Myths of Human Genetics

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INTRODUCTION

(This is the print version of the online Myths of Human Genetics, http://udel.edu/~mcdonald/mythintro.html. The online version is free, has color photos, and includes some links. This print version is available from www.lulu.com/product/18736867 for those who like paper.)

A fun way to teach the basics of genetics is to have students look at traits on themselves. Just about every biology student has, in one class or another, been asked to roll their tongue, look at their earlobes, or check their fingers for hair. Students can easily collect data on several different traits and learn about genes, dominant and recessive alleles, maybe even Hardy-Weinberg proportions. Best of all, these data don't require microscopes, petri dishes, or stinky fly food.

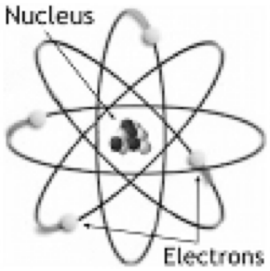
Unfortunately, what textbooks, lab manuals and web pages say about these human traits is mostly wrong. Most of the common, visible human traits that are used in classrooms do NOT have a simple one-gene, two-allele, dominant vs. recessive method of inheritance. Rolling your tongue is not dominant to non-rolling, unattached earlobes are not dominant to attached, straight thumbs are not dominant to hitchhiker's thumb, etc.

In some cases, the trait doesn't even fall into the two distinct categories described by the myth. For example, students are told that they either have a hitchhiker's thumb, which bends backwards at a sharp angle, or a straight thumb. In fact, the angle of the thumb ranges continuously, with most thumbs somewhere in the middle. This was clearly shown in the very first paper on the genetics of hitchhiker's thumb (Glass and Kistler 1953), yet 60 years later, teachers still ask students which of the two kinds of thumb they have.

In other cases, the trait really does fall into two categories, but it isn't determined by genetics. For example, students are asked to fold their arms, then told that the allele for having the right forearm on top is dominant. It is true that most people fall into two categories, right arm on top or left arm on top, but the very first study on the subject (Wiener 1932) clearly demonstrated that there is little or no genetic influence on this trait: pairs of right-arm parents are just about as likely to have right-arm children as are pairs of left-arm parents.

Some traits, such as tongue rolling, were originally described as fitting a simple genetic model, but later research revealed them to be more complicated. Other traits were shown from the very beginning to not fit the simple genetic model, but somehow textbook authors decided to ignore this. A quick search in the standard reference on human genetics, Online Mendelian Inheritance in Man (http://www.ncbi.nlm.nih.gov/omim), makes it clear that most of these traits do not fit the simple genetic model. I**t is an embarrassment to the field of biology education that textbooks and lab manuals continue to perpetuate these myths.**

Who's my daddy?



Not true.

Teachers might argue that using traits like tongue rolling and arm folding to teach genetics is a useful "lie-to-children," an oversimplification that is useful for teaching beginners, like teaching physics students that electrons are particles that rotate around the nucleus of an atom in little circular orbits. They can learn about polygenic traits, incomplete penetrance, and environmental effects in later biology classes, at the same time they're learning the quantum model of electrons in their physics classes.

However, there is another problem with teaching inaccurate human genetics. Let's say you tell your students that arm folding is a genetic trait, with the allele for right forearm on top (R) being dominant to left forearm on top (L). Results from a large number of studies show that about 11 percent of your students will be R children of two L parents; if they understand the genetics lesson correctly, they will think that either they were secretly adopted, or Mom was fooling around and Dad isn't their biological father. More of your students will reach this conclusion with each bogus genetic trait that you add to the lesson. I don't think this is a good way to teach genetics.

It is possible to use accurate genetic traits for a classroom exercise, such as blood types or DNA markers. However, some children are not the biological offspring of the people they think they are. The most common cause of this is a woman having an affair with someone other than her husband or partner. Several genetic studies, mostly in European and North American populations, have found that the rate of this "paternal discrepancy" is about 4 percent of all children (Bellis et al. 2005), which means that a typical classroom is likely to have at least one child who doesn't know that the person they call "Dad" isn't their biological father. Less commonly, children are not told they are adopted or are the product of artificial insemination or egg donation. So if you use blood groups or DNA for a classroom exercise in genetics, some of your students may find out that their family belongs on a lurid daytime talk show. While it can be medically important to know who your biological parents really are, a simple classroom demonstration of introductory genetics is not the way to learn this potentially traumatic information.

Alternatives

I prefer to use cat coat genetics to teach basic genetic concepts, because there are several easily visible traits whose genetics is well-established by cat breeders. I have my students use Petfinder.com to look at pictures of cats up for adoption in different cities; that way, they can look for geographic variation in allele frequency. It is easy to score several different traits from photographs, and if the students don't have access to computers, you can ask each student to describe the cat they know best. My experience has been that even students who don't own a cat have a friend's or neighbor's cat that they can describe with sufficient accuracy from memory.

Here are the easiest cat coat traits for students to identify. There are other traits that are rare (Siamese, polydactyly), difficult to score in photographs (agouti), or more complicated (genes affecting color patterns).

Locus	Genotypes	Phenotypes
L (hair length)	LL	Short hair
	Ll	Short hair
	ll	Long hair
W (white)	WW	completely white
	Ww	completely white
	ww	some colored hair
S (piebald spotting)	SS	some white hair
	Ss	some white hair
	ss	no white hair
D (dense pigment)	DD	black, brown, or orange
	Dd	black, brown, or orange
	dd	gray, light brown, or cream
O (orange)	OO (♀) or O- (♂)	orange or cream
	Oo (♀)	calico or tortoiseshell
	oo (♀) or o- (♂)	black or gray

At the *hair length* locus, there are some suggestions in the literature that the allele for long hair is more common in colder areas; this is something students can investigate using pictures of cats from different cities.

The *white* locus is useful because the dominant W allele, which produces all-white cats, is quite rare; this helps students understand that a "dominant" allele, in genetics, is one that determines the phenotype of the heterozygote, **not** the most common allele in the population. The *spotting*, *dense*, and *orange* loci cannot be scored in cats with the W allele.

At the *spotting* locus, the amount of white color in cats with the S allele can range from a few white toes, to white everywhere except for a colored patch on the forehead or tail. Some sources say that S/s heterozygotes have white on less than 50 percent of the body, while S/S homozygotes have white on more than 50 percent; it would be interesting for students to investigate this and see whether cats fall into three discrete categories (no white, much less than 50 percent white, much more than 50 percent white) or if there are a lot of cats with intermediate amounts of white. The pattern and amount of white may be affected by developmental accidents and modifier genes (see http://www.messybeast.com/bicolours.htm for much more information).

Gus is L? ww Ss D? O-

The *dense pigment* locus can be hard to score in badly lit photographs, but is pretty clear when you see the cat in person.

The *orange* locus is sex-linked (it is on the X chromosome). In the United States, cats with orange and black patches are called "calico" if they also have white from the *spotting* locus and "tortoiseshell" if they don't have white; in Australia, both kinds are called "tortoiseshell" or "torties." Male calico or tortoiseshell cats are extremely rare and are generally found in XXY males. Because all three genotypes can be distinguished in females, it is possible to test a sample of cats for fit to Hardy-Weinberg proportions.

Summary for worried parents

Can two parents who **fold their arms** with the left arm on top have a child who folds with the right arm on top? **Yes**.

Can two parents with **attached earlobes** have a child with unattached? **Yes**.

Can two parents who produce **red urine after they eat beets** have a child who makes yellow beet urine? **Yes**.

Can two parents with straight pinkies have a child with a **bent pinkie**? **Yes**.

Can two parents with smooth chins have a child with a **cleft chin**? **Yes**.

Can two parents without a bump inside their ear ("**Darwin's tubercle**") have a child with this bump? **Yes**.

Can two **blue-eyed** parents have a child with brown, green or hazel eyes? **Yes**.

Can two **red-haired** parents have a child with blond or brown hair? **Yes**.

Can two parents whose **hair whorls counterclockwise** on the back of their head have a child whose hair whorls clockwise? **Yes**.

Can two parents who **clasp their hands** with the left thumb on top have a child who clasps hands with the right thumb on top? **Yes**.

Can two parents with **hitchhiker's thumbs** have a child with straight thumbs? **Yes**.

Can two parents without **mid-digital hair** have a child with hair? **Yes**.

Can two parents who cannot taste the **bitter compound PTC** have a child who can taste it? **Yes**.

Can two parents with the **big toe longer** than the second toe have a child with the big toe shorter than the second toe? **Yes**.

Can two parents who cannot **roll their tongues** have a child who can? **Yes**.

Can two parents without **dimples** have a child with dimples? **Probably**, but no real research has been done.

Can two parents without a **widow's peak** have a child with a widow's peak? **Probably**, but no real research has been done.

Can two parents who produce non-stinky urine after they eat asparagus have a child who makes **stinky asparagus urine**? **Maybe not**; more research is needed.

Can two parents with **dry earwax** have a child with wet earwax? **No** (or at least it's very rare).

References

Glass, B., and J.C. Kistler. 1953. Distal hyperextensibility of the thumb. Acta Genetica 4: 192-206.

Wiener, A.S. 1932. Observations on the manner of clasping the hands and folding the arms. American Naturalist 66: 365-370.

ARM FOLDING

Right arm on top; left arm on top.

Most people have a strong preference when they fold their arms; they either have the left forearm on top (L) or the right forearm on top (R). Arm folding is sometimes used to illustrate basic genetics; the myth is that it is controlled by a single gene with two alleles, and the allele for R is dominant over the allele for L. I do not know how the myth got started. I don't know of any scientific papers that make this claim; the first paper to look at arm folding, Weiner (1932), clearly concludes that it has little or no genetic basis.

Arm folding as a character

Arm folding is easy to score; most people only fold their arms in one way, with the opposite way feeling very unnatural. In most populations, slightly more than half of people are L (McManus and Mascie-Taylor 1979). Some people, however, fold their arms either way; Weiner (1932) found that 3 out of 22 people folded their arms either way, while Reiss and Reiss (1998) reported that about 4 percent of their subjects had no preference.

Family studies

Wiener (1932) was the first to examine the genetic basis of arm folding by comparing parents and offspring, with the following results:

Parents	R offspring	L offspring	Percent R
R × R	31	50	38%
R × L	81	99	45%
L × L	58	70	45%

Each of the three kinds of matings has about the same proportion of R and L offspring, so Weiner (1932) concluded that there is no genetic basis for arm folding preference. If the myth were true, two L parents could not have an R child, but close to half of the children of LxL matings are R. For some reason, people kept doing family studies of arm folding, so that Reiss and Reiss (1998) were able to summarize the numbers from 12 studies:

Parents	R offspring	L offspring	Percent R
R × R	731	672	52%
R × L	1230	1497	45%
L × L	629	1038	38%

There is some association between parents and offspring, in that R x R parents have a higher proportion of R offspring than do L x L parents. All studies have found many R offspring of L x L parents and L offspring of R x R parents, so even if there is some genetic influence on arm folding, it is not a simple one-locus, two-allele genetic trait.

Twin studies

Reiss and Reiss (1998) summarized the results of two twin studies, which gave nearly identical results. About half of the pairs of monozygotic (identical) twins consist of one R twin and one L twin, and the same is true for dizygotic twins. This is further evidence for a lack of genetic influence on this trait.

	Monozygotic	Dizygotic
Both R	48	12
Both L	80	49
R + L	109	78

Conclusion

Family and twin studies clearly demonstrate that there is little genetic influence on arm folding, and it certainly is not the simple one-gene, two-allele trait described in the myth. You should not use arm folding to demonstrate basic genetics.

References

McManus, I.C., and C.G.N. Mascie-Taylor. 1979. Hand clasping and arm folding: A review and a genetic model. Annals of Human Biology 6: 527-558.

Reiss, M., and G. Reiss. 1998. Arm-folding in families: A review of literature and some new data. Psychologia 41: 81-87.

Wiener, A.S. 1932. Observations on the manner of clasping the hands and folding the arms. American Naturalist 66: 365-370.

ASPARAGUS URINE SMELL

After they eat asparagus, some people notice that their urine has a strong, unusual odor. Other people don't notice anything unusual. This was first thought to result from genetic variation in whether or not sulfur compounds in asparagus were secreted into the urine, with the allele for secreting being dominant. Later it was suggested that everyone secretes the compounds in their urine, but only some people can smell the compounds. Better-controlled experiments have shown that there is variation in both traits; some people secrete the compounds in their urine but can't smell them, while some people don't secrete the compounds but can smell them in other people's urine. This complication means that the ability to smell stinky compounds in one's own urine after eating asparagus is not a simple genetic trait. It is not known whether the two separate traits, secreting the compounds and being able to smell them, have a simple genetic basis.

Asparagus urine as a character

It has long been known that some people find that their urine has a strong, unusual smell after they eat asparagus, which is believed to result from sulfur-containing compounds (Nencki 1891). Other people don't notice anything unusual after eating asparagus. There are two possible explanations for this: some people excrete smelly compounds in their urine after eating asparagus, while other people don't excrete them; or some people can smell the compounds, while other people can't.

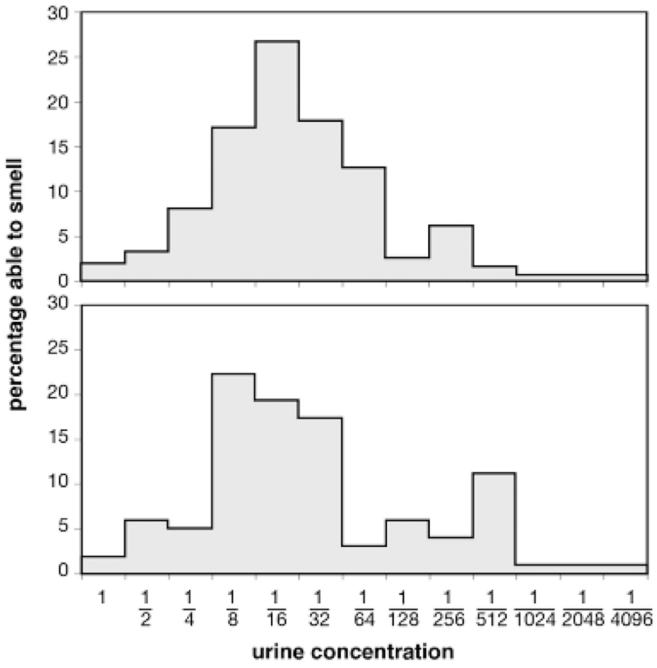
Unfortunately, many of the papers on asparagus urine are short and lack detail. Allison and McWhirter (1956) were the first to report that there was polymorphism, using a chemical test for methanethiol and finding that 46 out of 115 people were excretors. They did not say where there subjects where they were from (presumably Britain) or give any details about their chemical techniques. In response to criticism (Penrose 1957) of their study, Allison and McWhirter (1957) stated that methanethiol was clearly either present or absent; again, they gave no details about the chemical test they used.

Lison et al (1980) took asparagus urine from a single individual and asked 328 Israelis to smell different dilutions and find the lowest concentration that they could distinguish from tap water. They found a lot of variation among individuals, then divided them into "smellers" and "non-smellers." They also had 10 "smellers" smell the urine of 11 "non-smellers," and all of the smellers could distinguish asparagus urine from normal urine. Based on the second experiment, they concluded that everyone excretes the smelly substances, but there is variation in ability to smell them.

Hoffenberg (1983) took asparagus urine from a single individual and had 98 schoolchildren find the lowest concentration that they could distinguish from

tap water. The distribution was somewhat bimodal, but as in Lison et al. (1980), there was not a clear gap between smellers and non-smellers. Both Lison et al. (1980) and Hoffenberg (1983) asked people to distinguish between the odors of asparagus urine and tap water, not between asparagus urine and normal urine.

Mitchell et al. (1987) fed asparagus to 800 volunteers and then collected their urine. Three smellers then smelled their asparagus urine and a control sample of normal urine and classified the urine samples as smelly or not. All of the smellers agreed about the classification of all of the samples, which is a good indication that urine can be classified into two discrete types. Of the sample, 43% produced stinky urine.



Percentage of people able to tell the difference between the smell of diluted asparagus urine and the smell of tap water. Top, data from Lison et al. (1980); bottom, data from Hoffenberg (1983).

Waring et al. (1987) identified five people who produced stinky urine and three who did not, then used gas chromatography and found five sulfur compounds that were present in the stinky urine but not the non-stinky urine. This is further evidence that not everyone excretes the compounds.

Richer et al. (1989) fed asparagus to 103 French people and reported that all of them produced stinky urine. Unfortunately, they do not say how they assayed stinkiness.

Erickson et al. (2010) used a web-based survey form to ask customers of the 23andMe genomic company whether they had noticed an unusual smell after eating asparagus, and 63 percent of 4742 people said they had. Of course, it is unknown whether the people who hadn't noticed an odor were non-excretors or non-smellers.

