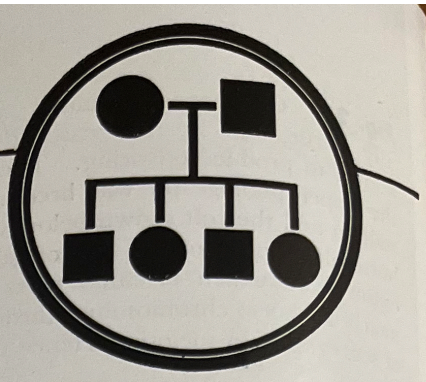
Winchester, A. M. 1951. Genetics: A survey of the principles of heredity. Houghton Mifflin Company, Cambridge, Massachusetts.

These are pages from Winchester (1951) that discuss many of the human traits covered in Myths of Human Genetics (http://megalorchestia.com/mythintro.html). Winchester's textbook is of historic interest, because I suspect that his confident assertions about the simple inheritance of these traits has been passed down from textbook to textbook. I'm not sure it all started with Winchester; someone interested in the history of biology education could do an interesting study on the treatment of these traits in textbooks over time. For dimples and widow's peak, Winchester (1951) is the first reference I could find on the trait, so I don't know where he got the idea that both traits are dominant. If you know of an earlier reference on the genetics of dimples or widow's peak, please email me. Thanks!

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CHAPTER TWENTY-FIVE



SURVEY OF HUMAN HEREDITY

To most students human heredity is naturally one of the most interesting phases of the study of genetics. Recognizing this fact, we have used the inheritance of human characteristics as illustrations of many genetic principles throughout this book. But a great many other human characteristics have been studied genetically besides those we have so far mentioned, and since some knowledge of these may be useful as well as interesting, we shall devote this chapter to a catalogue of some of these and to giving some information about their mode of inheritance.

At the outset it must be stressed once more that much of our information is incomplete regarding the inheritance of certain traits, and present conclusions are often tentative. This is to be expected because of the lack of evidence from experiments, but other difficulties also may prevent an easy classification of inherited characters. One of these difficulties is that the same trait may behave in different ways at different times. For instance, the inheritance of **syndactyly** (the presence of a flap of skin between the second and third toes) acts as a sex-linked recessive in many family pedigrees, but in some others it is inherited as an autosomal recessive. As a result we must assume that two of the genes involved in the growth of the skin on this part of the body may mutate to produce indistinguishable phenotypic effects. Indeed, there are even a few cases where *three* genes are known to produce similar effects — one an autosomal dominant, the second an autosomal recessive, and the third a sex-linked recessive. Since we know that many genes are involved in the production of nearly every characteristic, there is nothing surprising about this or contradictory to the principles of genetics which we have learned, though this variety of causes for the same effect does somewhat confuse the picture in those few cases where it exists.

The student of human heredity will find that many quantitative variations of human characteristics can be explained only by multiple gene inheritance. Multiple genes also participate in reducing the penetrance or altering the expressivity of many human variations which depend primarily on a single gene difference. Finally, surgery is often employed early in life to remedy inherited defects. This, of course, does not alter the genes, but it may make it very difficult to determine whether or not a person possessed a certain characteristic at birth. Clubfootedness, for instance, may be hereditary, but skilled orthopedic treatment and surgery in early childhood can correct the condition, so that an adult may not even know that he was born with clubfeet. Hence, these tabulations of the method of inheritance of human characteristics are presented with the warning that they must be taken with reservation as to their absolute accuracy. They may be of

value, nevertheless, as a preliminary guide in the survey of family histories which may extend our knowledge of human heredity.

Hair

Color

The great range of continuous variations in hair color suggests that this human character is influenced by multiple genes. Children resulting from marriages between persons of a dark haired race and persons with very blonde hair are typically dark-haired. This indicates an overall dominance of the genes for dark hair over the genes for light hair. In a mixed population, such as our own, the children of parents with opposite extremes of hair color are often intermediate because the dark-haired parents are heterozygous for a number of genes involved in this character. Marriages between two light blondes in our population almost invariably yield light blonde children, a fact which again points to the recessive nature of blonde genes as a whole.

Red hair, however, seems to result from a single pair of recessive genes. The children of two red-headed parents practically always have red hair. Also, red-haired children may be born to parents who are not red-headed and, in such families, approximately one-fourth of the children show this trait, as would be expected on the basis of simple Mendelian inheritance. Cockayne studied a number of families in which one parent had red hair and at least one child had red hair. This combination would indicate that the other parent was heterozygous. He found forty-six red-haired children and forty-eight children who did not have red hair in such families, a result which is very close to the 1:1 ratio which would be expected between homozygous recessive and heterozygous dominant persons (see pages 64–65). Incidentally, hair color may have some relation to the endocrine secretions. A baby born of a dark-haired mother may have dark hair at birth because of the influence of the hormones of the mother even though its genotype indicates blonde or red hair. When this first hair falls out and is replaced by a new growth the lighter color will be produced.

There seem to be two primary pigments in the hair, each of which is subject to quantitative variation through multiple genes and multiple alleles. One of these is a black melanin which may result in black, brown, sepia, or light blonde hair with varying shades between. Upon this is superimposed another melanin pigment which may range from sandy-red to yellow. Various genes influence the intensity and the quality of these pigments in such a way as to produce the wide variation in hair color which is characteristic of man.

We learned in Chapter 18 that the recessive gene for albinism interferes with the production of any melanin in the hair as well as in the skin and the eyes. This gene, therefore, is epistatic to all of the genes involved in the production of this pigment in the hair. Albinos often have some color in their hair as a result of other pigments. Negro albinos, especially, tend to have sandy-red hair, and white persons often have some reddish or golden-yellow pigmentation.

Under the general topic of hair color we may also mention the graying of hair. There is not complete agreement as to the exact cause of this phenomenon, for one theory holds that it results from the production of a substance known as leucokeratin, while another maintains that it is due to the formation of air bubbles in the hair shaft. We do know that the age at which the hair begins turning gray is influenced by heredity. It is difficult to establish the exact method of inheritance, however, because of the environmental effects of diet and other factors. In certain family pedigrees, premature grayness is inherited as an autosomal dominant.

