE IN HUMANS

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Most introductory biology texts include basic Mendelian genetics. Applying the principles of trait dominance and independent assortment of alleles to family and classmates rather than to peas often proves bewildering for both student and teacher. Although traits in the human population that exhibit Mendelian inheritance are relatively few in number, some are readily amenable to classroom study.

To be useful for classroom studies of human genetics, the model traits should meet most or all of the following prerequisites. The trait should be governed by a single gene. Its method of inheritance should be well documented. Procedures to determine possession of the trait should not be complex, and the trait should appear frequently enough within the classroom population to provide adequate examples. Finally, the trait should be one that will not cause embarrassment to those expressing it, that is, it should not involve a disease, deformity, or defect.

Some inherited characteristics suitable for the human genetics classroom that have well-documented modes of inheritance will be discussed here.

MIDDIGITAL HAIR

Genetic control of midphalangeal hair (Fig. 1) was first documented by C. H. Danforth in 1921. The presence of middigital hair is a dominant condition; therefore those lacking it are homozygous recessive for the trait. Students may require a hand lens to determine their phenotypes since the presence of even the slightest amount of hair qualifies as the dominant condition.

BENT LITTLE FINGER

Type A 3 brachydactylia, which was cataloged in 1965 by P. Dutta, is characterized by a shortened middle phalanx of the fifth finger. Because of the triangular shape of the imperfect middle segment, radial curvature of the fifth digit results. This trait is inherited as a dominant. Presence or absence of the trait can be determined by placing the slightly spread fingers on a flat surface and noting the alignment of the fifth finger in relation to the other four (Fig. 2). Those with the condition will notice a slight angling of the fifth finger toward the other digits.

TONGUE ROLLING

The inheritance of several tongue movement traits has been documented, researched, and disputed for the last half century. The ability to roll the tongue upward from the sides (Fig. 3) has received most emphasis in the literature. In 1940, A. H. Sturtevant reported two classes within the human population, roller and nonroller. The roller phenotype is dominant; thus individuals unable to perform this maneuver are homozygous recessive for the trait.

WIDOW'S PEAK

McKusick (1983) and most other researchers infer that the inheritance of widow's peak, a distinct downward point of the frontal hairline, occurs as a dominant in the human gene pool. Homozygous recessive individuals possess a straight hairline. This trait is expressed in varying degrees and occurs with enough frequency to be used in the classroom.

EARLOBE ATTACHMENT

The inheritance of a free or unattached earlobe (Fig. 4, left) is caused by a dominant gene. The homozygous recessive condition is expressed as direct attachment of the earlobe to the head (Fig. 4, right). Some researchers have suggested that size, shape, and attachment of the ears and lobes are all polygenetically controlled. However, most of the literature supports Mendelian inheritance concerning lobe attachment. Other genes, working singly or polygenetically, affect the size and shape of the ear and lobe.

FACIAL DIMPLES

The occurrence of natural indentations at the corners of the mouth (dimples) is controlled by a dominant allele. Those persons with

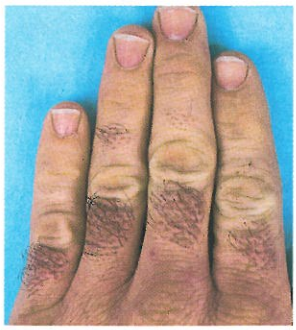

FIGURE 1 Middigital hair (dominant trait). All photographs are from our Biophoto® sheets Human Phenotypes 1 (17-4831) and Human Phenotypes 2 (17-4832).

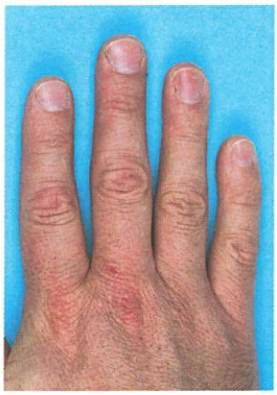

FIGURE 2 Bent little finger (dominant trait).

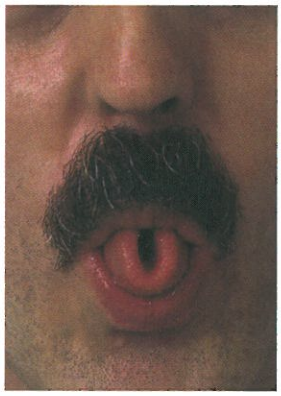

FIGURE 3 Tongue roller (dominant trait).

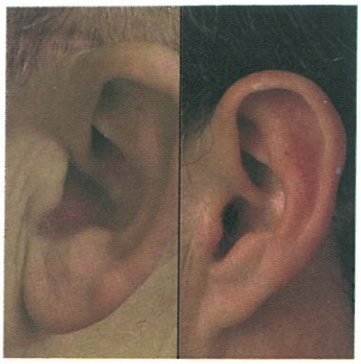

FIGURE 4 *Left,* free earlobe (dominant trait); *right,* attached earlobe (recessive trait).

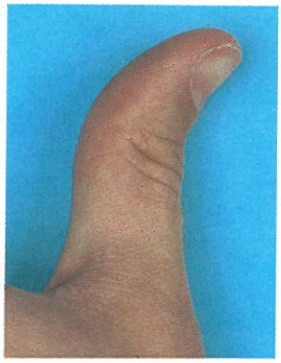

FIGURE 5 Hitchhiker's thumb (recessive trait).