Pelchat et al. (2011) asked people whether they could distinguish between asparagus and normal urine from other people in the study. They found that 3 out of 37 people produced asparagus urine that none of the other people could distinguish from normal urine, which means that some people really are non-excretors. Out of 31 people, 2 could not distinguish between the asparagus and normal urine of any of the subjects, meaning that there really are non-smellers. There was continuous variation in both excretion and smelling ability, as well; the asparagus urine of some people was easily detected by most of the people, while other people produced asparagus urine that only some people could detect.

Lison et al. (1980) and Richer et al. (1989) concluded that everyone excretes stinky compounds in their urine after eating asparagus, and the only variation is in ability to smell them. Other papers using chemical tests and carefully controlled smelling tests show that there is variation in excretion of sulfur compounds in urine after eating asparagus. The lack of detail in some of the papers makes it difficult to be sure, but it may be that everyone, if fed enough asparagus, excretes enough stinky compounds that a careful sniff by a sensitive nose can distinguish the asparagus urine from non-asparagus urine. It may be that only some people excrete large enough amounts of stinky compounds to be noticably unusual. Quantitative chemical analysis of sulfur compounds in the urine of a sample of asparagus eaters would be an obvious way to shed light on this.

It is clear that in addition to variation in excretion, there is also variation in the ability to smell the compounds. It is not clear whether this variation is continuous, or whether people can be clearly divided into smellers and non-smellers. Testing the ability of people to smell different concentrations of the sulfur compounds that are hypothesized to be the cause of the odor, such as methanethiol and dimethyl sulfide, would help resolve this question.

What is being smelled?

Despite a number of studies, it is not entirely clear what compounds are responsible for the odor in asparagus urine. Nencki (1891) identified methanethiol (a sulfur compound, also known as methyl mercaptan) in asparagus urine, and Allison and McWhirter (1956) found that is was present in the urine of some asparagus eaters and absent in others' urine. White (1975) used gas chromatography on methylene chloride extracts of asparagus urine and identified S-methyl thioacrylate and S-methyl 3-(methylthio)thiopropionate; he suggested that these compounds would be easily broken down into methanethiol, thus explaining the earlier results. He found that adding S-methyl thioacrylate and S-methyl 3-(methylthio)thiopropionate to normal urine made it smell like asparagus urine.

Waring et al. (1987) criticized previous studies for analyzing compounds in the liquid urine, not just those volatile enough to be smelled, and for using chemical extraction methods involving heat, time, and organic solvents that could cause chemical modifications in the urine. They used gas chromatography of the vapor above urine, and they identified five compounds that were present in the asparagus urine vapor of five excretors but absent in the vapor of three non-excretors: methanethiol, dimethyl sulphide, dimethyl disulphide, dimethyl sulphoxide, and dimethyl sulphone. By having smellers smell the different compounds, they found that a mixture of methanethiol and dimethyl sulfide was similar in smell to asparagus urine.

Leitner (2001) also used gas chromatography of vapors, and he identified twelve different sulfur compounds that were present in the vapors above asparagus urine but absent in normal urine. These included methanethiol and dimethyl sulphide, which Waring et al. (1987) identified as the sources of the distinctive odor.

Small, volatile molecules such as methanethiol and dimethyl sulfide would be lost in cooking, so there must be a more stable compound that is unique to asparagus and gets broken down in the body to produce the stinky compounds in asparagus urine. Asparagusic acid and its derivatives, such as dihydroasparagusic acid, are sulfur-containing compounds that are found in asparagus but not in related vegetables and which may act as plant growth inhibitors (Yanagawa et al. 1972) and to kill nematodes (Takasugi et al. 1975). Jansen (1948) fed 10 mg of dihydroasparagusic acid to two subjects and said their urine did not stink, but he did not say whether these two people were known to produce stinky urine after eating asparagus. Waring et al. (1987) fed asparagusic acid to two people known to produce stinky asparagus urine, and their urine stank; they also fed asparagusic acid to one person known to produce non-stinky asparagus urine, and that person's urine did not stink. While more detail about this experiment would be helpful, such as the amount of asparagusic acid used, it seems likely that the asparagusic acid in asparagus gets metabolized into smaller sulfur-containing compounds and excreted by some people. There are no data on whether non-excretors fail to absorb the asparagusic acid, fail to metabolize it, or fail to excrete the products of metabolism.

Family studies

Allison and McWhirter (1956) compared parents and offspring, using a chemical test to separate excretors (E) from non-excretors (NE):

Parents	E offspring	NE offspring
E × E	1	1
E × NE	5	7
NE × NE	0	11

They concluded that excretion of sulfurous compounds in urine after eating asparagus is controlled by a single gene, with the allele for excretion dominant over the allele for non-excretion.

Mitchell et al. (1987) performed a similar study, except they used three smellers to distinguish between the urine of excretors and non-excretors, and found similar results:

Parents	E offspring	NE offspring
E × E	19	3
E × NE	17	9
NE × NE	0	7

It would be better to have larger sample sizes and a more quantitative method of distinguishing excretors from non-excretors, but the available data are completely consistent with excretion being determined by a single gene with two alleles, with excretion dominant to non-excretion.

There are no family or twin studies on the ability to smell the stinky compounds in asparagus urine.

Molecular genetics

Erickson et al. (2010) surveyed customers of 23andMe, a company which genotyped the individuals at 535,076 single-nucleotide polymorphisms (SNPs). They asked 10,000 customers of northern European ancestry "Have you ever noticed a peculiar odor when you pee after eating asparagus?" and received 4737 responses. There was a statistically strong association between the answers and a region on chromosome 1 containing 10 olfactory receptor genes. At the SNP with the strongest association, a G/A polymorphism, 56.7% of GG, 70.9% of GA, and 74.0% of AA individuals reported stinky asparagus urine. This suggests that there is genetic variation in either excretion or ability to smell, but it is not clear which.

Pelchat et al. (2011) separated two traits, excreting stinky compounds and being able to smell them, and determined genotypes for the SNP for which Erickson et al. (2010) found the strongest association. They found an association of SNP genotype with the ability to smell asparagus urine, but no significant association with excreting.

Conclusion

It is clear that there is variation in two different traits: excretion of sulfur compounds in urine after eating asparagus, and ability to smell those compounds. This means that asking people whether their own urine smells odd after they eat asparagus is not a good way to study this. The limited amount of family data available suggests that excreting may be a simple one-gene character, with the allele for excreting dominant, but more work needs to be done. There is no family data on the smelling/non-smelling trait, only the genomic association study of Pelchat et al. (2011), so more work needs to be done on this trait as well.

References

Carrière, R. 1922. Über erbliche Orhformen, insbesondere das angewachsene Ohrläppchen. Zeitschrift für Induktive Abstammungs- und Vererbungslehre 28: 288-242.

Dutta, P., and P. Ganguly. 1965. Further observations on ear lobe attachment. Acta Genetica 15: 77-86.

El Kollali, R. 2009. Earlobe morphology: a simple classification of normal earlobes. Journal of Plastic, Reconstructive and Aesthetic Surgery 62: 277-280.

Hilden, K. 1922. Über die Form des Ohrläppchens beim Menschen und ihre Abhängigkeit von Erblanglagen. Hereditas 3: 351-357.

Lai, L.Y.C., and R.J. Walsh. 1966. Observations on ear lobe types. Acta Genetica 16: 250-257.

Mohanraju, C., and D.P. Mukherjee. 1973. Ear lobe attachment in an Andhra village and other parts of India. Human Heredity 23: 288-297.

Mowlavi, A., D.G. Meldrum, and B.J. Wilhelmi. 2004. Earlobe morphology delineated by two components: the attached cephalic segment and the free caudal segment. Plastic and Reconstructive Surgery 113: 1075-1076.[not seen yet]

Powell, E.F., and D.D. Whitney. 1937. Ear lobe inheritance: an unusual three-generation photographic pedigree chart. Journal of Heredity 28: 184-186.

Quelprud, T. 1934. Familienforschungen über Merkmale des äusseren Ohres. Zeitschrift fü Induktive Abstammungs- und Vererbungslehre 67: 296-299.

Wiener, A.S. 1937. Complications in ear genetics. Journal of Heredity 28: 425-426.

ATTACHED EARLOBE

Some people have earlobes that curve up between the lowest point of the earlobe and the point where the ear joins the head; these are known as "free" or "unattached" earlobes, as shown in the upper left of the picture below. Other people have earlobes that blend in with the side of the head, known as "attached" or "adherent" earlobes, as shown in the lower right.

Attached vs. free earlobes are often used to illustrate basic genetics. The myth is that earlobes can be divided into into two clear categories, free and attached, and that a single gene controls the trait, with the allele for free earlobes being dominant. Neither part of the myth is true.

Earlobes ranging from unattached (upper left) to attached (lower right).

Earlobes as a character

Classroom exercises on earlobe genetics say that there are two distinct categories, free (F) and attached (A). However, many of the papers on earlobe genetics have pointed out that there are many people with intermediate earlobes (Quelprud 1934, Wiener 1937, Dutta and Ganguly 1965). El Kollali (2009) classified earlobes into three types, based on whether the attachment angle was acute, right, or obtuse. To make the picture above, I searched for pictures of professional bicyclists (because they have short hair), found 12 with their ears showing, and arranged them from free to attached. It doesn't look to me as if there are just two categories; instead, there is continuous variation in the height of the attachment point (the "otobasion inferius") relative to the lowest point on the earlobe (the "subaurale"). My own earlobes are exactly halfway in between

the two extremes; I couldn't tell you whether my earlobes should be considered free or attached.

Family studies

Carrière (1922) and Hilden (1922) were among the first to study the genetics of earlobes, and they reached opposite conclusions. Carrière (1922) looked at 15 families and concluded that attached earlobes were dominant. However, all of the offspring of A x A matings had attached earlobes, and there were no F x F matings, so his data are consistent with either free or attached being dominant.

Powell and Whitney (1937) looked at one family and concluded that attached earlobes were recessive. Wiener (1937) responded by pointing out that the "arbitrary classification into two sharply defined types...gives a false picture, since all gradations between the two extremes are encountered." He divided earlobes into four arbitrary groups, from 0 (completely free) to 3 (completely attached). All possible matings, from completely 0 x 0 to 3 x 3, produced some intermediate earlobes. Wiener (1937) concluded that earlobes were determined by more than one gene, or by a singe gene with more than two alleles.

Lai and Walsh (1966) called earlobes in which the lowest point on the earlobe was the attachment point "attached," and they classified all other earlobes as "free." They recorded the following data on families in New Guinea:

Parents	F offspring	A offspring	Percent F
F × F	12	22	35%
F × A	72	114	39%
A × A	37	90	29%

If the myth were true, two parents with attached earlobes could not have a child with a free earlobe. There are slightly more A offspring from A x A matings, but the large numbers of F offspring from A x A matings and A offspring from F x F matings indicate that this is not a one-locus, two-allele trait.

Mohanraju and Mukherjee (1973) performed a similar study in India and found similar results:

Parents	F offspring	A offspring	Percent F
F × F	13	1	93%
F × A	7	7	50%
A × A	5	29	15%

They found a much stronger association between parents and offspring, but the five F offspring of A x A matings are inconsistent with the myth that this is a one-locus, two-allele trait.

Conclusion

Earlobes do not fall into two categories, "free" and "attached"; there is continuous variation in attachment point, from up near the ear cartilage to well below the ear. While there is probably some genetic influence on earlobe

attachment point, family studies show that it does not fit the simple one-locus, two-allele myth. You should not use earlobe attachment to demonstrate basic genetics.

References

Carrière, R. 1922. Über erbliche Orhformen, insbesondere das angewachsene Ohrläppchen. Zeitschrift für Induktive Abstammungs- und Vererbungslehre 28: 288-242.

Dutta, P., and P. Ganguly. 1965. Further observations on ear lobe attachment. Acta Genetica 15: 77-86.

El Kollali, R. 2009. Earlobe morphology: a simple classification of normal earlobes. Journal of Plastic, Reconstructive and Aesthetic Surgery 62: 277-280.

Hilden, K. 1922. Über die Form des Ohrläppchens beim Menschen und ihre Abhängigkeit von Erblanglagen. Hereditas 3: 351-357.

Lai, L.Y.C., and R.J. Walsh. 1966. Observations on ear lobe types. Acta Genetica 16: 250-257.

Mohanraju, C., and D.P. Mukherjee. 1973. Ear lobe attachment in an Andhra village and other parts of India. Human Heredity 23: 288-297.

Mowlavi, A., D.G. Meldrum, and B.J. Wilhelmi. 2004. Earlobe morphology delineated by two components: the attached cephalic segment and the free caudal segment. Plastic and Reconstructive Surgery 113: 1075-1076.[not seen yet]

Powell, E.F., and D.D. Whitney. 1937. Ear lobe inheritance: an unusual three-generation photographic pedigree chart. Journal of Heredity 28: 184-186.

Quelprud, T. 1934. Familienforschungen über Merkmale des äusseren Ohres. Zeitschrift fü Induktive Abstammungs- und Vererbungslehre 67: 296-299.

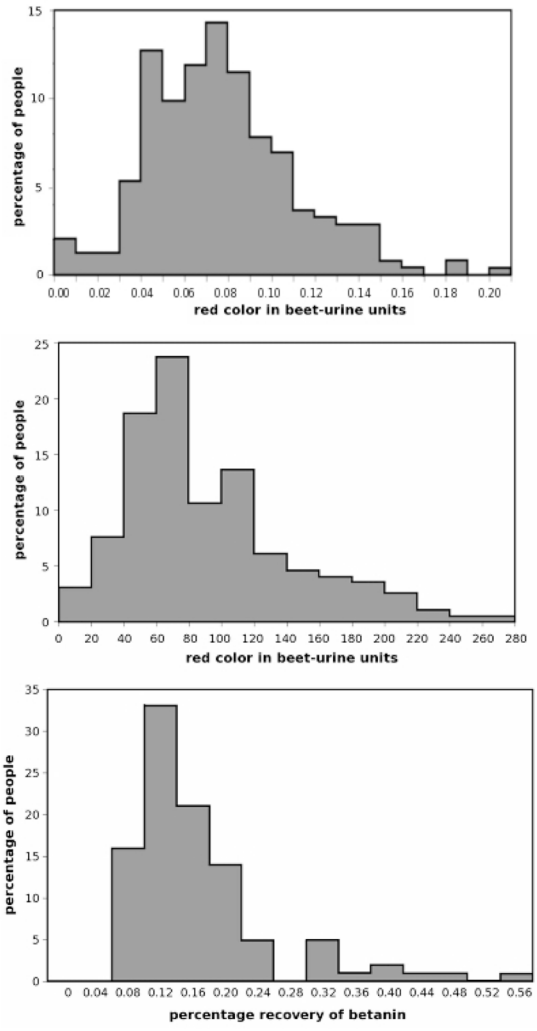
Wiener, A.S. 1937. Complications in ear genetics. Journal of Heredity 28: 425-426.

BEET URINE COLOR (BEETURIA)

After some people eat beets, their urine turns red, a harmless condition called beeturia or betaninuria. Because this looks like blood in the urine, someone who doesn't know it is caused by beets can become alarmed and go see a doctor. Other people have normal-looking yellow urine after they eat beets. The myth is that beeturia is caused by a single gene with two alleles, with the allele for beeturia being recessive.

Beeturia as a character

In early studies of beeturia (Allison and McWhirter 1956, Saldanha et al. 1960, Saldanha et al. 1962, Watson et al. 1963), people ate beets and then were classified as beeturic or non-beeturic based on the appearance of the urine. In general, people were classified as beeturic if there was any detectable redness in their urine. Forrai et al. (1968, 1982) measured the red color in urine with a photometer set to 530 nm, with the absorbance at the yellow wavelength of 660 nm subtracted to give "beet urine units." They found a broad distribution, but no separation into excretors and non-excretors, in a sample of 244 children (Forrai et al. 1968) and 198 twins (Forrai et al. 1982). Pearcy et al. (1991) conducted a similar study and came to the same conclusion, but they do not give their data. Watts et al. (1993) also found a distribution that was skewed but not bimodal.



Percentage of people with different amounts of betanine in their urine after eating beets. Top, data from Forrai et al. (1968); middle, data from Forrai et al. (1982); bottom, data from Watts et al. (1993).

Watson et al. (1963) and Tunnessen et al. (1969) found that beeturia was more common in people with iron deficiency, but Forrai et al. (1971) did not find a relationship between betanin and blood iron levels. Eastwood and Nyhlin (1995) gave non-beeturic subjects a mixture of betalaine and oxalic acid, and they became beeturic. Their interpretation was that the oxalic acid prevented betalaine from being decolorized in the stomach and colon, so that the variation among individuals in the redness of beet urine resulted from varying amounts of oxalic acid in the digestive system. They also found that vinegar-pickled beets caused more people to be beeturic than boiled beets, consistent with the role of acid in causing beeturia.