Some persons are born with a white forelock of hair which is given the genetic designation of **blaze.** This isolated island of white hair grows out from the central portion of the scalp where it joins the forehead and is usually combed back over the head to

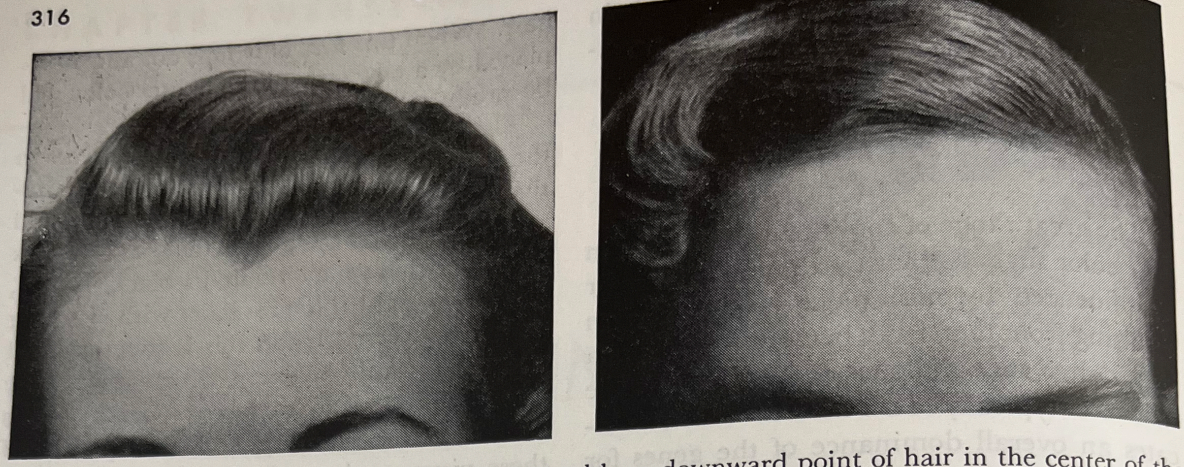


Fig. 208 Widow's peak (left) is characterized by a downward point of hair in the center of the forehead. The gene for this seems to be dominant over the gene for a continuous hair line (right).

produce a white streak in a head of hair otherwise normal in pigmentation. This condition results from an autosomal dominant gene. A similar condition, called **white spotting,** produces the white forelock and also produces colorless spots on the skin in other parts of the body. This also is inherited as a simple dominant.

Form of hair

The form of hair is dependent primarily upon its shape in cross-section. Straight hair is rounded, while wavy, curly, and kinky hair show progressive degrees of flattening. No doubt a number of genes are involved in hair form, but in the white race the evidence indicates that a single pair of alleles produce the difference between curly and straight hair, with the heterozygote showing wavy hair. In the children from a large number of marriages between straight-haired and wavy-haired persons, Cockayne found sixty-one straight to fifty-two wavy, a close approximation to the expected 1:1 ratio (see pages 66–67). When both parents had wavy hair he found straight, wavy, and curly in the ratio of 27:51:22, a close approximation to the expected 1:2:1 ratio (see pages 66–67). The kinky hair of the Negro race generally dominates over the hair form of the white race, although there may be some degree of intermediate expression.

Presence or absence of hair

Next to color and form, the most striking feature of hair concerns its presence or absence. **Baldness** is a characteristic which, without doubt, can be induced by environmental agents, such as disease, but the majority of people who are bald become so because of their genes. The condition is inherited as a result of a sex-influenced gene which is dominant in men and recessive in women. The pattern of baldness which develops and the age at which it begins is also influenced by heredity. There is another condition, hairlessness (**hypotrichosis**), which is characterized by the complete absence of hair on the head from birth or shortly thereafter and a scanty growth of hair on other parts of the body. Such a condition is inherited as an autosomal recessive (see pages 61–62) which seems similar in its effects to genes for hairlessness in domestic mammals, such as cats. A dominant form of hairlessness has also been reported from some human races including Australian natives. The hairlessness of the Mexican hairless dog is also due to a dominant gene.

The opposite condition, excessive amounts of hair on the body (**hypertrichosis**), may be produced by a dominant gene in man. Also, other genes may produce excessive amounts of hair only on certain body parts. Heavy,

bushy eyebrows result from a dominant autosomal gene. Hypertrichosis of the ears is a trait induced by a gene on the non-homologous portion of the Y-chromosome. Hence it appears in men only and passes directly from father to son. The amount of hair on the face and body is variable among the different races. The Ainus of northern Japan are the hairiest people on earth, the white race comes next, followed by the Negroes, while the American Indians, Mongoloids, and Eskimos have little hair on the face and body. The intermediate condition of race hybrids suggest multiple gene inheritance for these differences.

The growth of hair on the middle joint of the fingers is an interesting human characteristic which may easily be studied by amateur geneticists. **Middigital hair** may occur on fingers in various combinations. It is most common on the ring finger; next comes the ring finger plus the middle finger; next comes the ring finger, plus the middle finger, plus the little finger; and least common is hair on all four fingers. A multiple allele hypothesis has been proposed to account for the inheritance of these combinations. This hypothesis assumes the presence of hair on all four fingers as dominant to its presence on three; on three, in turn, being dominant to its presence on two; two dominant to one; and one dominant to none. In making such a study, a hand lens should be used to bring out small hairs that might not be easily visible with the naked eye.

Arrangement of hair on the head

Near the top part of the back of the head there is a crown or **whorl of hair** which rotates in a clockwise direction in most people. In a few cases, however, the whorl may be counterclockwise as a result of a recessive gene. Occasionally, a person will have two whorls rather than one. This seems to be inherited as a recessive character also. At the center of the forehead the hairline may dip down to form a point which is called a **"widow's peak"** (see Fig. 208). This characteristic is inherited as a dominant character in some families.