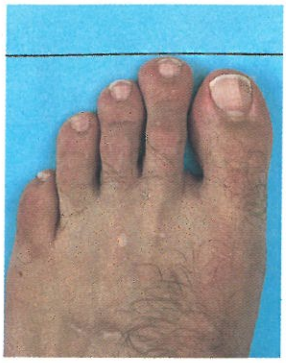

FIGURE 6 Short big toe (dominant trait).

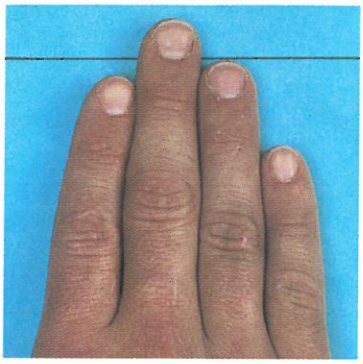

FIGURE 7 Short index finger (dominant trait in males, recessive in females).

the phenotypic absence of facial dimples generally possess the homozygous recessive genotype. Facial dimples may be inherited as an irregular dominant, but the mechanisms of transmission are solid enough for educational applications.

PTC Tasting

One of the most thoroughly documented human genetic traits is the ability or inability to taste the chemical phenylthiocarbamide (PTC, $C_7H_8N_2S$), also known as phenylthiourea. A strong, absorbent tissue or paper is saturated with a solution of PTC (0.5 g per liter), air-dried, and cut into small strips approximately 1 x 4 cm. The strip of dry, saturated paper is then placed on the tongue, allowing time for adequate mixture of the saliva and PTC crystals. (Note: a control taste test should be made prior to the PTC taste test to eliminate any confusion between tastes of treated and untreated papers.)

About 70 percent of the white American population can discern a definite bitter taste. Almost 91 percent of the black American population falls into the PTC taster phenotype. The PTC taster gene has been designated as T; therefore tasters are either homozygous dominant (TT) or heterozygous dominant (Tt). Nontasters carry the tt genotype.

Hitchhiker's Thumb

In 1953 B. Glass and J. C. Kistler classified distal hyperextensibility of the thumb (Fig. 5) as recessive. Considerable variation exists in the expression of this gene. For classroom purposes, those individuals who cannot bend at least one thumb backward about 45 degrees are probably carrying the dominant gene.

Length of Big Toe

In 1964 A. R. Kaplan claimed that the relative lengths of the hallux (big toe) and second toe are Mendelian traits. Those individuals possessing a long hallux are considered genotypically recessive for the trait. Possession of the dominant gene results in a short big toe in relation to the second toe (Fig. 6). Rarely, the phenotype may differ on each foot. If one or both halluces are either equal to or greater in length than the second toe, the recessive gene is present.

Discussion and observation of this trait should be left to the instructor's discretion, taking into consideration individual feelings about personal hygiene.

Sex-Influenced Traits

Sex-influenced alleles are those whose dominance is affected or altered by the sex of the bearer.

Relative Finger Lengths

The relative lengths of the index and ring fingers was thoroughly researched by H. W. Kloepfer in 1946 and by V. R. Phelps in 1952. The allele for a short index finger (Fig. 7) in relation to the fourth finger is dominant in males and recessive in females. In rare instances phenotypic expression may vary between left and right hands. If one or both index fingers are either equal to or greater in length than the fourth finger, the recessive genotype is present in males and the dominant in females.

Pattern Baldness

The expression of pattern baldness is characterized by gradually thinning hair on the top of the head, culminating in a fringe of hair low on the head. This condition will obviously have few classroom representatives; however, most, if not all, students should be familiar with the phenotype. Pattern baldness is an excellent example of a sex-influenced autosomal trait. The controlling gene acts as a dominant in males and a recessive in females. Phenotypic expression of the heterozygous condition usually occurs only in the presence of higher testosterone levels.

Sex-Affiliated Inheritance

The difference between the XX and XY genotypes in the human population creates some interesting and complex possibilities. Traits carried on the sex chromosomes fall into two categories: sex-linked (X-linked) and holandric.

Genes located exclusively on the X chromosome are termed sex-linked. Genes on the X chromosome are represented twice in females and once in males. Therefore phenotypic expression of a recessive gene would be expected to occur more often in males.

Some Mendelian-inherited X chromosome conditions include hemophilia A and B, red-green color blindness, diabetes insipidus, white forelock, juvenile muscular dystrophy, night blindness, parkinsonism, and retinitis pigmentosa. Each of the above is governed by a recessive gene located on the X chromosome.

Holandric traits are carried only on the Y chromosome and are therefore expressed only in males. Current research has uncovered few examples of holandric inheritance patterns. One example is hypertrichosis or hairy ears. This trait is complicated by the degree of variability of the phenotype, however. Expression may range from a few sparse hairs to thick, heavy growth.

Classroom examples of individuals possessing conspicuous traits carried by the sex chromosomes are few. Discussion of sex-affiliated inheritance in the genetics classroom should not be overlooked, however.

Further Reading

Burns, G. W., The Science of Genetics, 4th edition, Macmillan Publishing Co., Inc., New York, 1980.

McKusick, V. A., Mendelian Inheritance in Man, Johns Hopkins University Press, Baltimore, 1983.