Family studies

Allison and McWhirter (1956) visually divided people into beeturic (B) and non-beeturic (NB) and looked at a number of families, with the following results:

Parents	B offspring	NB offspring
B × B	14	2
B × NB	2	2
NB × NB	0	6

Because all six offspring of B x B matings were beeturic, they concluded that beeturia was caused by a recessive allele.

Saldanha et al. (1962) looked at a larger number of families:

Parents	B offspring	NB offspring	Percent B
B × B	18	4	82%
B × NB	15	19	44%
NB × NB	17	38	31%

The 17 non-beeturic offspring of B x B matings do not fit the idea that beeturia is caused by a recessive allele. Saldanha et al. (1962) considered people with "very weak" amounts of red pigment in their urine as beeturic, while Allison and McWhirter (1956) only counted people who were "distinctly positive" for beeturia.

Twin studies

Forrai et al. (1982) fed pairs of twins uniform amounts of beet juice and measured the red pigment in their urine, rather than just classifying them as beeturic or non-beeturic. They found that monozygotic twins were not more similar to each other than dizygotic twins. If the amount of red pigment was determined by genetic variation, monozygotic twins should be more similar to each other, so this suggests that beeturia is not strongly affected by genetics.

Conclusion

The careful measurements of Forrai et al. (1982) and Watts et al. (1993) show that people cannot be divided into two distinct categories, beeturic and non-beeturic; instead, there is a continuous range of variation in the redness of urine after eating beets. The twin study of Forrai et al. (1982) suggests that this variation is not strongly determined by genetics. Beeturia is not a simple one-locus, two-allele trait.

References

Allison, A. C., and K. G. McWhirter. 1956. Two unifactorial characters for which man is polymorphic. Nature 178: 748-749.

Eastwood, M. A., and H. Nyhlin. 1995. Beeturia and colonic oxalic acid. Quarterly Journal of Medicine 88: 711-717.

Forrai, G., D. Vágújfalvi, and P. Bölcskey. 1968. Betaninuria in childhood. Acta Paediatrica Academiae Scientiarum Hungaricae 9: 43-51.

Forrai, G., D. Vágújfalvi, J. Lutter, E. Benedek, and E. Soós. 1971. No simple association between betanin excretion and iron deficiency. Folia Haematologica 95: 245-248.

Forrai, G., G. Bankovi, and D. Vágújfalvi. 1982. Betaninuria: a genetic trait? Acta Physiologica Academiae Scientiarum Hungaricae 59: 265-282.

Geldmacher-von Mallinckrodt, M., M. T. Aiello, and M. V. Aiello. 1967. Quantitative erfassung und klinische bedeutung der betaninurie. Zeitschrift für Klinische Chemie und Klinische Biochemie 5: 264-270.

Pearcy, R. M., S. C. Mitchell, and R. L. Smith. 1991. Beetroot and red urine. Biochemical Society Transactions 20: 225.

Saldanha, P. H., L. E. Magalhães, and W. A. Horta. 1960. Race differences in the ability to excrete beetroot pigment (betanin). Nature 187: 806.

Saldanha, P. H., O. Frota-Pessoa, and L. I. S. Peixoto. 1962. On the genetics of betanin excretion. Journal of Heredity 53: 296-298.

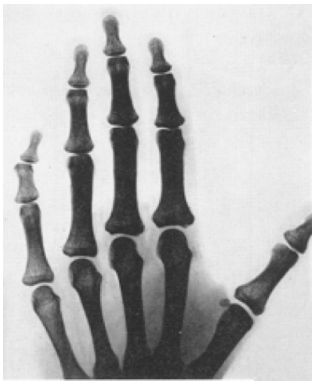
Tunnessen, W. W., C. Smith, and F. A. Oski. 1969. Beeturia. American Journal of Diseases of Children 117: 424-426.

Watson, W. C., R. G. Luke, and J. A. Inall. 1963. Beeturia: its incidence and a clue to its mechanism. British Medical Journal 2: 971-973.

Watts, A. R., M. S. Lennard, S. L. Mason, G. T. Tucker, and H. F. 1993. Beeturia and the biological fate of beetroot pigments. Pharmacogenetics 3: 302-311.

BENT LITTLE FINGER

Some people's little fingers bend in towards the ring fingers (B), while in other people they are straight (S). The myth is that little fingers can be clearly divided into two categories, bent and straight, and that the trait is controlled by one gene with two alleles, with the allele for B being dominant. Neither part of the myth is true.



X-ray of hand with bent little finger (from Hersh et al. 1953).

Bent little finger as a character

The technical name for a little finger that bends in towards the ring finger is clinodactyly. When the little finger bends in towards the palm and can't be straightened out, it is known as streblomicrodactyly, streblodactyly or camptodactyly.

Little fingers range from perfectly straight to bending inwards at a sharp angle. It is not clear whether fingers fall into two discrete categories or there is a continuous range of pinky angle. Hersh et al. (1953) said that bent little fingers bend inward at an angle of 15 to 30 degrees. They found that only 4 out of a sample of 4,304 people had what they considered to be bent little fingers. Marden et al. (1964) identified about 1% of healthy newborns as having bent little fingers.

Family studies

Hersh et al. (1953) identified 51 families in which one or more children had bent little fingers. In 47 of the families, one parent had bent little fingers and the other had straight. Hersh et al. (1953) concluded that bent little finger was caused by a single dominant allele, but the four families in which both parents of a B child were S are inconsistent with this.

Dutta (1965) found two extended families with bent little fingers. Six children of S x S parents were all S, while 22 out of 34 children of B x S parents were B. This fits the model of bent little finger being caused by a single dominant allele, but the number of families is very small. Leung and Kao (2003) found one more extended family in which bent little fingers were common; they concluded that it fit the model of B caused by a dominant allele, but the data also fit a model in which it is recessive.

Conclusion

If only extremely bent little fingers are considered, as done by Hersh et al. (1953), then the bent little finger trait is too rare to be useful for illustrating basic genetics in classrooms. If fingers with a more moderate bend are counted, then there is no clear dividing line between bent and straight, plus no evidence that the trait is genetic. In either case, you should not use bent little finger to demonstrate basic genetics.

References

Dutta, P. 1965. Inheritance of radially curved little finger. Acta Genetica et Statistica Medica 15: 70-76.

Hersh, A.H., F. DeMarinis, and R.M. Stecher. 1953. On the inheritance and development of clinodactyly. American Journal of Human Genetics 5: 257-268.

Leung, A.K.C, and Kao, C.P. 2003. Familial clinodactyly of the fifth finger. Journal of the National Medical Association 95: 1198-1200.

Marden, P.M., Smith, D.W., and McDonald, M.J. 1964. Congenital anomalies in the newborn infant, including minor variations: a study of 4,412 babies by surface examination for anomalies and buccal smear for sex chromatin. Journal of Pediatrics 64: 357-371.

CHEEK DIMPLES

When some people smile, they have dimples in one or both cheeks. Other people don't have dimples. This is occasionally said to be a simple genetic trait; the myth is that dimples are controlled by one gene with two alleles, and the allele for dimples is dominant. I do not know how the myth got started; I have searched the literature thoroughly, and I have not found any scientific papers with any genetic evidence on cheek dimples.

Cheek dimples.

Cheek dimples as a character

Dimples vary in how obvious they are. Wiedemann (1990) suggests that their appearance may be affected by circulation, body weight or muscle tone, although he does not cite any evidence for this. He describes several individuals who had dimples as children and lost them by adulthood. This would make it impossible to divide people into two distinct types, having dimples vs. not having dimples.

Family studies

Winchester (1951) claimed that dimples are dominant to non-dimples, but with the complicating factor of variable expression. He did not cite any studies as evidence for this assertion, and I have not been able to find any.

Conclusion

The presence of dimples may change during an individual's lifetime, and there is no published evidence for a genetic basis for dimples. Therefore you should not use dimples to demonstrate basic genetics.

References

Wiedemann, H-R. 1990. Cheek dimples. American Journal of Medical Genetics 36: 376.

Winchester, A. M. 1951. Genetics: A survey of the principles of heredity. Houghton Mifflin Company, Cambridge, MA.

Cleft Chin

Some people have a prominent dimple or crease in the front of the chin, called a cleft chin (or "butt chin"); others do not. This is sometimes said to be controlled by a single gene with two alleles, with cleft chin (C) dominant to smooth chin (S). The small amount of genetic data that is available shows that this myth is not true; in addition, there are many chins that are intermediate between clearly cleft and smooth.

Cleft chin as a character

Cleft chins come in a variety of shapes, including vertical furrows, Y-shaped furrows, and round dimples (Günther 1939). They also vary in depth from barely noticable to extremely prominent (Bhanu and Mahhotra 1972). I looked through a gallery of United States Senators (because they have large official portraits online) and picked out the ones that had some hint of a cleft chin, along with a few smooth-chinned senators for comparison. As you can see, there are a lot of people with vague depressions in the middle of the chin, and few with really unambiguous furrows or dimples. I find it hard to draw a clear boundary between cleft and smooth chins in these pictures.

Which of these would you call a cleft chin?

The frequency of cleft chin varies widely among different populations; Indian populations range from 4 to 71 percent cleft chin (Bhanu and Malhotra (1972). Günther (1939) recorded cleft chins in 9.6 percent of German men and 4.5 percent of German women. The large difference in frequency between men and women does not fit the simple genetic model of cleft chin being determined by a single autosomal (non-sex-linked) gene. Bhanu and Malhotra (1972) recorded the frequency of cleft chin in Indian boys and men of different ages. They said there was no difference in frequency of cleft chin among age groups, using a chi-square test, but when I analyze the data using logistic regression, I get a significant (P=0.03) increase in cleft chin with age; about 5% of boys 6 to 10 years old have cleft chins, while 10% of men over 35 have a cleft chin. This change with age is also evidence against the simple genetic model.

Family studies

Lebow and Sawin (1941) first suggested that cleft chin was a genetic character, based on data from a single family. They suggested that cleft chin was recessive, although they admitted that they didn't have definitive evidence.

There is an unpublished dissertation by Pfannenstiel (1951), which I haven't seen, that concludes that cleft chin is dominant, but affected by the environment (cited in Beckman et al. [1960]). Beckman et al. (1960) report the following data from Swedish families (C is cleft, S is smooth):

Parents	C offspring	S offspring	Percent C
C × C	10	1	91%
C × S	24	28	46%
S × S	4	33	11%

If the myth were true, two parents with smooth chins could not have a child with a cleft chin. While there is definitely a strong genetic influence, as parents with cleft chins have a higher proportion of cleft-chin offspring than do parents without cleft chins, the 4 cleft-chinned offspring from smooth X smooth parents do not fit the myth that cleft chin is determined by a dominant allele.

Conclusion

Many people have chins that are intermediate between clearly cleft and clearly smooth, and the family studies do not fit the myth that cleft chin is caused by a dominant allele. You should not use cleft chin to demonstrate basic genetics.

References

Beckman, L., J.A. Böök, and E. Lander. 1960. An evaluation of some anthropological traits used in paternity tests. Hereditas 46: 543-569.

Bhanu, V., and K.C. Malhotra. 1972. A population genetic study of cleft chin in India. American Journal of Physical Anthropology 37: 367-372.

Günther, H. 1939. Anomalien und Anomaliekomplexe in der Gegend des ersten Schlundbogens. Zeitschrift für menschliche Vererbungs- und Konstitutionslehre 23: 43-52.

Lebow, M.R., and P.B. Sawin. 1941. Inheritance of human facial features: a pedigree study involving length of face, prominent ears and chin cleft. Journal of Heredity 32: 127-132.

DARWIN'S TUBERCLE

Some people have a small bump on the inside of their upper ear. This is known as "Darwin's tubercle," or "Darwin's bump," because Charles Darwin mentioned it in his book The Descent of Man (Darwin 1879). Darwin's tubercle is sometimes used to illustrate basic genetics; the myth is that it is controlled by a single gene with two alleles, and the allele for Darwin's tubercle is dominant. The data in the only two genetic studies on this trait show that the myth is not true, so I'm not sure how the myth got started.

Ears with and without Darwin's tubercle.

Darwin's tubercle as a character

About 10 percent of Spanish adults (Ruiz 1986), 40 percent of Indian adults (Singh and Purkait 2009), and 58 percent of Swedish schoolchildren (Hildén 1929) have this trait. Singh and Purkait (2009) divide the tubercle into "nodosity", "enlargement" and "projection." The differences among these are not obvious. When I look at ears, some people have a very obvious bump, while others have just a slight swelling on the margin of the ear; I think it would be difficult to cleanly divide ears into two categories, with or without Darwin's tubercle. In addition, some people have a bump on one ear but not the other (Quelprud 1936), and it is not clear how to classify them.

Family studies

Quelprud (1936) looked at presence (P) and absence (A) of Darwin's tubercle in a large number of German families. Some individuals had a bump on just one ear; including them in P, the results were:

Parents	P offspring	A offspring	Percent P
P × P	88	59	60%
P × A	101	60	63%
A × A	22	27	45%

There are somewhat more A offspring from A x A matings than from P x P or P x A matings, suggesting that there is some genetic influence on the trait. However, if the myth were true, two parents without Darwin's tubercle could not have a child with the tubercle. The large number of P offspring from A x A matings, and

the large number of A offspring from P x P matings, are completely inconsistent with the simple genetic model of one locus with two alleles.

The only other family study that I'm aware of is Beckman et al. (1960), who found the following:

Parents	P offspring	A offspring	Percent P
P × P	1	3	25%
P × A	10	28	26%
A × A	14	44	24%

These data are also inconsistent with the simple two-allele genetic model. They even suggest that there is no genetic influence on the trait at all.

Twin studies

Quelprud (1936) looked at Darwin's tubercle in identical twins. He found 58 pairs of twins where both were P and 32 pairs where both were A. There were 26 pairs of twins in which one twin had a Darwin's tubercle on one or both ears, while the other twin did not. This suggests that whether a person has Darwin's tubercle depends in part on developmental accidents or other environmental influences, not just genetics.

Conclusion

The family and twin studies strongly indicate that Darwin's tubercle is not determined by a single gene with two alleles, and there may be very little genetic influence on the trait at all. You should not use Darwin's tubercle to demonstrate basic genetics.

References

Darwin, C. 1871. The descent of man and selection in relation to sex. London: John Murray.

Hildén, K. 1929. Studien über das Vorkommen der darwinschen Ohrspitze in der Bevölkerung Finnlands. Fennia 52: 3-39.

Quelprud, T. 1936. Zur erblichkeit des darwinschen höckerchens. Zeitschrift für Morphologie und Anthropologie 34: 343-363.

Ruiz, A. 1986. An anthropometric study of the ear in an adult population. International Journal of Anthropology 1: 135-143.

Singh, P., and R. Purkait. 2009. Observations of external ear--an Indian study. Homo--Journal of Comparative Human Biology 60: 461-472.

EARWAX

Some people have earwax that is wet, sticky and yellow or brown; other people's earwax is dry, crumbly and grayish. Variation at a single gene determines which kind of earwax you have; the allele for wet earwax is dominant over the allele for dry earwax. The allele for dry earwax appears to have originated by mutation in northeastern Asia about 2,000 generations ago, then spread outwards because it was favored by natural selection. It is very common in eastern Asia, becomes much less common towards Europe, and is rare in Africa.

Earwax type is not used very often to illustrate basic genetics, but unlike most human characters that are used (tongue rolling, attached earlobes, etc.), it really is controlled by a single gene with two alleles.

You can do a Google Images search for "earwax" if you want to get grossed out, but I'd rather look at kittens. Plus, in Japan, wet earwax is known as "cat earwax."

Earwax as a character

Earwax, also known as "cerumen," is produced by glands that line the ear canal. It gradually moves out of the ear, carrying dead skin cells and other debris. Most people have one of two types of earwax. Wet earwax is sticky and yellowish brown to dark brown in color, while dry earwax is crumbly and gray to tan. A small number of people, generally less than 1 percent, have earwax that is intermediate or unclassifiable (Matsunaga 1962, Petrakis et al. 1967; Ing et al. 1973; Petrakis et al. 1990). Petrakis et al. (1986) found that the proportion of white women with dry earwax increased with age, which would suggest that earwax type was not determined solely by genetics, but Nakajima and Hirano (1969) did not find any change with age in Japan.

Family studies

Matsunaga (1962) wrote the first English-language paper on the polymorphism in earwax type, reviewing a large body of Japanese-language literature that dated back to the early 1930s. Data on Japanese families showed the following, where "W" is wet earwax and "D" is dry earwax:

Parents	W offspring	D offspring	Percent W
W × W	35	12	74%
W × D	205	195	51%
D × D	0	634	0%

If the allele for dry is recessive, two parents with dry earwax could not have a child with wet earwax, and this is what the data show. In addition, because the allele for wet earwax is rare in this sample, most W parents would be WD heterozygotes. You would then expect about a 3:1 ratio of W to D in offspring of WxW matings, and about a 1:1 ratio of W to D in WxD matings. The data also fit this prediction. These data support the conclusion that earwax is one-locus, two-allele trait, with the W allele being dominant.