Eyes

Color

The color of the iris of the eye is a clearly defined human character which draws much interest. Indeed, it was one of the first human characters which was suggested as an example of Mendelian inheritance in man. Davenport, in 1907, studied a number of family pedigrees and suggested that brown eyes are dominant over blue eyes in the white race. There are many variations of eye color, however, which may be influenced by other genes. The blue color of the iris is due to a pigment in the back (or retinal layer) of the iris as seen through a semiopaque, colorless layer in the front of the iris. This blue layer is present in most persons, but in those with darker eyes, it will be masked by the development of melanin in the front part of the iris. Nearly all babies of the Caucasian race are born with blue eyes, which may darken later as the melanin pigment develops in the front layer of the iris. Gray eyes seem to be a variation of the blue. Green and hazel eyes appear when the melanin partially masks the reflection from the rear of the iris. Brown and "black" (dark brown) represent an almost complete masking. There is also variation in the distribution of the melanin — it may be smoothly distributed, it may be in spots, or it may occur in a ring around the outer edge of the iris.

In most families where both parents have pure blue or gray eyes, the children all have blue or gray eyes, thus indicating the recessive nature of this character. The darker colors generally are dominant over the blue or gray, but there is considerable variation in the expression of the darker colors because of modifying genes. Of course, when the recessive gene for albinism is homozygous there is no formation of melanin. In some albinos the iris is pink because the blood in the retinal layer becomes visible. In other albinos there

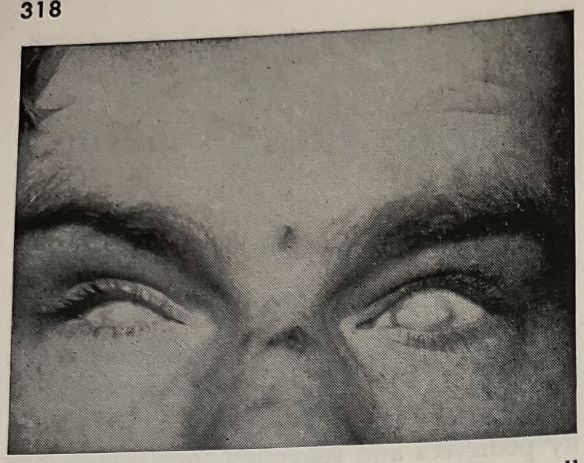


Fig. 209 *Microphthalmia*, the possession of small non-functional eyes, was inherited as a sex-linked recessive in four generations of a family studied by F. E. Stephens in Utah.

are some reflecting bodies in the retinal layer and the eyes will be a pale blue.

Vision

Nearsightedness, **myopia,** is a very prevalent hereditary defect of the eyes. It may be brought about by either one of two independent factors. When the eyeball is too long, a normal lens will bring distant objects to a focus in front of the retina, thus causing a fuzziness of the image upon the retina. Such a condition seems to be inherited as an autosomal recessive. An excessive curvature of the cornea is a less common cause of nearsightedness, and seems to be caused by a dominant gene. There is also a sex-linked form of myopia which has a reduced penetrance (see pages 265–266).

Farsightedness, **hyperopia,** results when the eyeball is too short for the curvature of the lens. In a normal eye, when the muscles within the eye are relaxed, the image from distant objects is in focus on the retina. When the eyeball is too short, however, the point of focus is behind the retina. Hence, there must be a constant strain on the eye muscles to accommodate for normal vision, and it is sometimes impossible to bring nearby objects into sharp focus at all. This condition seems to be a simple dominant character.

Astigmatism is a defect of vision caused by unequal curvature of the cornea, which causes objects in one plane to be in sharper focus than objects in another plane. It also seems to be inherited as a dominant.

Blindness is, of course, the most serious of eye defects. It may be induced by many environmental causes and it may be inherited in many different forms. Keeler lists over twenty different hereditary causes of blindness. One of the most common of these is **glaucoma,** which is responsible for more hereditary blindness than any other cause. In glaucoma the fluid within the chambers of the eye develops a great internal pressure which results in destruction of the optic nerve. This is a case in which preventive surgery may be used to inhibit the development of an inherited defect. An eye operation may provide a channel for a better circulation of the humors within the eye and thus relieve the undue pressure on the optic nerve. The character is dominant, but usually does not make its appearance until middle age, although in some families it may appear as early as fifteen years of age. **Cataract** is another eye defect which leads to blindness. The condition is characterized by the development of an opaque condition of the lens or the cornea. We know that there are environmental agents which may lead to the production of cataract, but it is undoubtedly influenced by heredity also. The tendency to develop cataract is inherited as a dominant, and the age at which it is likely to develop is also inherited. In some families it is even present at birth. Another inherited cause of blindness is **retinitis pigmentosa**, which is characterized by the development of a pigment alongside the blood vessels of the retina. The condition is found to result from a dominant gene in some families and from a recessive in others. Both these genes seem to be incompletely sex-linked. **Optic atrophy,** or degeneration of the optic nerve, which also results in blindness, is due to a recessive sex-linked gene. **Micropthalmia,** illustrated in Figure 209, is an inherited condition characterized by the presence of very small, non-