Petrakis et al. (1967) performed a similar study in American Indians, with similar results:

Parents	W offspring	D offspring	Percent W
W × W	32	6	84%
W × D	20	9	69%
D × D	0	42	0%

and so did Nakajima and Hirano (1969) in Japan:

Parents	W offspring	D offspring	Percent W
W × W	27	3	90%
W × D	137	109	56%
D × D	0	345	0%

Molecular genetics

Tomita et al. (2002) used eight Japanese families to determine that the gene for wet/dry earwax is on chromosome 16, near the centromere. Yoshiura et al. (2006) then found the gene responsible: ABCC11 (ATP-binding cassette, subfamily C, member 11). The allele for wet earwax has a G at site 538 of the coding region, which causes an arginine at position 180 in the amino acid sequence; most dry alleles have an A at site 538, coding for a glycine. There is another, less common allele, a deletion of 27 nucleotides in exon 29, that also causes dry earwax. Toyoda et al. (2009) stained the ABCC11 protein in various tissues and found that there was less of the protein in individuals with the AA (dry earwax) genotype, suggesting that the A allele causes a loss of function of the ABCC11 protein.

```
              Ile Ala Ser Val Leu Gly Pro Ile Leu Ile Ile Pro
wet earwax    ATT GCC AGT GTA CTC GGG CCA ATA TTG ATT ATA CCA
dry earwax    ATT GCC AGT GTA CTC AGG CCA ATA TTG ATT ATA CCA
                                  Arg
```

Bases 523-558 of the DNA sequence of ATCC11, along with the amino acid sequence. The DNA polymorphism at site 538 causes the amino acid difference that determines earwax type.

The allele for dry earwax has frequencies close to 100% in people from northern China and Korea, is intermediate in people from Japan, southern Asia, and the Americas, is uncommon in Europe, and is almost completely absent in Africa (Petrakis et al. 1971, Ibraimov 1991, Yoshiura et al. 2006, Ohashi et al. 2011). There are very few polymorphisms in the human genome that show this much difference in allele frequency between one geographic area and another. Ohashi et al. (2011) used patterns of variation at two nearby microsatellite loci and estimated that the allele for dry earwax originated as a new mutation about 2000 generations ago and has spread due to natural selection, with dry earwax individuals having a relative fitness about 1 percent higher than wet earwax individuals. Wet earwax is associated with warmer areas in Europe, Asia and the Americas (Ohashi et al. 2011), so there seems to be something about cooler climates that favors the dry earwax allele.

Earwax and body odor

There are two kinds of sweat glands: eccrine sweat glands, which are found throughout the skin, and apocrine sweat glands, which are found in the armpits and groin. Eccrine sweat glands produce sweat that is mostly water and salt, and it does not contribute very much to body odor. Apocrine sweat contains proteins and lipids; when bacteria on the skin metabolize apocrine sweat, they produce body odor. The earwax glands (ceruminous glands) are a form of apocrine gland.

In Japan, wet earwax has long been associated with greater body odor (Adachi 1937, Matsunaga 1962). Yoo et al. (2006) found that 860 out of 896 patients who consulted a doctor about axillary osmidrosis (the medical term for stinky armpits) had wet earwax, in a population where wet earwax is uncommon. Nakano et al. (2009) genotyped the polymorphism in the ABCC11 gene in Japanese axillary osmidrosis patients and found the genotypes that cause wet earwax in 78 out of 79, while only 35 percent of the general population had wet earwax genotypes; Inone et al. (2010) found similar results in a smaller study. Martin et al (2010) analyzed sweat from people of different ABCC11 genotypes and found that several compounds that are precursors for body odor are absent or in lower concentrations in sweat from people with the dry earwax genotype.

Earwax and breast cancer

Like the earwax glands, the mammary glands are a form of apocrine gland, and the ABCC11 gene is expressed in both. Petrakis (1971) noticed that breast cancer was less common in geographic areas with high frequencies of dry earwax. He found a higher frequency of wet earwax in Japanese-American women with breast cancer than in a control group of Japanese-American women, although the difference was not statistically significant. Ing et al. (1973) surveyed a much larger sample of breast cancer patients and controls in Hong Kong and did not find a significant difference in the frequency of wet earwax.

Ota et al. (2010) compared ABCC11 genotypes in Japanese breast cancer patients and controls and did find a significantly higher frequency of the allele for wet earwax in breast cancer patients; the genotypes for wet earwax were in 25

percent of the cancer patients and 17 percent of the controls. There was no difference in ABCC11 genotype frequency between breast cancer patients and controls in large samples of German (Lang et al. 2011), Australian or Italian (Beesley et al. 2011) women, but fewer than 2 percent of women in those populations have dry earwax.

There is additional evidence that the ABCC11 polymorphism affects the mammary glands. Breast fluids can be collected from some women who are not producing milk, and Petrakis et al. (1981) found that both Asian and Caucasian women with wet earwax were more likely to produce breast fluids. Petrakis et al. (1990) confirmed this pattern, but found that there was a larger difference between wet and dry earwax women for Asian immigrants to the United States than for U.S.-born Asian-Americans, suggesting that the environment also plays a role. Miura et al. (2007) found that women with dry earwax were less likely to produce colostrum (the breast milk produced around childbirth), and those who did produced smaller volumes than women with wet earwax.

Conclusion

Unlike most of the human characters that are used to demonstrate simple genetics principles, wet vs. dry earwax really is controlled by one gene with two alleles. Several factors make it especially attractive as a classroom topic: the gene has been identified, the biochemical basis for the variation is fairly well understood, there is evidence for strong natural selection, and there are links to human health (body odor and possibly cancer). There are, however, two problems with using earwax type in classroom exercises. One is that the allele for dry earwax has a frequency in western European populations of 10 to 20 percent, and it is virtually absent from African populations. Because dry earwax is recessive, this means that a classroom of students of European and African ancestry may show no variation; everyone could have wet earwax. Classrooms in northern China or Korea, on the other hand, may contain only students with dry earwax, because the allele for dry earwax is at nearly 100 percent in those areas.

The second problem with using earwax type in a genetics exercise is that having a bunch of students sitting in a classroom, digging wax out of their ears and waving it around, would be kind of disgusting.

References

Adachi, B. 1937. Das Ohrenschmalz als Rassenmerkmal und der Rassengeruch ("Achselgeruch") nebst dem Rassenunterschied der Schweissdrüsen. Zeitschrift für Rassenkunde und die Gesamte Forschung am Menschen 6: 273-307.

Beesley, J., S. E. Johnatty, X. Q. Chen, A. B. Spurdle, P. Peterlongo, M. Barile, V. Pensotti, S. Manoukian, P. Radice, and G. Chenevix-Trench. 2011. No evidence for an association between the earwax-associated polymorphism in ABCC11 and breast cancer risk in Caucasian women. Breast Cancer Research and Treatment 126: 235-239.

Ibraimov, A.I. 1991. Cerumen phenotypes in certain populations of Eurasia and Africa. American Journal of Physical Anthropology 84: 209-211.

Ing, R., L. Petrakis, and H. C. Ho. 1973. Evidence against association between wet cerumen and breast cancer. Lancet 1: 41.

Inone, Y., T. Mori, Y. Toyoda, A. Sakurai, T. Ishikawa, Y. Mitani, Y. Hayashizaki, Y. Yoshimura, H. Kurahashi, and Y. Sakai. 2010. Correlation of axillary osmidrosis to a SNP in the ABCC11 gene determined by the Smart Amplification Process (SmartAmp) method. Journal of Plastic, Reconstructive and Aesthetic Surgery 63: 1369-1374.

Lang, T., et al. (13 co-authors). 2011. The earwax-associated SNP c.538G>A (G180R) in ABCC11 is not associated with breast cancer risk in Europeans. Breast Cancer Research and Treatment, e-pub ahead of print.

Martin, A., M. Saathoff, F. Kuhn, H. Max, L. Terstegen, and A. Natsch. 2010. A functional ABCC11 allele is essential in the biochemical formation of human axillary odor. Journal of Investigative Dermatology 130: 529–540.

Matsunaga, E. 1962. The dimorphism in human normal cerumen. Annals of Human Genetics 25: 273-286.

Miura, K., K. Yoshiura, S. Miura, T. Shimada, K. Yamasaki, A. Yoshida, D. Nakayama, Y. Shibata, N. Niikawa, and H. Masuzaki. 2007. A strong association between human earwax-type and apocrine colostrum secretion from the mammary gland. Human Genetics 121: 631–633.

Nakajima, A., and I. Hirano. 1969. Distribution and inheritance of earwax types: a study on inhabitants in Awa District, Chiba Prefecture. Japanese Journal of Human Genetics 13: 201-207.

Nakano, M., N. Miwa, A. Hirano, K. Yoshiura, and N. Niikawa. 2009. A strong association of axillary osmidrosis with the wet earwax type determined by genotyping of the ABCC11 gene. BMC Genetics 10:42.

Ohashi, J., I. Naka, and N. Tsuchiya. 2011. The impact of natural selection on an ABCC11 SNP determining earwax type. Molecular Biology and Evolution 28: 849-857.

Ota, I. et al. (17 co-authors). 2010. Association between breast cancer risk and the wild-type allele of human ABC transporter ABCC11. Anticancer Research 30: 5189-5194.

Petrakis, N.L. 1971. Cerumen genetics and human breast cancer. Science 173: 347-349.

Petrakis, N. L., K. T. Molohon, and D. J. Tepper. 1967. Cerumen in American Indians: genetic implications of sticky and dry types. Science 158: 1192-1193.

Petrakis, N.L., U. Pingle, S. J. Petrakis, and S. L. Petrakis. 1971. Evidence for a genetic cline in earwax types in Middle East and south Asia. American Journal of Physical Anthropology 35: 141-144.

Petrakis, N. L., V. L. Ernster, S. T. Sacks, E. B. King, R. J. Schweitzer, T. K. Hunt, M.-C. King. 1981. Epidemiology of breast secretion: Association with breast cancer risk factors and cerumen type. Journal of the National Cancer Institute 67: 277-284.

Petrakis, N.L., S. L. Wiesenfeld, and L. Flander. 1986. Possible influence of age on the expression of the heterozygous cerumen phenotype. American Journal of Physical Anthropology 69: 437-440.

Petrakis, N. L., E. B. King, M. Lee, and R. Miike. 1990. Cerumen phenotype and proliferative epithelium in breast fluids of United States-born vs. immigrant Asian women: a possible genetic-environmental interaction. Breast Cancer Research and Treatment 16: 279-285.

Tomita, H., K. Yamada, M. Ghadami, T. Ogura, Y. Yanai, K. Nakatomi, M. Sadamatsu, A. Masui, N. Kato, and N. Niikawa. 2002. Mapping of the wet/dry earwax locus to the pericentromeric region of chromosome 16. Lancet 359: 2000-2002.

Toyoda, Y., et al. (12 co-authors). 2009. Earwax, osmidrosis, and breast cancer: why does one SNP (538 g> a) in the human ABC transporter ABCC11 gene determine earwax type? FASEB Journal 23:2001–2013.

Yoo, W. M., N. S. Pae, S. J. Lee, T. S. Roh, S. Chung, and K. C. Tark. 2006. Endoscopy-assisted ultrasonic surgical aspiration of axillary osmidrosis: a retrospective review of 896 consecutive patients from 1998 to 2004. Journal of Plastic, Reconstructive and Aesthetic Surgery 59: 978-982.

Yoshiura, K., et al. (39 co-authors). 2006. A SNP in the ABCC11 gene is the determinant of human earwax type. Nature Genetics 38: 324-330.

EYE COLOR

One of the oldest myths in human genetics is that having blue eyes is determined by a single gene, with the allele for blue eyes recessive to the allele for non-blue eyes (green, brown, or hazel). Many people who know nothing else about genetics think that two blue-eyed parents cannot have a brown-eyed child.

Eye color as a character

The color of the iris is determined by the amount of melanin, the ratio of eumelanin (which is dark brown) to pheomelanin (which is reddish), and the way the melanin is distributed in the eye. Irises with little melanin appear blue due to scattering of light by collagen fibers in the iris. Blue, gray, green and hazel eyes are only common in people of European ancestry; other people's eyes are various shades of brown.

Many studies divide eye colors into three categories: blue (or blue and gray); green and hazel; and brown. This has been criticized as an oversimplification (Brues 1975), and eye colors have been divided into nine categories (Mackey et al. 2011) or the hue and saturation values quantified (Liu et al. 2010). Eye color can change dramatically in the first few years of life, as many babies are born with blue eyes but then develop green or brown eyes (Matheny and Dolan 1975), and changes can also occur later in life (Bito et al. 1997, Liu et al. 2010). Some people have a blue or green iris with a brown ring around the pupil (Sturm and Larsson 2009), which makes the classification of eye color even more complicated.

Family studies

Davenport and Davenport (1907) were the first to suggest that blue eye color was caused by a recessive allele. They claimed that whenever both parents had blue eyes, all of the children have blue eyes, but their data actually included two hazel-eyed offspring of blue-eyed parents. The authors said "we suspect [these] to be of a blue type," whatever that means.

Hurst (1908) divided eyes into just two types, "simplex" (S, blues and some grays, with no pigment on the outer surface of the iris) and "duplex" (D, all other colors). He found the following results:

Parents	D offspring	D offspring	Percent D
D × D	240	18	93%
D × S	187	137	58%
S × S	0	101	0%

Because there are no "duplex" (non-blue-eyed) offspring of two blue-eyed parents, these data fit the model of blue eyes being caused by a recessive allele at one gene.

Holmes and Loomis (1909) criticized the earlier work, saying that eye color varies continuously, and dividing it into categories is arbitrary. Out of 52

offspring of two blue-eyed parents in their data, one had brown eyes and two had gray eyes, which does not fit the idea that blue eyes are caused by a recessive allele. Boas (1918) found an even larger number of non-blue-eyed offspring of two blue-eyed parents, 26 out of 223. Surprisingly, there don't seem to have been any parent-offspring studies of eye color since then, at least none that I could find.

Molecular genetics

A number of groups surveyed associations of single-nucleotide polymorphisms with eye color, with fairly consistent results: variation in the HERC2 and OCA2 genes, which are next to each other on chromosome 15, plays a major role in determining eye color. However, variation in at least 10 other genes, plus complicated interactions between these genes, also influences eye color (reviewed in Sturm and Larsson 2009, with more recent results in Liu et al. 2010 and Pospiech et al. 2011). The large number of genes that influence eye color means that it is not the simple character controlled by a single gene that the myth describes.

Conclusion

Eye color is not an example of a simple genetic trait, and blue eyes are not determined by a recessive allele at one gene. Instead, eye color is determined by variation at several different genes and the interactions between them, and this makes it possible for two blue-eyed parents to have brown-eyed children.

References

Bito, L. Z., A. Matheny, K. J. Cruickshanks, D. M. Nondahl, and O. B. Carino. 1997. Eye color changes past early childhood: the Louisville Twin Study. Archives of Ophthalmology 115: 659-663.

Boas, H. M. 1918. Inheritance of eye color in man. American Journal of Physical Anthropology 2: 15-20.

Brues, A. M. 1975. Rethinking human pigmentation. American Journal of Physical Anthropology 43: 387-391.

Davenport, G. C., and C. B. Davenport. 1907. Heredity of eye color in man. Science 26: 589-592.

Holmes, S. J., and H. M. Loomis. 1909. The heredity of eye color and hair color in man. Biological Bulletin 18: 50–65.

Hurst, C. C. 1908. On the inheritance of eye-colour in man. Proceedings of the Royal Society of London B 80: 85-96.

Liu, F., et al. (20 co-authors). 2010. Digital quantification of human eye color highlights genetic association of three new loci. PLOS Genetics 6: e1000934.

Mackey, D. A., C. H. Wilkinson, L. S. Kearns, and A. W. Hewitt. 2011. Classification of iris colour: review and refinement of a classification schema. Clinical and Experimental Ophthalmology 39: 462-471.

Matheny, A. P., and A. B. Dolan. 1975. Changes in eye color during early childhood: sex and genetic differences. Annals of Human Biology 2: 191-196.

Pospiech, E., J. Draus-Barini, T. Kupiec, A. Wojas-Pelc, and W. Branicki. 2011. Gene-gene interactions contribute to eye colour variation in humans. Journal of Human Genetics 56: 447-455.

Sturm, R. A., and M. Larsson. 2009. Genetics of human iris colour and patterns. Pigment Cells and Melanoma Research 22: 544-562.

HAIR COLOR

Some people have red hair, and some have hair that is various shades of blond or brown. The myth is that red hair is determined by a single gene, with the allele for red being recessive to alleles for other colors.