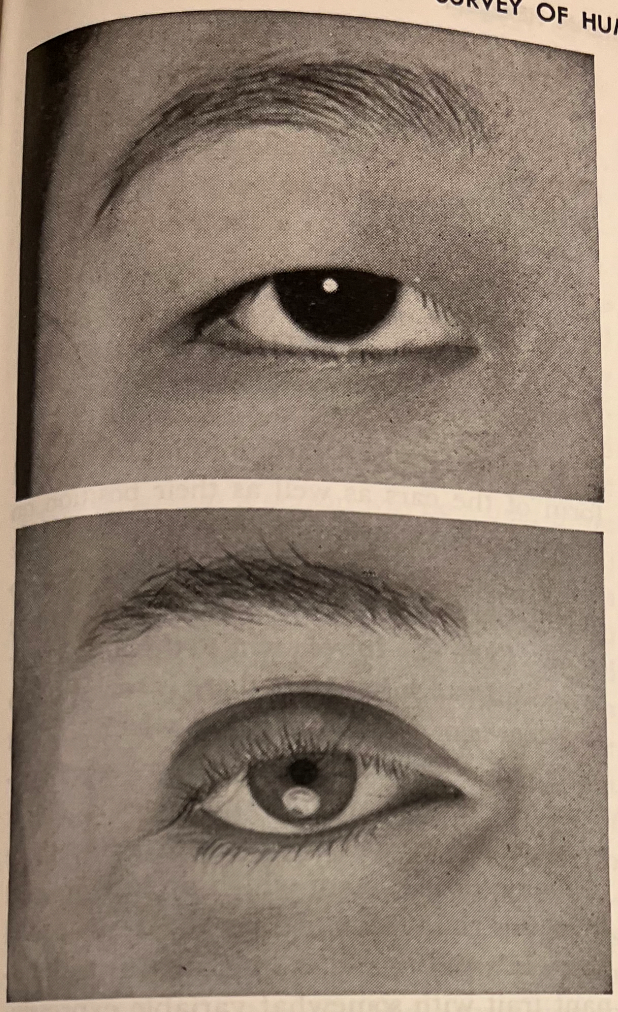


Fig. 210 The Mongolian eye-fold (above), compared with a typical Caucasian eye (below). Note that the upper eyelid is folded downward, extending over the point of origin of the eyelashes.

functional eyes. It seems to be due to a recessive, sex-linked gene.

Red-green color blindness is inherited as a sex-linked recessive as we learned in Chapter 9 (page 117). Detailed studies of this condition, however, show that there are different variations of this type of color blindness. Two gene loci very close together on the chromosome seem to be involved, and there seem to be three multiple alleles for each of the two loci. The normal allele, of course, is dominant over the other two in each case, and the gene for a less extreme form of color-blindness is dominant over the gene for a more extreme form in each case. It appears that the dominance is not complete, however, for it has been found that some women carrying one of the color-blind genes at one of the two loci, but with a normal allele, have poor color aptitude. In a study made by Waaler in Oslo, Norway, it was found that of about 9,000 boys, about 6 per cent showed one of the four forms of red-green color blindness.

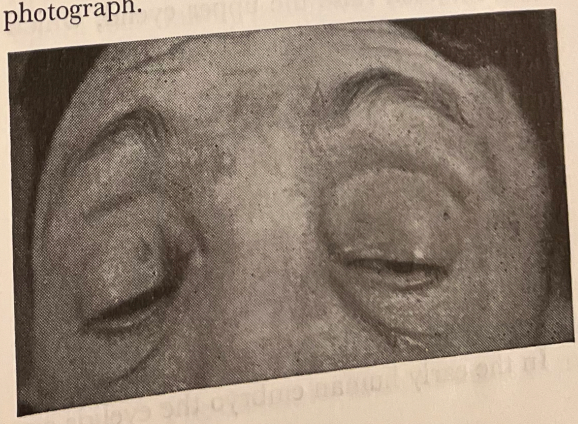
There is also a **total color blindness,** which results from the action of a recessive gene located on the homologous portions of the *X*- and *Y*-chromosomes. Another sex-linked recessive gene results in an impairment of vision known as **congenital night blindness.** Persons showing this character see very well in good light, but very poorly in dim light. Night-blindness may also result from a deficiency of vitamin A in the diet, but feeding vitamin A in large amounts to a person who is congenitally night-blind brings no relief from this condition.

In some persons vision is impaired by an uncontrolled movement of the eyes, known as **nystagmus.** The eyes may undergo up and down, lateral, or rolling movements. In some families this character is inherited as a sex-linked recessive, in others as an autosomal dominant.

Eyelids

The eyes of the specialized Mongoloids (Chinese, Japanese, and so on) are commonly referred to as "slant-eyes." Actually the position of the eyes is the same as in the other races, but there is a fold of skin from the upper eyelid which extends down over the inner corner of the eye to produce this distinctive

Fig. 211 *Ptosis*, or drooping eyelids. This man can open his eyes only as far as shown in this photograph.



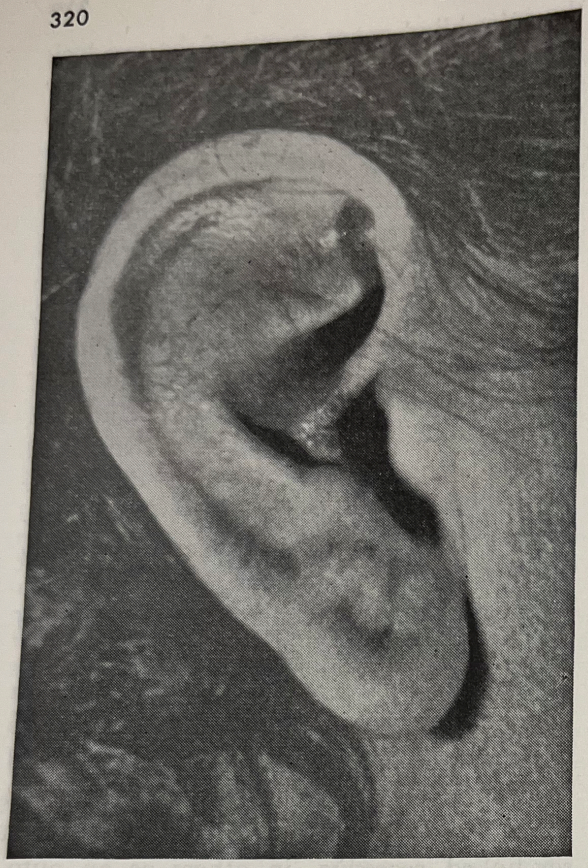


Fig. 212 Natural "earring holes." Note the pit on the ear at the exact point where ladies once punctured their ears for earrings. This is due to an autosomal dominant with incomplete penetrance and variable expressivity. It appears on only one ear in some persons who express the character.

appearance (see Fig. 210). Interracial marriages indicate that this character is inherited as a simple dominant. Another inherited defect of the upper eyelid is called **ptosis,** or drooping eyelids (see Fig. 211). Persons with this condition are unable to use the muscles which raise the upper eyelid, which therefore remains drooped down over the eye and leaves only a small slit between the upper and lower lid for vision. There are various degrees of expressivity of this gene. In some persons the drooping is sufficient only to give a sleepy appearance, but in others it is so pronounced that the head must be thrown back for them to see straight ahead. This condition appears generally to be a dominant.

In the early human embryo the eyelids are fused, but normally they separate at about the seventh month of development. There is an inherited condition called **cryptophthalmos** in which the lids fail to separate and the baby is born unable to open its eyes. This appears to be due to a recessive autosomal gene.

Ears

Form

Many continuous variations in the size and form of the ears as well as their position on the head indicate multiple gene inheritance of ear characteristics. A few of these seem to correspond primarily to variations in a single gene. **Free ear lobes** seem to dominate over **attached ear lobes,** but there is variation in the degree of freedom of those which are not attached. The outer, rolled rim varies continuously in its size, and sometimes is almost lacking. In some persons, moreover, there is a very distinct point which projects inward from this rim. As noted earlier this is called **Darwin's point** and is inherited as a dominant trait with somewhat variable expressivity, since it shows in only one ear in many persons. There is also some reduced penetrance. Nearly all persons have a small enlargement of the cartilage at this point, but it usually does not stand out enough to show distinctly. Keeler reports an interesting pedigree showing inheritance of natural **"earring holes"** (see Fig. 212). In the center of the lobe of the ear of an affected person there is a distinct depression in exactly the same spot where the young ladies of times past pierced their ears for earrings. This condition is inherited as an autosomal dominant with somewhat incomplete penetrance and variable expressivity. About half the people showing the character express it on only one side.

Hearing

The outer portions of the ear are the more noticeable, but the important functional parts are embedded in the bone within the

skull. **Deafness** is, of course, the most serious defect of the ears. Since hearing involves the correct coordination of many parts of the ear, deafness may result from a number of causes. Various types of ear infections and other environmental agents may induce deafness. One inherited form of deafness, however, is present at birth. Since children so afflicted cannot hear the spoken word, they normally do not learn to speak and become deaf-mutes. At least two genes are known which produce such deafness, and both of them are recessive, so that a person will be a deaf-mute if he is homozygous for either of the two genes. Some special training institutions have been developed to teach such persons to speak and to read lips. The author once held a rather lengthy conversation with a person who had been so trained, only to learn later that the person had heard not a word that was said. Usually, however, the speech of persons born deaf is noticeably different from that of persons with normal hearing. Another inherited form of deafness makes its appearance after maturity, beginning with a slightly defective hearing and progressing to total or near total deafness in old age. This is known as **otosclerosis** and is inherited as a dominant. It is usually accompanied by a ringing, or buzzing, in the ears. It is induced by an abnormal growth of bone around the bones of the middle ear which transmit sound vibrations and make hearing possible. This type of deafness is usually first noticed at about thirty years of age. Still another type of deafness results from atrophy of the auditory nerve, which transmits the impulses from the hearing organs of the ears to the brain. This usually begins to manifest itself at about forty years of age and progresses for several years, finally resulting in complete deafness. It seems to be inherited as an autosomal dominant.

Nose

We often hear the expression, "As plain as the nose on your face," which emphasizes the fact that the nose is one of the most conspicuous of facial features. As with so many other human characteristics, we find continuous variations of many sorts in the length, width, and shape of the tip of the nose which result from multiple genes. A number of the characteristics of the nose which show simpler inheritance may be mentioned. A high, convex bridge of the nose seems to be dominant over a straight bridge or a concave bridge. The root of the nose, the point at which it joins the forehead, in some persons is quite high, forming a suitable attachment for eyeglasses; while in others it is low. There is variability in this character, but high root seems to dominate low root. A straight tip of the nose seems to be dominant over an upturned tip. The wings which spread out from the point of the nose are also quite variable, and may be wide and spreading or narrow. The wide spread wings seem to show dominance over the more narrow wings. Also, some wings may be higher than the septum of the nose, so that the openings of the nostrils are clearly visible from a side view. In other cases the wings are level with or lower than the septum which divides the nostrils. Some geneticists think that the high wings are recessive to the lower types.

Mouth

Lips

The lips form the gateway to the mouth and are a very prominent part of a person's facial expression. Numerous multiple genes determine the general shape and size of the lips. Full lips seem to be dominant over thin lips in some family pedigrees. **Harelip** is one of the most common abnormalities of the lips. When the upper lip is first formed in early embryogenesis it consists of three portions, two lateral and one median. During the second month of embryonic life these processes normally fuse to form the upper lip. On some occasions, however, they fail to fuse, most frequently on the left side, and a baby is born with a distinct cleft extending up toward the nose. The deformity takes its name from

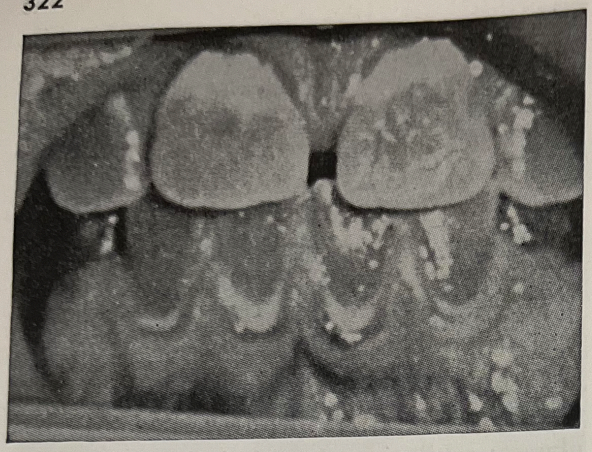


Fig. 213 Opalescent dentine. The dentine is soft and the enamel is bluish. The teeth wear down easily. A dominant gene seems to be responsible for this condition.

the mouth of the hare, but the cleft in the animal's upper lip is median, while that in a human lip lies to one side or the other of the median line. Harelip is often accompanied by a cleft palate in the roof of the mouth and connecting the mouth cavity with the nasal cavity, and it is more common in men than in women. The evidence indicates that the condition is inherited, although there seems to be some variation in penetrance. Some authorities consider it to be due to a recessive gene which has greater penetrance in the male than in the female. Surgery performed early in life may correct harelip and cleft palate so that the condition is not evident.