Hair color as a character

Hair color is determined by the amount of eumelanin (which is dark brown) and pheomelanin (which is reddish). The amount of eumelanin ranges continuously from very little, producing light-blonde hair, to large amounts, producing black hair. People with large amounts of pheomelanin have red hair, which can range from pale red ("strawberry blond") to bright red to reddish brown.

Most studies divide hair color into a small number of categories, such as blond, red, and brown. Reed (1952) criticized the subjectiveness of this approach and used a reflectance spectrophotometer to measure the amount of light reflected by hair at different wavelengths. He found that there was no clear separation of hair into two categories; instead there were some individuals with intermediate hair that could not easily be classified as red or non-red. Reed (1952) also examined hair under a microscope and found the reddish pheomelanin granules that are common in red hair were also in some individuals with brown hair. This suggests that when the red pheomelanin pigment is present, whether a person has red hair is determined by the amount of brown eumelanin pigment that the person also has.

Family studies

The variety of hair colors makes it difficult to summarize the results of family studies in detail. Davenport and Davenport (1909) found numerous examples of two brown-haired parents having red-haired offspring, which would suggest that it is determined by a recessive allele, but Neel (1943) found 13 out of 114 offspring of two red-haired parents to have non-red hair. Reed (1952) reviewed the various hypotheses that had been proposed, including that red was recessive; that red was dominant; that red was dominant, but could be masked by brown; or that red was usually recessive but could sometimes be dominant.

Molecular genetics

Valverde et al. (1995) surveyed DNA sequence variation in the melanocortin 1 receptor (MC1R) gene. They found several amino acid variants that were found in red-haired people but rare in non-red people. Box et al. (1997) identified the three most common amino acid polymorphisms that are associated with red hair: R151C, R160W, and D294H. This shorthand means that the common amino acids at positions 151, 160 and 294 in the protein are arginine (R), arginine, and aspartic acid (D), while the amino acids cysteine (C), tryptophan (W), and histidine (H) are found in redheads. Most alleles have only one of these

three red-associated amino acids; for example, some alleles have cysteine at position 151 but arginine and aspartic acid at positions 160 and 294.

There are a large number of rare amino acid polymorphisms in the MC1R gene, some of which may also be associated with red hair (Beaumont et al. 2007). Sulem et al. (2007) surveyed genetic variation throughout the genome of a large sample of Icelanders and found that MC1R is the only gene with a strong association with red hair. However, knowing the genotype of an individual at the MC1R locus is not enough to predict whether they have red hair. Beaumont et al. (2007) found that only 74% of individuals who were homozygous for tryptophan at position 160 have red hair, while 4% of individuals who were heterozygous for this amino acid had red hair. Box et al. (1997) found five pairs of dizygotic twins which had identical genotypes for the MC1R gene, yet one twin had red hair and the other didn't. Sulem et al. (2007) used the variation at the MC1R gene to try to predict hair color, and about a third of the individuals who were predicted to have red hair actually had blond or brown hair.

Evolution

The alleles associated with red hair are fairly common in northern European populations; in Britain and Ireland, the R151C allele has a frequency of about 10 percent, while R160W and D294H are at 9 and 2 percent (Gerstenblith et al. 2007). Red hair is rare in other populations, which has led to speculation that the alleles for red hair were favored by selection by differing amounts of ultraviolet radiation, since red hair is associated with pale skin (Jablonski and Chaplin 2010) and is most common in areas with gloomy winters. Harding et al. (2000) applied several statistical tests for DNA sequence data and found no evidence that the large amount of amino acid variation at MC1R resulted from positive selection; in particular, the large number of amino acid sites that vary within human populations are comparable to the large number of amino acid differences between human and chimp MC1R. Harding et al. (2000) did not find any amino acid variation within African populations, so they concluded that there is strong negative selection there (new amino acid mutations are selected against). They concluded that the variation outside of Africa reflects a relaxation of negative selection allowing new alleles to drift in frequency, rather than new alleles being favored. In contrast, Savage et al. (2008) concluded that MC1R was affected by positive selection, based on the greater geographic variation in allele frequency than most human genes, greater levels of polymorphism, and an unusually large number of low frequency polymorphisms.

Conclusion

Red hair color is not an example of a simple genetic trait. While the amount of red pigment may be mainly determined by one gene (MC1R), there are a large number of different MC1R alleles, and other genes affecting the amount of brown pigment that plays a major role in determining hair color. The complicated genetics means that it is possible for two red-haired parents to have non-red-haired children.

References

Beaumont, K. A., S. L. Shekar, R. A. Newton, M. R. James, J. L. Stow, D. L. Duffy, and R. A. Sturm. 2007. Receptor function, dominant negative activity and phenotype correlations for MC1R variant alleles. Human Molecular Genetics 16: 2249-60.

Box, N., J. Wyeth, L. O'Gorman, N. Martin, and R. Sturm. 1997. Characterization of melanaocyte stimulating hormone receptor variant alleles in twins with red hair. Human Molecular Genetics 6:1891–1897.

Davenport, G. C., and C. B. Davenport. 1909. Heredity of hair color in man. American Naturalist 43: 193-211.

Gerstenblith, M. R., A. M. Goldstein, M. C. Fargnoll, K. Peris, and M. T. Landi. 2007. Comprehensive evaluation of allele frequency differences of MC1R variants across populations. Human Mutation 28: 495-505.

Harding, R. M., et al. (11 co-authors). 2000. Evidence for variable selective pressures at MC1R. American Journal of Human Genetics 66: 1351-1361.

Jablonski, N.G., and G. Chaplin. 2010. Human skin pigmentation as an adaptation to UV radiation. Proceedings of the National Academy of Sciences 107: 8962-8968.

Neel, J. V. 1943. Concerning the inheritance of red hair. Journal of Heredity 34: 93-96.

Reed, T. E. 1952. Red hair colour as a genetical character. Annals of Eugenics 17: 115-139.

Savage, S. A., M. R. Gerstenblith, A. M. Goldstein, L. Mirabello, M. C. Fargnoll, K. Peris, and M. T. Landi. 2008. Nucleotide diversity and population differentiation of the melanocortin 1 receptor gene, MC1R. BMC Genetics 9: 31.

Sulem, P., et al. (25 co-authors). 2007. Genetic determinants of hair, eye and skin pigmentation in Europeans. Nature Genetics 39: 1443-1452.

Valverde, P., E. Healy, I. Jackson, J. L. Rees, and A. J. Thody. 1995. Variants of the melanocyte-stimulating hormone receptor gene are associated with red hair and fair skin in humans. Nature Genetics 11: 328-330.

HAIR WHORL

Clockwise hair whorl; counterclockwise whorl.

When viewed from above and behind the head, many people's hair whorls in either a clockwise (CW) or counterclockwise (CCW) direction. This is sometimes used to illustrate basic genetics; the myth is that whorl direction is controlled by a single gene with two alleles, and the allele for clockwise is dominant to the allele for counterclockwise. Several studies have data that fit this myth fairly well, but with enough exceptions that the myth can't be completely true.

While hair whorl is occasionally used to illustrate basic genetics, it gets more attention because some studies have suggested that counterclockwise whorls are more common in left-handed people than in right-handed, and other studies have suggested that counterclockwise whorls are more common in gay men than in straight men. Other studies have given conflicting results, and it is not clear yet whether there is any relationship between hair-whorl direction and either handedness or sexuality.

Hair whorl as a character

In people with short, straight hair, a single whorl is usually fairly obvious. Clockwise whorls are most common; estimates of the frequency of clockwise whorls range from 51 percent in Japan (Klar 2009) to 65 percent of undergraduate psychology students in the United Kingdom (Annett 1985), 69 percent of Nigerians (Ucheya and Igweh 2005), 74 percent of German schoolboys (Bernstein 1925), 81 percent of students in the United States (Lauterbach and Knight 1927), 92 percent of the "general population" in Maryland (Klar 2003), and 94 percent of newborns in the United States (Wunderlick and Heerema 1975). Some people have two or more whorls; Lauterbach and Knight (1927) found 5 percent of white Massachusetts schoolchildren to have double whorls, while Schwarzburg (1927) found 5.4 percent of Germans to have double whorls. Wunderlich and Heerema (1975) found double whorls in 1.5 percent of white newborns, while Ucheya and Igweh (2005) found double whorls in 2 percent of Nigerian men.

The direction of the whorl can be difficult to determine. Rahman et al. (2009) had two people independently judge photographs of whorls, and for about 4 percent of subjects, one observer called the whorl clockwise while the other called it counterclockwise. The whorl can be particularly hard to see in people with long or curly hair. Lauterbach (1925) says "frequently the hair requires considerable combing in order to discover the natural whorl... Long, fine hair sometimes assumes a false whorl but the true whorl can always be located

close to the scalp." Ziering and Krenitsky (2003) reported that 78 percent of women had what they called a "diffuse" pattern instead of a whorl. Wunderlich and Heerema (1975) could see a hair whorl in only 10 percent of black newborns, and Ziering and Krenitsky (2003) reported that 80 percent of African-American men had a diffuse pattern instead of a whorl. Ucheya and Igweh (2005), however, identified a whorl in all 500 of their sample of Nigerian men.

Family studies

Bernstein (1925) compared parents and offspring, with the following results (omitting double whorls):

Parents	CW offspring	CCW offspring	Percent CW
CW × CW	58	12	83%
CW × CCW	32	8	80%
CCW × CCW	3	8	27%

He concluded that hair whorl was a simple Mendelian trait, with the allele for clockwise whorl dominant to the allele for counterclockwise whorl. However, if the myth were true, two parents with counterclockwise whorls could not have a child with a clockwise whorl, so the three CW children of CCW x CCW parents do not fit the myth.

Schwarzburg (1927), Kloepfer (1946), Beckman et al. (1960), and Sharma (1985) collected similar data. Adding all of their numbers to those of Bernstein (1925) yields the following:

Parents	CW offspring	CCW offspring	Percent CW
CW × CW	385	56	87%
CW × CCW	140	57	71%
CCW × CCW	3	16	16%

The data clearly indicate that there is a genetic influence on the direction of the hair whorl, because CW x CW parents have a much higher percentage of CW offspring than do CCW x CCW parents. However, the three CW offspring of CCW x CCW matings, which all come from Bernstein (1925), do not fit the simple model that CW is completely dominant. Because other researchers have shown that it can be difficult to determine the direction of whorl (Rahman et al. 2009), it is possible that the 3 people who do not fit the model really had CCW whorls but were observed incorrectly (or one of their parents was incorrectly called CCW).

Klar (2003) proposed a "random recessive" model for the inheritance of hair whorl, in which there are two alleles, R for clockwise whorl and r for random whorl direction. In this model, RR and Rr individuals have clockwise hair whorls, while half of rr individuals have clockwise whorls and half have counterclockwise whorls. Klar (2005) reanalyzed the data of Schwarzburg (1927) and concluded that the data fit the random-recessive model better than the model in which CW is dominant. However, Klar's random-recessive model predicts that all CCW individuals have the rr genotype, so all of the offspring of CCW x CCW matings should be rr and therefore half the offspring of CCW x

CCW matings should be CCW. Instead, the data in the table above show three CW and 16 CCW offspring from CCW x CCW matings. This is significantly different from the prediction of Klar's random-recessive model (exact binomial test, P=0.004). So the data don't fit Klar's random-recessive model very well, either.

Twin studies

Rife (1933) looked at 13 pairs of monozygotic twins. Seven of the pairs had opposite whorls: clockwise in one twin, counterclockwise in the other. Sharma (1985) looked at the hair whorls of 27 pairs of identical twins and found one pair with opposite whorls. The pairs of twins that differ in their whorl direction do not fit the simple model of the trait being completely determined by genetics.

Hair whorl and handedness

Klar (2003) surreptitiously observed people in Maryland shopping malls, most of whom would be right-handed; of the 500 people with clearly visible, single whorls, 8.4% had counterclockwise whorls. He also surveyed 49 non-righthanded people (this includes left-handed and ambidextrous individuals) and found that 44.9% had counterclockwise whorls. Klar (2003) concluded that the much higher proportion of counterclockwise whorls in left-handed people fit a random recessive model. In this model, a person with an RR or Rr genotype is right-handed and has a clockwise whorl. A person with an rr genotype has a 50 percent chance of being left-handed and a 50 percent chance of having a counterclockwise whorl; therefore half of all left-handed people would have a counterclockwise whorl, and half of all people with a counterclockwise whorl would be left-handed.

Klar's work received a lot of media attention, and led to several followup studies and renewed attention to some earlier work. In all of these, the hair whorl was observed at close range with the permission of the subject, rather than being observed surreptitiously from a distance the way Klar did. I summarize the data below; "CCW" is counterclockwise whorl, "RH" is right-handed, "NRH" is non-right-handed, "N" is the number of RH or NRH individuals. "Sig" is whether the difference in CCW percentage between RH and NRH is statistically significant. I omitted people with double whorls from the percentages.

Reference	Subjects	CCW% in RH	N	CCW% in NRH	N	sig?
Lauterbach and Knight (1927)	White children	19.3	877	20.6	63	no
Newman (1934)	American	15	?	21	?	no
Annett (1985)	British adults	34.9	209	38.0	38	no
Scott et al. (2005)	Pennsylvania whites	13.8	123	11.1	36	no
Beaton and Mellor (2007)	Welsh men	12.8	125	54.2	48	yes
Jansen et al. (2007)	German soldiers	18.3	981	19.7	127	no
Schmidt et al. (2008)	German children	10.2	177	32.5	40	yes
Perelle et al. (2009)	New York adults	15.6	179	22.7	66	no
Schwartz et al. (2010)	North Am. men	18.7	1168	16.0	206	no

If Klar's random recessive model were correct, you'd expect to see a very low frequency of counterclockwise whorls in righthanded people, while about half of non-righthanded people would have counterclockwise whorls. The results of

Beaton and Mellor (2007) and Schmidt et al. (2008) support Klar's model, but the other studies do not.

The results from the different studies are puzzling; there is no obvious difference in the way the studies were conducted, or the populations studied, that would explain why some studies find a difference and other studies do not. For now, it is not clear whether there is an association between handedness and hair whorl direction. This could be an interesting class project, if enough care was taken to be unbiased in observing the direction of whorl. Rahman et al. (2009) are a good example of how to do this; they took a photograph of each person's whorl, then had two people who were unaware of the purpose of the study independently judge whether it was clockwise or counterclockwise.

Hair whorl and male sexuality

Klar (2004) surreptitiously recorded the direction of hair whorl at a beach near Rehoboth Beach, Delaware that is popular among gay men. Out of 272 men with single whorls, 29.8 percent had counterclockwise whorls. This was a higher proportion than the 9.1 percent counterclockwise he counted in 328 men from malls, stores, and the beach at Atlantic City, most of whom would be straight. This result got a lot of attention in the popular press (France 2007) and is mentioned on a lot of web pages about "gaydar." However, two more rigorous studies did not find a significant difference between gay and straight men in the proportion of counterclockwise whorls. Rahman et al. (2009) and Schwartz et al. (2010) separated gay and straight men based on a questionnaire, not which beach they went to, and determined their whorl type based on close examination, not from a distance. Rahman et al. (2009) found 18 percent of gay and 14 percent of straight men to have counterclockwise whorls, while Schwartz et al. (2010) found 19.7 percent in gay men and 17.2 percent in straight men. In both studies, the observer determined whether the whorl was clockwise or counterclockwise without knowing whether the subject was gay or straight, which may be an important difference between these studies and that of Klar (2004).

Conclusion

It's hard to determine which way the hair whorls in people with long or curly hair, and the data do not fit the simple genetic model perfectly. So you should not use hair whorl direction to demonstrate basic genetics.

References

Annett, M. 1985. Left, right, hand and brain: The right shift theory. London, UK: Erlbaum.

Beaton, A.A., and G. Mellor. 2007. Direction of hair whorl and handedness. Laterality 12: 295-301.

Beckman, L., J.A. Böök, and E. Lander. 1960. An evaluation of some anthropological traits used in paternity tests. Hereditas 46: 543-569.

Bernstein, F. 1925. Beiträge zur Mendelistischen Anthropologie. II. Quantitative Rassenanalyse auf Grund von statistischen Beobachtungen über den Derhsinn des Kopfhaarwirbels. Sitzungsberichte der Prüssischen Akademie der Wissenschaften. 71-82.

France, D. 2007. The science of gaydar. New York, June 24, 2007, pp. 32-37, 98-99.

Jansen, A., H. Lohman, S. Scharfe, C. Sehlmeyer, M. Deppe, and S. Knecht. 2007. The association between scalp hair-whorl direction, handedness and hemispheric language dominance: Is there a common genetic basis of lateralization? NeuroImage 35: 853-861.

Klar, A.J.S. 2003. Human handedness and scalp hair-whorl direction develop from a common genetic mechanism. Genetics 165: 269-276.

Klar, A.J.S. 2004. Excess of counterclockwise scalp hair-whorl rotation in homosexual men. Journal of Genetics 170: 2027-2030.