Teeth

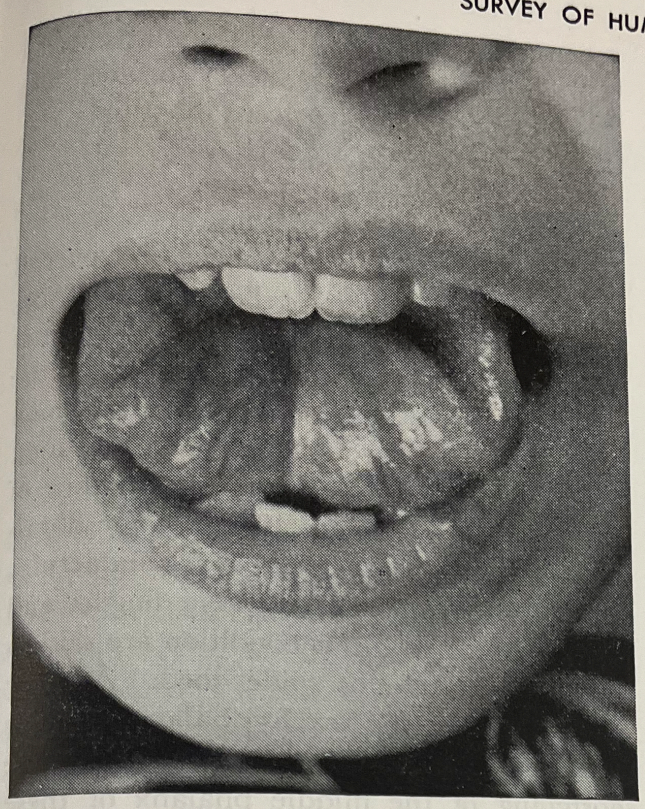
There are many genes which influence the development and conformation of the teeth. Variations in single genes may produce pronounced effects. One dominant gene causes the absence of some of the upper incisors and molars. Another dominant gene causes the absence or reduction of the two lateral incisors in the upper jaw. A sex-linked recessive gene causes the absence of the canines and perhaps some other surrounding teeth. A dominant sex-linked gene causes the absence of enamel on the teeth. This causes them to wear down rapidly so that they often barely protrude through the gums. Still another dominant gene causes defective dentine, so that the dentine is soft and opalescent, the enamel has a bluish color, and the teeth wear down easily (see Fig. 213). Brown enamel of the teeth results from a recessive sex-linked gene. Tooth decay is without doubt influenced by environmental factors such as diet, but the susceptibility of the teeth to decay seems to be due to a dominant gene.

Tongue

As with most other body characteristics the shape and size of the tongue responds to so many different genes that it is not possible to isolate many effects of individual genes. Some people have the distinctive ability to roll their tongue into a U-shape when they extend it from the mouth (see Fig. 214). This ability seems to depend upon a dominant gene. The sense of **taste** is also associated with the tongue. It has been discovered that people vary in their ability to taste a certain chemical, phenylthio-carbamide. To some this chemical has a distinctly bitter taste, to others it is quite tasteless. This is an easily measured human characteristic which has been extensively investigated. The ability to taste this substance is dominant, the inability recessive. About 70 per cent of the American people are tasters.

Chin and Cheeks

The chin is often thought to denote some aspects of a person's character — the pugnacious, aggressive, protruding chin and the weak, timid, receding chin — but the genes which affect this portion of the human anatomy seem to have no direct relationship to inherited traits of personality. The receding chin seems to be inherited as a recessive trait. The chin also varies in its length, from the lower lip to its lowest point, with multiple genes making the variation continuous. Some persons have a distinct depression, or dimple, in the lower part of the chin, as illustrated in Figure 215. This seems to be inherited as a dominant. One pedigree showed



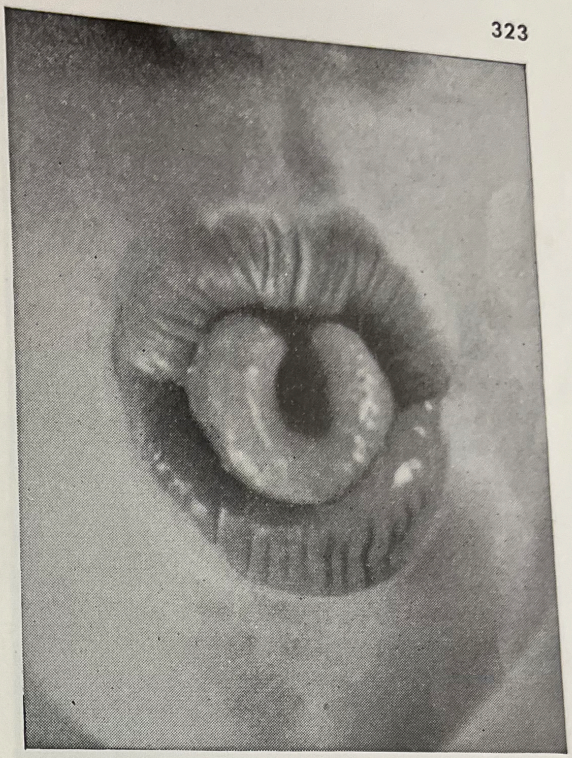


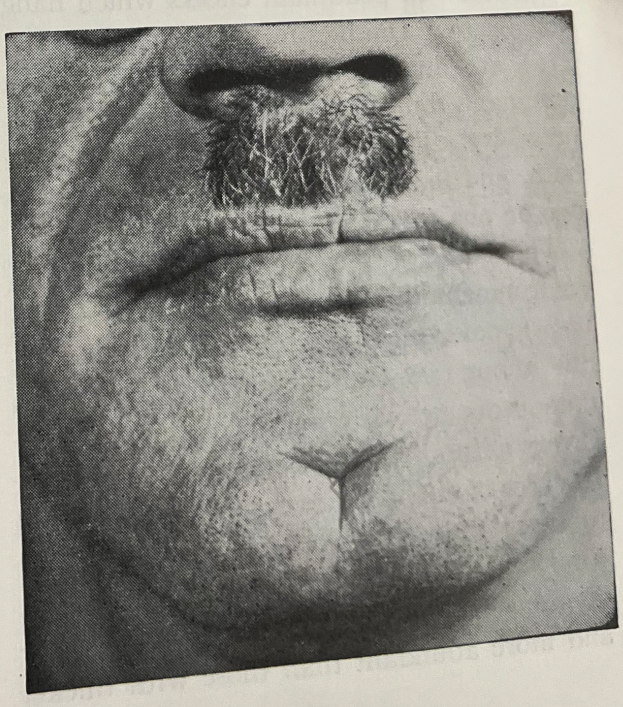
Fig. 214 Two forms of tongue-rolling. The ability to fold the tip of the tongue backward is found in less than one person in a thousand, but the majority of people can roll the tongue in a U-shape. A dominant gene seems to be involved in each case.

this character in an extreme form; there was actually a hole in the chin bone at this point. It too was found to be a dominant trait.

From a front view, the chin may be square or rounded, wide or narrow, with variations between. A study of the Hapsburg dynasty of Europe shows that for six centuries there was a rather consistent transmission in the royal family of a narrow, protruding chin, together with an overhanging lower lip. Dominance of this trait certainly seems to be indicated. A double chin, however, clearly reveals the mutual influence of heredity and environment — inheritance of the necessary genes plus excessive intake of fattening foods.

The cheek bones may be high, low, or intermediate. Many races, such as the American Indians, are characterized by high cheek bones. The gene complex for high cheek bones seems to be dominant. There is quantitative variation in the extent of the fat pads of the cheeks, both as to thickness and placement. In some persons the pads are so low

Fig. 215 Cleft chin. There is a hole in the bone of the chin of this man just at the point where the cleft occurs. The character is inherited as a dominant.



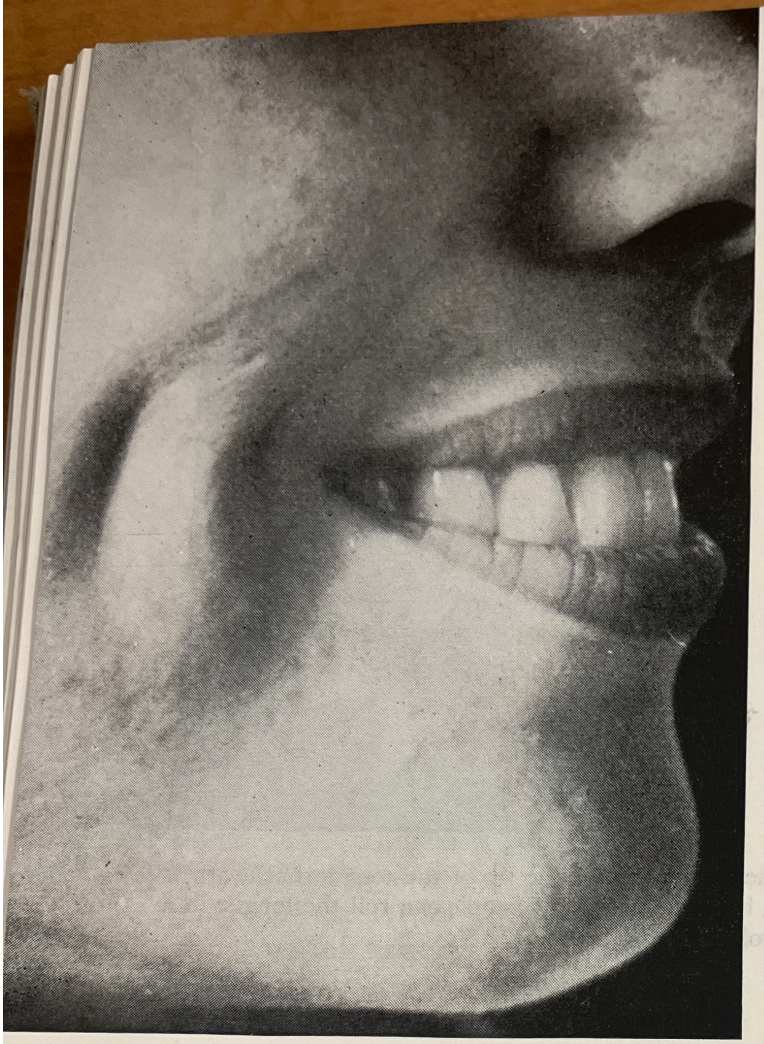


Fig. 216 Dimpled cheeks seem to be inherited as a dominant, although there is variable expressivity.

as to result in pendulant cheeks which hang downward. Dimples in the cheeks (see Fig. 216) are inherited also, apparently as a dominant, but with some variation in expressivity. They may occur on one cheek or both, and in rather rare cases there may be two on one cheek.