Klar, A.J.S. 2005. A 1927 study supports a current genetic model for inheritance of human scalp hair-whorl orientation and hand-use preference traits. Genetics 170: 2027-2030.

Klar, A.J.S. 2009. Scalp hair-whorl orientation of Japanese individuals is random; hence, the trait's distribution is not genetically determined. Seminars in Cell and Developmental Biology 20: 510-513.

Kloepfer, H.W. 1947. An investigation of 171 possible relationships in man. Annals of Human Genetics. 32-70.

Lauterbach, C.E. 1925. Studies in twin resemblance. Genetics 10: 525-568.

Lauterbach, C.E., and J.B. Knight. 1927. Variation in whorl of the head hair. Journal of Heredity 18: 107-115.

Mishra, S.K., and M.K. Singhal. 1995. The scalp hair whorl: its relation with age, sex, handedness and cephalic index. Journal of the Anatomical Society of India 44: 134-143.

Newman, H.H. 1934. Dermatoglyphics and the problem of handedness. American Journal of Anatomy 55: 277-322.

Perelle, I.B., Ehrman, L., and Chanza, M. 2009. Human handedness and scalp hair whorl direction: no evidence for a common cause. Laterality 14: 95-101.

Rahman, Q., Clarke, K., and Morera, T. 2009. Hair whorl direction and sexual orientation in human males. Behavioral Neuroscience 123: 252-256.

Rife, D.C. 1933. Genetic studies of monozygotic twins. III. Mirror imaging. Journal of Heredity 24: 443-446.

Schmidt, H., Depner, M., and Kabesch, M. 2008. Medial position and counterclockwise rotation of the parietal scalp hair-whorl as a possible indicator for non-right-handedness. Scientific World Journal 8: 848-854.

Schwartz, G., Kim, R.M., Kolundzija, A.B., Rieger, G., and Sanders, A.R. 2010. Biodemographic and physical correlates of sexual orientation in men. Archives of Sexual Behavior: 39: 93-109.

Schwarzburg, V. W. 1927. Statistische untersuchungen uber den menschlichen scheitelwirbel und seine vererbung. Zeitsch. F. Morphol. U. Anthropol. 26: 195-223.

Scott, N.M., S.M. Weinberg, K. Neiswanger, C.A. Brandon, and M.L. Marazita. 2005. Hair whorls and handedness: informative phenotypic markers in nonsyndromic cleft lip with or without cleft palate (NS CL/P) cases and their unaffected relatives. American Journal of Medical Genetics 136A: 158-161.

Sharma, K. 1985. Inheritance of occipital hair whorls in man: a twin and family study. Indian Journal of Physical Anthropology and Human Genetics 11: 17-23.

Ucheya, R.E., and J.C. Igweh. 2005. Hair whorl patterns on the posterior aspect of the scalp among Nigerians. Journal of Experimental and Clinical Anatomy 4: 21-24.

Wunderlich, R.C., and N.A. Heerema. 1975. Hair crown patterns of human newborns. Clinical Pediatrics. 14: 1045-1049.

Ziering, C., and G. Krenitsky. 2003. The Ziering whorl classification of scalp hair. Dermatologic Surgery 29: 817-821.

HAND CLASPING

When people clasp their hands, almost all have a strong preference; either the right thumb is on top (R) or the left thumb is on top (L). Hand-clasping is sometimes used to illustrate basic genetics; the myth is that hand-clasping is controlled by one gene with two alleles, and the allele for L is dominant. I do not know how this myth started, as from the first study (Lutz 1908) onwards, it has been clear that hand-clasping preference does not fit this simple myth.

Left thumb on top; right thumb on top.

Hand clasping as a character

Most people have a strong preference for clasping their hands in one way, either with the left thumb on top (L) or the right thumb on top (R). To most people, it feels unnatural to clasp the hands in the opposite way, making it a very easy trait to observe. Surveys indicate that roughly half of the people studied are R and half are L (Wiener 1932, Freire-Maia et al. 1958, Lai and Walsh 1965, Reiss 1999). Reiss (1998, 1999) reviewed nearly 100 publications that have surveyed hand-clasping frequencies in populations around the world. There were a few populations with particularly high or low frequencies of left-over-right claspers, but most populations had between 40 and 75 percent L. The proportion of people with no preference for L or R was about 1 percent.

Family studies

Reiss (1999) summarized data from 18 studies of parents and offspring, with the following totals across all studies:

Parents	L offspring	R offspring	Percent L
L × L	1252	880	59%
L × R	2309	2573	47%
R × R	1298	2815	32%

Reiss (1999) concluded that there may be some genetic basis for this character (because LxL matings produce more L offspring than do RxR matings), but it is not a simple one-gene, two-allele genetic character. If the myth were true, two R parents could not have an L child, but almost a third of the children of RxR matings are L. In the first study on this character, Lutz (1908) reached the same

conclusion based on the same kind of data, and it is not clear what the purpose was of the 17 family studies done in the subsequent 90 years.

Twin studies

Reiss (1999) summarized data from four twin studies. In all of the studies, there are many pairs of twins where one is L and the other is R, indicating that there is little genetic influence on this character:

	Monozygotic	Dizygotic
Both L	106	101
Both R	103	67
L + R	157	131

Conclusion

From the very first study, almost a hundred years ago, it has been clear that hand clasping is not a simple genetic trait. This has been repeatedly demonstrated using both family studies and twin studies. It is not clear where the idea that L is dominant over R came from, unless it is the unfortunately common misconception that common = dominant. You should not use hand clasping to demonstrate basic genetics.

References

Freire-Maia, N., A. Quelce-Salgado, and A. Freire-Maia. 1958. Hand clasping in different ethnic groups. Human Biology 30: 281-291.

Lai, L.Y.C., and R.J. Walsh. 1965. The patterns of hand clasping in different ethnic groups. Human Biology 37: 312-319.

Lutz, F.E. 1908. The inheritance of the manner of clasping the hands. American Naturalist 42.495: 195-196.

Reiss, M. 1998. Händefalten--eine Übersicht. Anthropol. Anz. 56: 163-.

Reiss, M. 1999. The genetics of hand-clasping: A review and a familial study. Annals of Human Biology 26: 39-48.

Wiener, A.S. 1932. Observations on the manner of clasping the hands and folding the arms. American Naturalist 66: 365-370.

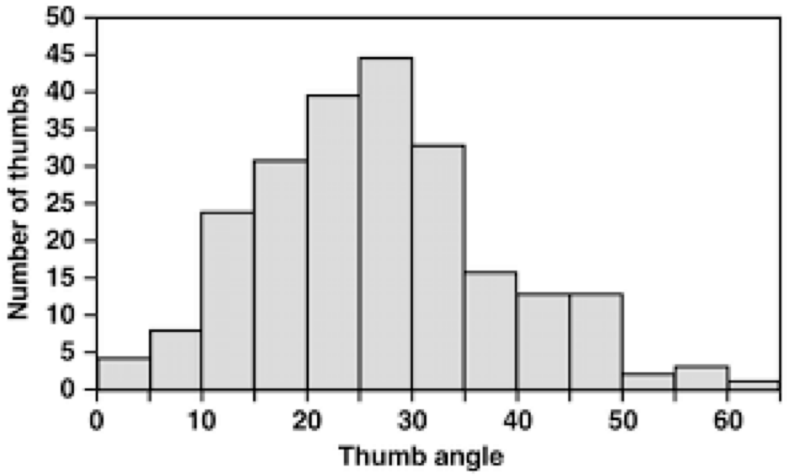
HITCHHIKER'S THUMB

Thumbs ranging from straight to hitchhiker.

Some people have "hitchhiker's thumbs," which bend backwards with a large angle between the two segments (phalanges). The myth is that there are just two kinds of thumbs, straight thumbs (S) and hitchhiker's thumbs (H), and the trait is controlled by a single gene with two alleles, with the allele for S being dominant. This was proposed by Glass and Kistler (1953) and has been the subject of very little research since then. Hitchhiker's thumb is often used to demonstrate genetics, but neither part of the myth is true: thumbs don't fall into two discrete categories, and the trait is not controlled by a single gene.

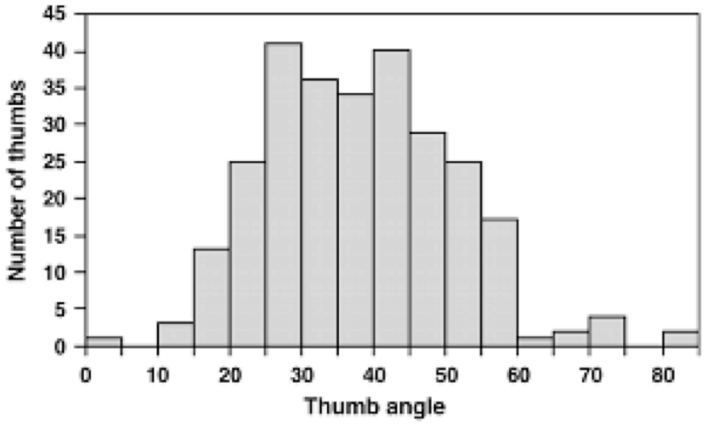
Hitchhiker's thumb as a character

Harris and Joseph (1949) measured the angle between the first and second phalanges of the thumb on X-rays of 294 individuals. They found a continuous distribution, with most individuals having intermediate values, not the two distinct kinds of thumbs described in the myth:



Distribution of thumb angles. Data for right thumbs for males and females combined (Harris and Joseph 1949).

Glass and Kistler (1953) did a similar study, except they used a protractor held against the outside of the thumb to measure the thumb angle, and obtained the following results:



Distribution of thumb angles. Data for right thumbs for males and females combined (Glass and Kistler 1953).

Glass and Kistler (1953) arbitrarily called all thumbs with an angle equal to or greater than 50 degrees "hitchhiker's thumbs." They gave no explanation for why they picked 50 degrees as the dividing point. They noted that many individuals would then have one hitchhiker's thumb and one straight thumb; they classified these individuals as having the hitchhiker's thumb trait. Glass and Kistler (1953) had different people measure the thumbs, and the repeated measurements often differed by several degrees, which would mean that many people would be considered to have a hitchhiker's thumb by one observer but not by a different observer.

I searched the internet for pictures of thumbs (it was easy, because lots of people give the thumbs-up sign when they get their picture taken) and arranged them from straightest to most bent. As you can see, there's a range of thumb angles, from straight to nearly 90 degrees, with no clear division between hitchhiker and non-hitchhiker thumbs.

Family studies

Glass and Kistler (1953), having decided that anyone with one or both thumbs having an angle equal to or greater than 50 degrees had the hitchhiker's thumb trait, collected the following family data:

Parents	S offspring	H offspring	Percent S
S × S	281	32	90%
S × H	71	37	66%
H × H	1	30	3%

They concluded that thumb type was a simple Mendelian trait, with the allele for S dominant. The single individual who didn't fit this model (the S offspring of two H parents) was explained as an example of incomplete penetrance, meaning that other genes or non-genetic factors also influence the trait. Glass and Kistler (1953) were too polite to mention incorrect paternity as another possible explanation.

The only other family study on hitchhiker's thumb that I am aware of is Beckman et al. (1960):

Parents	S offspring	H offspring	Percent S
S × S	50	8	86%
S × H	18	17	51%
H × H	3	4	43%

If the myth were true, two parents with hitchhiker's thumb could not have a child with a straight thumb. The three S offspring of H x H parents are inconsistent with a two-allele model in which the allele for S is dominant.

Conclusion

While there is a genetic influence on thumb angle, thumbs do not divide into two categories, hitchhiker's and non-hitchhiker's. Instead, the thumb angle varies continuously, with most thumbs having intermediate values. You should not use hitchhiker's thumb to demonstrate basic genetics.

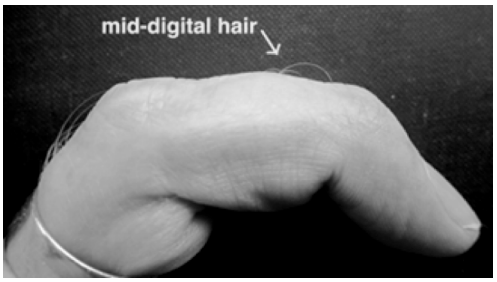
References

Beckman, L., J.A. Böök, and E. Lander. 1960. An evaluation of some anthropological traits used in paternity tests. Hereditas 46: 543-569.

Glass, B., and J.C. Kistler. 1953. Distal hyperextensibility of the thumb. Acta Genetica 4: 192-206.

Harris, H., and J. Joseph. 1949. Variation in extension of the metacarpo-phalangeal and interphalangeal joints of the thumb. Journal of Bone and Joint Surgery 31: 547-559.

MID-DIGITAL HAIR



Some people have hair on the back of the middle segment of some fingers. Individuals with hair on the middle segment of at least one finger are considered to have the trait mid-digital hair (H); other people have no mid-digital hair (N).

Mid-digital hair is often used to illustrate basic genetics; the myth is that the presence or absence of mid-digital hair is controlled by a single gene with two alleles, and the allele for H is dominant. Several genetic studies show that this myth is not true.

Mid-digital hair as a character

The population frequency of people with mid-digital hair ranges from 2 percent in Eskimos to about 75 percent in people of northern European ancestry (Saldanha and Guinsburg 1961). Danforth (1921) was the first to examine mid-digital hair as a character. (Garn [1951] said "middle phalangeal hair" would be more accurate, as "mid-digital hair" implies hair on the middle finger; the scientific literature mostly uses his term, while genetics textbooks and web pages tend to use "mid-digital hair.") He pointed out the difficulties in scoring this trait: the hairs can be on all four fingers, or just one, and can range from numerous and thick to scarce and very fine. In some cases, hair follicles are present but no hairs are visible, which Danforth and most subsequent researchers counted as having hair present. Hair is most common on the ring finger, then the middle finger and pinkie; mid-digital hair is rare on the index finger.

Bernstein and Burke (1942) reported that mid-digital hair was present in about half of females under 21, with a slightly higher percentage in males, but less than 20 percent of women over 21 had mid-digital hair. They suggested that housework wore away the mid-digital hair and hair follicles, which would complicate its use as a genetic trait.

Ikoma (1973) surveyed the left hands of over 7,500 people in Japan. About 24 percent had at least one finger with mid-digital hair. The most common patterns were hair on just the ring finger or the ring finger and middle fingers, with middle, ring, and pinkie fingers being somewhat less common. Less than one percent of people had mid-digital hair on their index finger.

Saldanha and Guinsburg (1961) reported that over 10 percent of a sample of Brazilians had mid-digital hair on one hand but not the other. Other studies (Danforth 1921, Bernstein and Burks 1942, Ikoma 1973) only looked at the left hand, so they would have classified some people as N who actually had mid-digital hair on their right hand.

Family studies

Danforth (1921) was the first to examine the genetic basis of mid-digital hair by comparing parents and offspring. Unfortunately, his sample of 80 families did not include any N x N matings. He concluded that his data were fairly consistent with H being dominant, but with some evidence that multiple genes or environmental factors influenced the trait.

Bernstein and Burks (1942) collected family data, with the following results:

Parents	H offspring	N offspring	Percent H
H × H	60	12	83%
H × N	54	55	50%
N × N	20	214	9%

They suggested that H was dominant over N. If this were true, two N parents could not have a H child. Bernstein and Burks (1942) say the 20 H offspring of N x N matings could be explained if some of the "N" mothers were really H women whose mid-digital hair follicles had been worn away by housework.

Kloepfer (1946), Bernstein (1949), and Beckman and Böök (1959) collected family data, with similar results:

Parents	H offspring	N offspring	Percent H
H × H	38	12	76%
H × N	26	16	62%
N × N	1	7	13%

Kloepfer (1946)

Parents	H offspring	N offspring	Percent H
H × H	114	14	89%
H × N	46	33	58%
N × N	9	51	15%

Bernstein (1949)

Parents	H offspring	N offspring	Percent H
H × H	42	13	76%
H × N	31	26	54%
N × N	9	21	30%

Beckman and Böök (1959)

Hindley and Damon (1973) collected family data in the Solomon Islands. They note that they did not use a magnifying glass, so they may have missed small, fine hairs and follicles without hair. They also had blood group data, so they were confident that all the parents were correctly identified (no hidden adoptions or false paternity). They obtained the following results:

Parents	H offspring	N offspring	Percent H
H × H	6	10	38%
H × N	17	21	45%
N × N	11	13	46%

Unlike previous studies, these data show no genetic influence on mid-digital hair; about the same proportion of children have mid-digital hair for all three mating types.

Twin studies

Sommer (1971) looked at the fingers of 28 pairs of identical twins, and found one pair in which one twin had mid-digital hair while the other twin did not. This is evidence that the trait is affected by the environment as well as genetics.

Conclusion

Family studies show that mid-digital hair is not a simple genetic trait. It may be that the genetics of the trait are more complicated, or it may be that there is an environmental influence (such as hand work wearing away the hairs). In either case, you should not use mid-digital hair to demonstrate simple genetics.

References

Beckman, L., and J.A. Böök. 1959. Distribution and inheritance of mid-digital hair in Sweden. Hereditas 45: 215-220.

Bernstein, M.M., and B.S. Burke. 1942. The incidence and mendelian transmission of mid-digital hair in man. Journal of Heredity 33: 45-53.

Danforth, C.H. 1921. Distribution of hair on the digits in man. American Journal of Physical Anthropology 4: 189-204.

Garn, S.M. 1951. The use of middle-phalangeal hair in population studies. American Journal of Physical Anthropology 9: 325-334.

Hindley, S.W., and A. Damon. 1973. Some genetic traits in Solomon Island populations. IV. Mid-phalangeal hair. American Journal of Physical Anthropology 39: 191-194.

Ikoma, E. 1973. A genetic study on digital hair. Japanese Journal of Human Genetics 18: 259-273.

Kloepfer, H.W. 1946. An investigation of 171 possible relationships in man. Annals of Eugenics. 13: 35-71.

Saldanha, P.H., and S. Guinsburg. 1961. Distribution and inheritance of middle phalangeal hair in a white population of Sao Paulo, Brazil. Human Biology 33: 237-249.

Sommer, K. 1971. Untersuchungen zur Genetik des Merkmals "Fingerbehaarung." Humangenetik 11: 155-168.

PTC Tasting

To some people, small amounts of the compounds phenylthiocarbamide (PTC) or propylthiouracil (PROP) taste very bitter; other people do not taste these compounds. The myth is that there are only two kinds of people, tasters and non-tasters, and that the trait is controlled by a single gene, with the allele for tasting dominant over the allele for non-tasting.

PTC tasting as a character

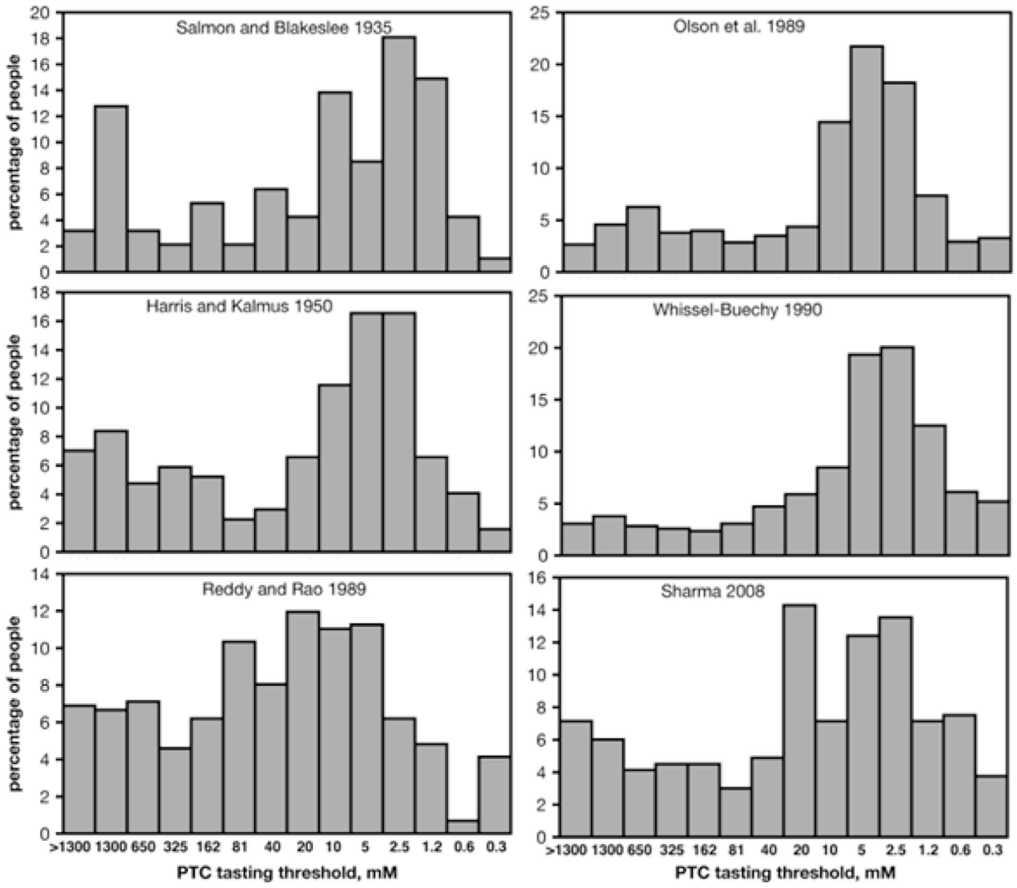
Fox (1932) was working in a lab with phenylthiocarbamide (PTC) when a colleage complained about the bitter taste of the chemical dust Fox was spreading around. Fox insisted that it was tasteless; to settle the disagreement, he had other colleagues taste the PTC and discovered that it had a strong bitter taste for some people, while others found it tasteless. This led to a large body of research on PTC tasting; Guo and Reed (2001) review the subject and cite 392 references, and there has been more work done since then. For most of the myths in this series, I had to dig deep to find every obscure scrap of information, but for PTC, I am just skimming the surface. The reviews by Guo and Reed (2001), Kim and Drayna (2004) and Wooding (2006) are good places to start if you'd like more information.

Testing methods

Some of the early studies put PTC crystals directly on the tongue, while others used solutions of PTC or paper soaked in PTC and then dried. However, some people would be classified as tasters with one technique and non-tasters with a different technique (Hartmann 1939, Lawless 1980).

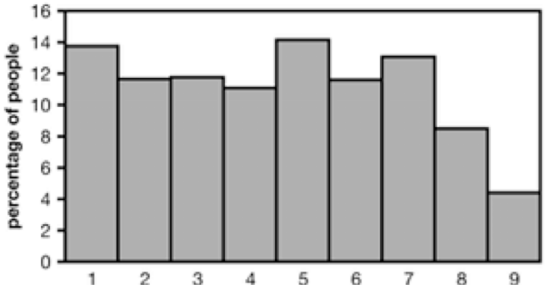
The most common method for measuring the ability to taste PTC involves finding the weakest concentration of PTC that tastes different from plain water (Blakeslee 1932). The technique of Harris and Kalmus (1950) for threshold measurement has been widely used, sometimes with small modifications. The subject is given a two-fold dilution series of PTC, starting with the weakest concentration and going up until they say they can taste it. The subject is then asked to sip four PTC solutions of that concentration and four plain waters, and identify which are PTC. If they get it correct, the next weaker solution is tried; if they get it incorrect, the next stronger solution is tried. The weakest PTC solution that the subject can correctly identify is the threshold.

Harris and Kalmus (1950) found that the distribution of PTC tasting thresholds was bimodally distributed, but there were some intermediate individuals. Other studies have found similar results, a bimodal distribution with some intermediate individuals (Blakeslee 1932, Salmon and Blakeslee 1935, Falconer 1947, Olson et al. 1989, Whissell-Buechy et al. 1990, Guo et al. 1998, Drayna et al. 2003).



Percentage of people with different PTC-tasting thresholds. >1300" indicates people who could not taste PTC at the highest concentration, 1300 mM.

A different way to measure PTC tasting is to give each subject an intermediate concentration of PTC solution, then ask them to rate it on a numeric scale, such as 0 (no taste) to 7 (very strong taste) (Lawless 1980). This category rating method is much quicker than the threshold detection method of Harris and Kalmus (1950) and exposes the subjects to much less PTC. It may also do a better job of separating people into two distinct categories, taster and non-taster (Lawless 1980), but it has not been used very often.



Percentage of people with different scores on a PTC bitterness scale. Data from the three genotypes in Khataan et al. (2009)

In classrooms, the usual way to test PTC tasting is by having students taste a piece of paper that has been soaked in PTC. Some individuals who are classified as tasters using paper are non-tasters using a threshold test, and vice

versa (Hartmann 1939, Lawless 1980). Khataan et al. (2009) asked subjects to taste a piece of paper containing 3 μg of PTC and rate it from 1 (not at all bitter) to 9 (extremely bitter). The variation among 911 subjects was not at all bimodal.

Individual variation

Salmon and Blakeslee (1935) measured the PTC threshold of 12 people repeatedly over a period of several days. Each subject had different thresholds at different times; in the most extreme case, one person sometimes had a threshold of 0.4 mM while another time had a threshold of 100 mM. In other subjects tested repeatedly throughout a day, the threshold varied by as much as eightfold over as short as 15 minutes.

Whissell-Buechy (1990) retested 30 subjects after one year. Using a dividing line between taster and non-taster based on a much larger sample, three of the 30 retested individuals would have changed between "taster" and "non-taster."

A number of studies have reported that the sensitivity to PTC is lower (the threshold concentration is higher) in older people (Harris and Kalmus 1950, Olson et al. 1989, Whissel-Buechy 1990). Kalmus and Trotter (1962) retested 110 people after about 15 years and found an increase in the average threshold; two people changed between taster and non-taster.

PTC versus PROP

Wheatcroft and Thornburn (1972) pointed out that PTC has been found to be toxic in rats, and that while the Harris and Kalmus (1950) protocol recommends spitting out the test solutions, a complete non-taster who swallowed all of the solutions would swallow an amount of PTC that is frighteningly close to a lethal dose. Many studies have used propylthiouracil (PROP) instead of PTC. Like PTC, PROP tastes very bitter to some people and is tasteless to others. Lawless (1980) found that there was a general agreement between the two compounds: most people who found a dilute solution of PTC to be bitter also found a dilute solution of PROP to be bitter. However, the range of thresholds for PROP was narrower and the distribution less bimodal than for PTC, making it even more difficult to separate people into tasters and non-tasters with PROP. Bufe et al. (2005) found that the association with TAS2R38 haplotypes was stronger for PTC than for PROP.

Family studies

Blakeslee (1932) divided individuals into PTC tasters (T) and non-tasters (NT) and obtained the following results from a set of 103 families:

Parents	T offspring	NT offspring	Percent NT
T × T	109	22	83%
T × NT	42	32	57%
NT × NT	0	22	0%

Because there were no taster offspring of two non-taster parents, this fits the model of PTC tasting being determined by two alleles of a single gene, with tasting being dominant over non-tasting.

Merton (1958) examined 60 Norwegian families and found similar results, except that there were five taster offspring of two non-taster parents:

Parents	T offspring	NT offspring	Percent NT
T × T	73	16	82%
T × NT	36	14	72%
NT × NT	5	30	14%

and Das (1958) examined 126 Indian families:

Parents	T offspring	NT offspring	Percent NT
T × T	191	47	80%
T × NT	137	71	66%
NT × NT	4	37	10%

Das (1958) took a closer look at the four taster offspring of two non-taster parents. One had a PTC threshold right on the borderline between taster and non-taster. The other three came from a single mother; blood groups revealed that her husband was not the biological father of her three children.

Olson et al. (1989) also found some taster offspring of two non-taster parents:

Parents	T offspring	NT offspring	Percent NT
T × T	194	19	91%
T × NT	117	28	81%
NT × NT	18	26	41%

They examined blood group data on families of 11 of the taster offspring of two non-taster parents, and three of the offspring were the result of extra-pair mating; the other eight really were taster offspring of two non-taster biological parents.

If PTC tasting were a simple one-gene, two-allele genetic character, with tasting completely dominant to non-tasting, then two non-tasting parents could not have a tasting child. Both Merton (1958) and Olson et al. (1989) found some tasting offspring of non-tasting parents, so the trait must be more complicated than the myth says. The discrepancy could be due to more complicated genetics, involving multiple alleles or multiple genes, or some kind of environmental influence.

Twin studies

Kaplan et al. (1967) tested the taste threshold for PROP on pairs of twins, and found that pairs of monozygotic twins were more similar than pairs of dizygotic twins. However, out of 75 pairs of monozygotic twins, seven of the pairs had one taster twin and one non-taster twin. Martin (1975) looked at 28

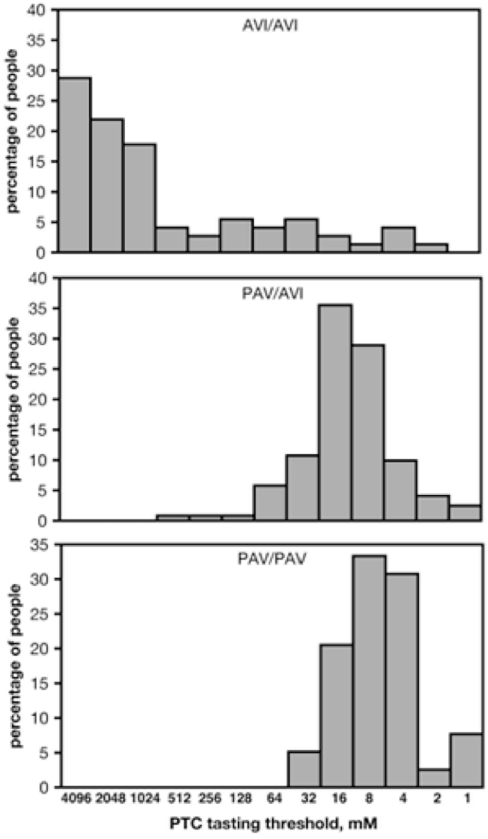
pairs of monozygotic twins and did not find any taster/non-taster pairs. Sharma (2008) found that none of a sample of 66 monozygotic twin differed by more than 3 units on the Harris-Kalmus scale of PTC threshold, while 26 out of 75 dizygotic twins differed by more than 3 units.

Molecular genetics

Drayna et al. (2003) used linkage to DNA polymorphisms in 26 large families and found that much of the variation in PTC tasting was associated with chromosome 7, while some variation was associated with chromosome 16.

Kim et al. (2003) extended the linkage mapping of Drayna et al. (2003) and found that variation in a single gene, TAS2R38, explains much of the variation in PTC tasting. The gene has a single exon that is 1002 base pairs long. There are three amino acid polymorphisms: alanine/proline (A/P) at amino acid position 49, valine/alanine (V/A) at position 262, and isoleucine/valine (I/V) at position 296. The three polymorphisms have 8 possible combinations (haplotypes), but two make up more than 95 percent of all haplotypes in European and Asian populations: AVI and PAV, named after the amino acids at positions 49, 262 and 296. AVI/AVI homozygotes are mostly non-tasters, while AVI/PAV heterozygotes and PAV/PAV homozygotes are mostly tasters. The AAV haplotypes was present at a frequency of 3 percent in Europe, while AAV, AAI and PVI were found only in sub-Saharan African populations. Several nonhuman primate species had the PAV haplotype, meaning that the AAV and other haplotypes originated after the human lineage split from the chimp lineage.

The average PTC tasting thresholds were lowest for PAV/PAV homozygotes, slightly higher for AVI/PAV heterozygotes, and much higher for AVI/AVI homozygotes (Kim et al. 2003). There was, however, considerable overlap, suggesting that PTC tasting threshold is affected by other genes or environmental factors.



Percentage of people with PTC tasting thresholds, for the three common haplotypes at the TAS2R38 gene. Combined data from the Utah and NIH samples in Kim et al. (2003).

Wooding et al. (2004) sequenced the coding region of TAS8R38 in 165 humans, one common chimp and one gorilla. They applied a number of statistical tests for evidence of natural selection. There were more intermediate-frequency polymorphisms than expected for a human gene, which suggests that balancing selection may be affecting the gene.

Bufe et al. (2005) gave people different concentrations of PTC and asked them to rate the bitterness. At the higher concentrations, there was no overlap in bitterness score between AVI/AVI homozygotes and PAV/PAV homozygotes, but AVI/PAV heterozygotes had a range of bitterness scores that overlapped both homozygotes. They did the same experiment with PROP and found a much weaker association with TAS2R38 genotype; the highest concentration of PROP tasted very bitter to some AVI/AVI individuals and did not taste bitter to some PAV/PAV individuals.

Conclusion

PTC tasting is largely determined by a single gene, TAS2R8, with two common alleles, and the allele for tasting is mostly dominant over the allele for non-tasting. However, both classical family and twin studies, and modern molecular genotyping, show that there are other genes or environmental factors that influence PTC tasting. As a result, there is a continuous range of PTC tasting, not absolute separation into tasters and non-tasters. PTC tasting would be a fascinating subject for an advanced genetics class, but it does not fit the one-gene, two-allele myth well enough to be used to demonstrate simple Mendelian genetics.

References

Blakeslee, A. F. 1932. Genetics of sensory thresholds: taste for phenyl thio carbamide. Proceedings of the National Academy of Sciences of the USA 18: 120-130.

Bufe, B., P. A. S. Breslin, C. Kuhn, D. R. Reed, C. D. Tharp, J. P. Slack, U.-K. Kim, D. Drayna, and W. Meyerhof. 2005. The molecular basis of individual differences in phenylthiocarbamide and propylthiouracil bitterness perception. Current Biology 15: 322-327.

Das, S. R. 1958. Inheritance of the PTC taste character in man: An analysis of 126 Rarhi Brahmin families of West Bengal. Annals of Human Genetics 22: 200-212.

Drayna, D., H. Coon, U.-K. Kim, T. Elsner, K. Cromer, B. Otterud, L. Baird, A. P. Peiffer, and M. Leppert. 2003. Genetic analysis of a complex trait in the Utah Genetic Reference Project: a major locus for PTC taste ability on chromosome 7q and a secondary locus on chromosome 16p. Human Genetics 112: 567-572.

Falconer, D.S. 1947. Sensory threshholds for solutions of phenyl-thio-carbamide: results of tests on a large sample, made by R.A. Fisher. Annals of Eugenics 13: 211-222.

Fox, A. L. 1932. The relationship between chemical constitution and taste. Proceedings of the National Academy of Sciences of the USA 18: 115-120.

Guo, S. W., and D. R. Reed, 2001 The genetics of phenylthiocarbamide perception. Annals of Human Biology 28: 111–142.

Guo, S. W., F. M. Shen, Y. D. Wang, and C. J. Zheng. 1998. Threshold distributions of phenylthiocarbamide (PTC) in the Chinese population. Annals of the New York Academy of Sciences 855: 810-812.

Harris, H., and H. Kalmus. 1950. The measurement of taste sensitivity of phenylthiourea (PTC). Annals of Eugenics 15: 24-31.

Hartmann, G. 1939. Application of individual taste difference towards phenyl-thio-carbamide in genetic investigations. Annals of Eugenics 9: 123-135.

Kalmus, H., and W. R. Trotter. 1962. Direct assessment of the effect of age on PTC sensitivity. Annals of Human Genetics 26: 145-149.

Kaplan, A. R., R. Fischer, A. Karras, F. Griffin, W. Powell, R. W. Marsters, and E. V. Glanville. 1967. Taste threshholds in twins and siblings. Acta Geneticae Medicae et Gemollologiae 16: 229-241.

Khataan, N. H., L. Stewart, D. M. Brenner, M. C. Cornelis, and A. El-Sohemy. 2009. TAS2R38 genotypes and phenylthiocarbamide bitter taste perception in a population of young adults. Journal of Nutrigenetics and Nutrigenomics 2: 251-256.

Kim, U.-K., and D. Drayna. 2004. Genetics of individual differences in bitter taste perception: lessons from the PTC gene. Clinical Genetics 67: 275-280.

Kim, U. K., E. Jorgenson, H. Coon, M. Leppert, N. Risch, and D. Drayna. 2003. Positional cloning of the human quantitative trait locus underlying taste sensitivity to phenylthiocarbamide. Science 299: 1221-1225.

Lawless, H. 1980. A comparison of different methods used to assess sensitivity to the taste of phenylthiocarbamide (PTC). Chemical Senses 5: 247-256.

Martin, N. G. 1975. Phenylthiocarbamide testing in a sample of twins. Annals of Human Genetics 38: 321-326.

Merton, B. B. 1958. Taste sensitivity to PTC in 60 Norwegian families with 176 children: confirmation of the hypothesis of single gene inheritance. Acta Genetica et Statistica Medica 8: 114-128.

Olson, J. M., M. Boehnke, K. Neiswanger, A. F. Roche, and R. M. Siervogel. 1989. Alternative genetic models for the inheritance of the phenylthiocarbamide taste deficiency. Genetic Epidemiology 6: 423-434.

Salmon, T.N., and A. F. Blakeslee. 1935. Genetics of sensory thresholds: variations within single individuals in taste sensitivity for PTC. Proceedings of the National Academy of Sciences of the USA 21: 78-83.

Sharma, K. 2008. Comparing sensory experience in bitter taste perception of phenylthiocarbamide within and between human twins and singletons: Intrapair differences in thresholds and genetic variance estimates. Anthropologischer Anzeiger 66: 211-224.

Wheatcroft, P. E. J., and C. C. Thornburn. 1972. Toxicity of the taste testing compound phenylthiocarbamide. Nature New Biology 235: 93-94.

Whissell-Buechy, D. 1990. Effects of age and sex on taste senstitivity to phenylthiocarbamide (PTC) in the Berkeley Guidance sample. Chemical Senses 15: 39-57.

Wooding, S. 2006. Phenylthiocarbamide: A 75-year adventure in genetics and natural selection. Genetics 172: 2015-2023.

Wooding, S., U.-K. Kim, M. J. Bamshad, J. Larsen, L. B. Jorde, and D. Drayna. 2004. Natural selection and molecular evolution in PTC, a bitter-taste receptor gene. American Journal of Human Genetics 74:637-646.

TOE LENGTH

In some people, the big toe is longer than the second toe (here called "L," for long big toe), while other people have the big toe shorter than the second toe ("S"). This is sometimes said to controlled by one gene with two alleles, with the allele for S dominant to the allele for L. There is no good evidence for this myth; the small number of studies of toe length give contradictory results.

Toe length as a character

I did an image search for painted toenails and arranged the images from longer big toes to longer second toes. As you can see, the relative length of the big and second toes varies continuously; there aren't just two categories of toe length. Some studies have found about 5 percent of the populations sampled to have the big toe and second toe equal in length (Romanus 1952, Turgut et al. 1997). Hawkes (1914) said the big and second toes were the same length in only 0.1 percent of people. She also said about 6 percent of people had the big toe longer on one foot and the second toe longer on the other foot.

Toes ranging from big toes longer (upper left) to big toe shorter (lower right).

Family studies

Hawkes (1914) compared British parents and offspring, with the following results (individuals who were L on one foot and S on the other are omitted):

Parents	S offspring	L offspring
S × S	8	1
S × L	10	58
L × L	0	154

She concluded that L was "irregularly dominant" over S. In reality, these data are consistent with S being dominant over L. If L were dominant, some of the L x L matings would be of two heterozygotes, and some of their offspring would be S.

Beckman et al. (1960) compared Swedish parents and offspring, with the following results:

Parents	S offspring	L offspring
S × S	3	1
S × L	6	31
L × L	3	56

While there is clearly a genetic component, the one L offspring of S x S parents does not fit the model of S being recessive, while the three S offspring of L x L parents do not fit the model of S being dominant.

Papadopoulos and Damon (1973) performed a similar study on four tribes in the Solomon Islands:

Parents	S offspring	L offspring	Percent S
S × S	74	46	62%
S × L	63	102	38%
L × L	51	137	27%

In this study, S parents are more likely to have S offspring than are L parents, so there seems to be some genetic influence on toe length. However, if the myth were true, two S parents could not have an L child, yet more than a quarter of the children of S x S matings are L. The large number of S individuals with two L parents is inconsisent with the myth that this is a simple one-gene, two-allele genetic character, with S completely dominant to L. The difference between the results from Great Britain, which fit the simple two-allele model, and the results from the Solomon Islands is puzzling; it is possible that the genetics of toe length are just more complicated in the Solomon Islands than in Great Britain.

Twin studies

Kaplan (1964) found that out of 63 pairs of monozygotic twins, none had one L twin and one S twin, while 11 out of 44 pairs of dizygotic twins had one L and one S twin. This clearly indicates a strong genetic influence on this trait, although it does not indicate whether toe length is controlled by one or more than one gene.

Conclusion

Whether the big toe is longer or shorter than the second toe is influenced by genetics, but it may be determined by more than one gene, or by a combination of genetics and the environment. You should not use toe length to demonstrate basic genetics.

References

Beckman, L., J.A. Böök, and E. Lander. 1960. An evaluation of some anthropological traits used in paternity tests. Hereditas 46: 543-569.

Hawkes, O. A. M. 1914. On the relative lengths of the first and second toes of the human foot, from the point of view of occurrence, anatomy and heredity. Journal of Genetics 3: 249-274.

Kaplan, A. R. 1964. Genetics of relative toe lengths. Acta Geneticae Medicae et Gemellologiae 13: 295-304.

Papadopoulos, C. C., and A. Damon. 1973. Some genetic traits in Solomon Island populations. III. Relative toe lengths. American Journal of Physical Anthropology 39: 185-190.

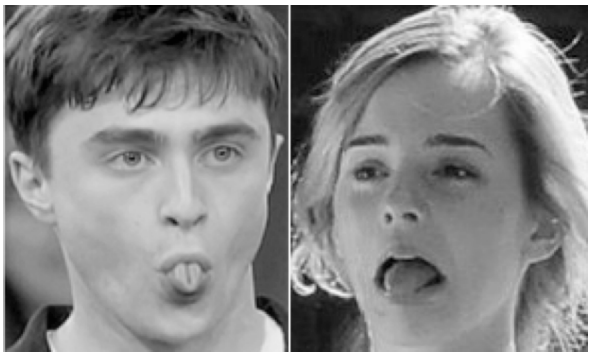
Romanus, T. 1952. Frequency of long second toe in Swedish children from Uppland province. Acta genetica 3: 159-167.

TONGUE ROLLING

Some people can roll their tongue into a tube, and some people can't. This is one of the most common traits that biology teachers use to demonstrate basic genetic principles. Alfred Sturtevant (one of the pioneers of *Drosophila* genetics) described tongue rolling as a simple two-allele character, with the allele for rolling (usually given the symbol T or R) being dominant over the allele for non-rolling (t or r) (Sturtevant 1940). Many studies have shown that the myth is incorrect, but tongue rolling remains a popular subject in genetics classes.

Tongue rolling as a character

Most people, when first asked, either can easily roll their tongue (here called "R"), or cannot roll it at all ("NR"). The proportion of people who can roll their tongue ranges from 65 to 81 percent, with a slightly higher proportion of tongue-rollers in females than in males (Sturtevant 1940, Urbanowski and Wilson 1947, Liu and Hsu 1949, Komai 1951, Lee 1955). However, some people, especially children, cannot roll their tongue when first asked but later learn to do so (Sturtevant 1940). Komai (1951) found that the proportion of tongue-rollers among Japanese schoolchildren increased from 54 percent at ages 6-7 to 76 percent at age 12, suggesting that over 20 percent of the population learns to tongue-roll during that age range. That some people learn to roll their tongues after first being unable to is the first evidence that this is not a simple genetic character. There are also some people who can only slightly roll the edges of their tongue and cannot easily be classified as rollers or non-rollers (Reedy et al. 1971).



Rolled tongue; non-rolled tongue.

Family studies

Sturtevant (1940) compared parents and offspring, with the following results:

Parents	R offspring	NR offspring
R × R	28	5
R × NR	33	22
NR × NR	4	9

He concluded that tongue rolling was at least partially genetic, with rolling dominant to non-rolling, despite the four R offspring of NR x NR parents.

Komai (1951) performed a similar study with much larger sample sizes, and found similar results:

Parents	R offspring	NR offspring	Percent R
R × R	928	104	90%
R × NR	468	217	68%
NR × NR	48	92	34%

In both family studies, individuals with tongue-rolling parents are much more likely to be tongue-rollers than individuals with non-rolling parents. It is difficult to imagine how the common family environment could influence tongue-rolling, so this resemblance between relatives suggests that there is a large genetic influence on tongue-rolling.

However, if this trait were a simple one-gene, two-allele genetic character, with rolling completely dominant to non-rolling, then two non-rolling parents could not have a rolling child. Both studies found rolling offspring of non-rolling parents, so the trait must be more complicated than the myth says. The discrepancy could be due to more complicated genetics, involving multiple alleles or multiple genes, or some kind of environmental influence.

Twin studies

Matlock (1952) found that out of 33 pairs of monozygotic (identical) twins, 7 pairs consisted of one R and one NR twin. This clearly establishes that there are important non-genetic influences on tongue rolling, and it convinced Sturtevant (1965) that tongue rolling was not determined solely by genetics. Reedy et al. (1971) and Martin (1975) also found numerous pairs of monozygotic twins who differed in tongue rolling. Dizygotic twins were twice as likely to differ in tongue-rolling ability as monozygotic twins (Reedy et al. 1971), which is additional evidence that there is some genetic influence on this trait.

	Matlock 1952 MZ	Reedy et al. 1971 MZ	Martin 1975 MZ	Reedy et al. 1971 DZ
Both R	18	43	15	81
Both NR	8	11	5	21
R + NR	7	7	8	30

Conclusion

Family studies clearly demonstrate that tongue rolling is not a simple genetic character, and twin studies demonstrate that it is influenced by both genetics and the environment. Despite this, tongue rolling is probably the most commonly used classroom example of a simple genetic trait in humans. Sturtevant (1965) said he was "embarrassed to see it listed in some current works as an established Mendelian case." You should not use tongue rolling to demonstrate basic genetics.

References

Komai, T. 1951. Notes on lingual gymnastics. Frequency of tongue rollers and pedigrees of tied tongues in Japan. Journal of Heredity 42: 293-297.

Lee, J. W. 1955. Tongue-folding and tongue-rolling in an American Negro population sample. Journal of Heredity 46: 289-291.

Liu, T. T., T. C. Hsu. 1949. Tongue-folding and tongue-rolling in a sample of the Chinese population. Journal of Heredity 40: 19-21.

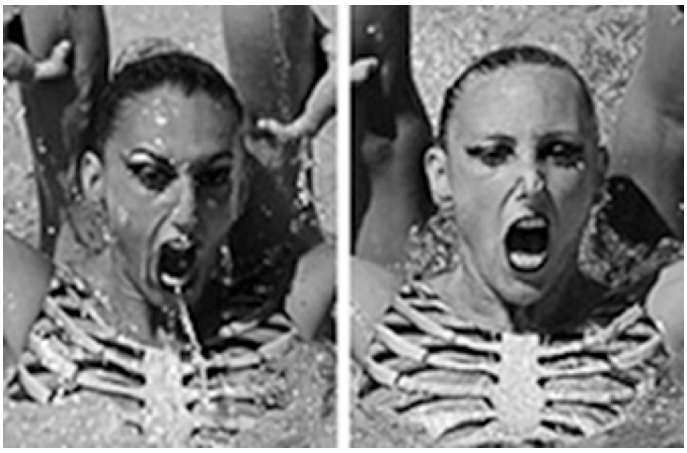
Martin, N. G. 1975. No evidence for a genetic basis of tongue rolling or hand clasping. Journal of Heredity 66: 179-180.

Matlock, P. 1952. Identical twins discordant in tongue-rolling. Journal of Heredity 43: 24.

Sturtevant, A. H. 1940. A new inherited character in man. Proceedings of the National Academy of Sciences USA 26: 100-102.

Sturtevant, A. H. 1965. A History of Genetics. Harper and Row, New York, NY.

WIDOW'S PEAK



Widow's peak (left); straight hairline (right).

Some people have a prominent V-shaped point at the front of their hairline, called a widow's peak, while other people have a hairline that goes straight across. Widow's peak is sometimes used to illustrate basic genetics; the myth is that it is controlled by one gene with two alleles, and the allele for widow's peak is dominant over the allele for straight hairline. There is no evidence that this myth is true. I do not know how the myth got started; I have searched the literature thoroughly, and I have not found any scientific papers that make this claim.

Widow's peak as a character

I found only two papers that have looked at widow's peak in the general population. Smith and Cohen (1973) looked at photographs of male medical students and concluded that 32 out of 1039 (3%) had a "slight but noticeable" widow's peak and one had a "more distinctive and obvious" widow's peak. Nusbaum and Fuentefria (2009) looked at 360 women in hair salons and concluded that 81% of them had a widow's peak.

Unfortunately, neither of these sets of authors defined what they counted as a widow's peak. To find a picture of people with and without a widow's peak, I searched for images of synchronized swimmers (because they wear their hair pulled back, and because they look goofy). I had to look at a lot of pictures before I found the clear widow's peak in the picture above, so I think that Nusbaum and Fuentefria (2009) must have used a very loose definition of widow's peak if they counted 81% of women as having one. Unless there is a very large difference between males and females, Smith and Cohen (1973) must have used a much stricter definition in their study.

In addition to ambiguities about who does or does not have a widow's peak, there is the problem of age. The hairline of many men recedes over time, and it often recedes more slowly in the middle. It could therefore be difficult to distinguish between a receding hairline and a true widow's peak in adult men.

Family and twin studies

There are several papers on people with rare genetic disorders who are more likely to have a widow's peak; see the OMIM entry at http://www.ncbi.nlm.nih.gov/omim/194000. However, I do not know of any family or twin studies of widow's peak in the general population. Unless I've failed to find some old paper, there is no published evidence that widow's peak has a genetic basis.

Conclusion

It is hard to draw a firm line between widow's peak and straight hairline, and there is no published evidence about whether it is influenced by genetics. You should not use widow's peak to demonstrate simple genetics.

References

Nusbaum, B.P., and S. Fuentefria. 2009. Naturally occurring female hairline patterns. Dermatologic Surgery 35: 907-913.

Smith, D.W., and M.M. Cohen. 1973. Widow's peak scalp-hair anomaly and its relation to ocular hypertelorism. Lancet 2: 1127-1128.