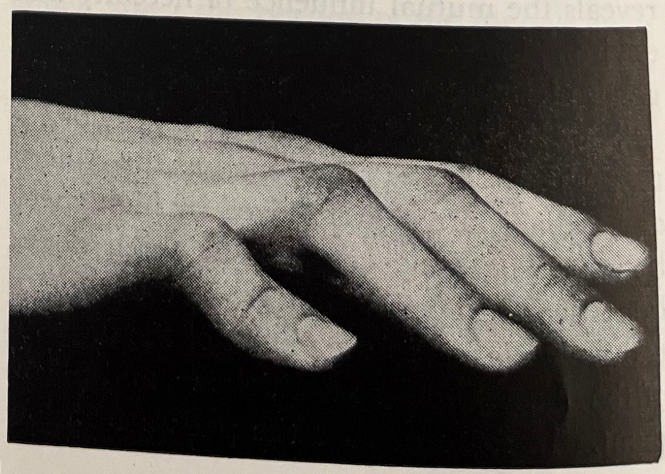
The color of the cheeks is certainly an environmentally induced characteristic among the female members of our population, but among men and in children of both sexes there is distinct variation in coloration. Rosy cheeks naturally accompany a thin skin over this area of the body, for that allows the blood to show through. Popular opinion to the contrary, this does not mean that such individuals have a blood supply which is richer and more abundant than those with thicker skin and less rosy cheeks. Darwin reported a family in which the tendency to blush and the manner of blushing were inherited.

Hands and Feet

Length of fingers and toes

There are genes which produce overall variations in the length of the fingers and toes relative to the hand or foot, and other genes which influence the relative lengths of the individual fingers and toes. Most genes which influence the fingers also influence the toes, but there are a few which affect only one of the two types of digits. There is one dominant gene for a trait, called **arachnodactyly,** which produces extremely long fingers and toes. Persons with this condition are said to be spider-fingered or spider-toed. Another dominant gene, for **brachyphalangy,** causes very short fingers, because of an extreme shortening of the middle phalanx of these digits. On the basis of a study of the children of one couple both of whom had the condition, this gene is believed to be lethal when homozygous. Another dominant gene causes a shortening of the fingers and toes through the apparent absence of the middle joint. This is known as **brachydactyly.** X-ray photographs show that this trait involves a fusion of a rudimentary middle joint with one of the other joints of the digits. Still another dominant gene, **symphalangy,** causes a fusion of some of the joints without any shortening. This produces stiff fingers. An inter-

Fig. 217 Camptodactyly, flexed fingers which can be extended no further than shown here.



esting sex-influenced character concerns the relative length of the middle finger in relation to the other fingers of the hand. A long middle finger seems to be inherited as a dominant in men and as a recessive in women. (See Chapter 9, pages 125–126, for a more detailed description of this trait.) A **second** toe longer than the big toe also is inherited, but no pedigrees seem to be available to indicate the type of inheritance.

Position of fingers and toes

Some persons are unable to straighten out their fingers because of an abnormal shortening of the flexor tendons (see Fig. 217). Possession of such flexed fingers, known as **camptodactyly,** is inherited as a dominant with reduced penetrance. The possession of crooked little fingers is a recessive trait. Hypermobility of thumb joints (see Fig. 218) is another characteristic seemingly inherited as a dominant.



Fig. 219 Mono-digital mutation of the hands and feet of a family in India. The mother is shown at the right, with her two daughters and two granddaughters. The child in the center has normal hands and feet. The character is transmitted as a dominant, but these people think it appears because of a lack of religious zeal on the part of the prospective mother.

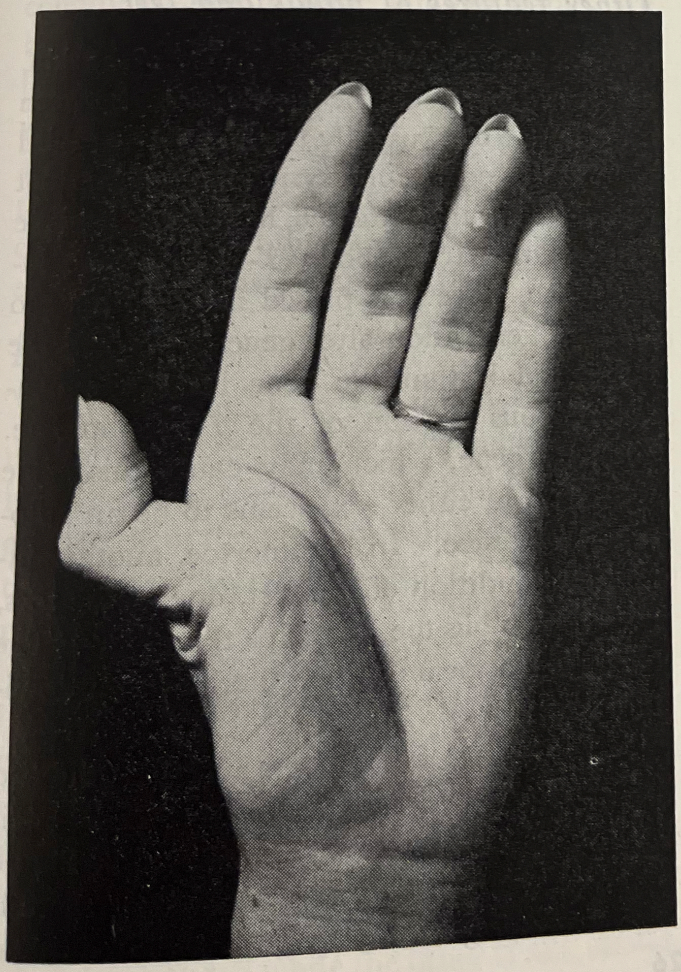


Fig. 218 Hypermobility of the joints of the thumb. This extreme mobility seems to be inherited as a dominant.

Odd numbers of fingers and toes

Polydactyly, or the presence of extra fingers and toes, was one of the first inherited traits to be traced in a human pedigree (see Fig. 8, page 13), and we have a very complete pedigree for this trait, collected by Maupertuis in the first half of the eighteenth century. The extra digit may be appended to the little finger or toe, or it may be attached to the thumb or the big toe. The condition is inherited as a dominant with variable expressivity. Some persons show it in their hands, but not on their feet, and vice versa. Another dominant gene results in a reduction of the total number of fingers. This seems to be due to a fusion of digits and also varies in its expressivity. The most extreme expression results in **"lobster claw,"** a condition in which there are only two digits. Other persons

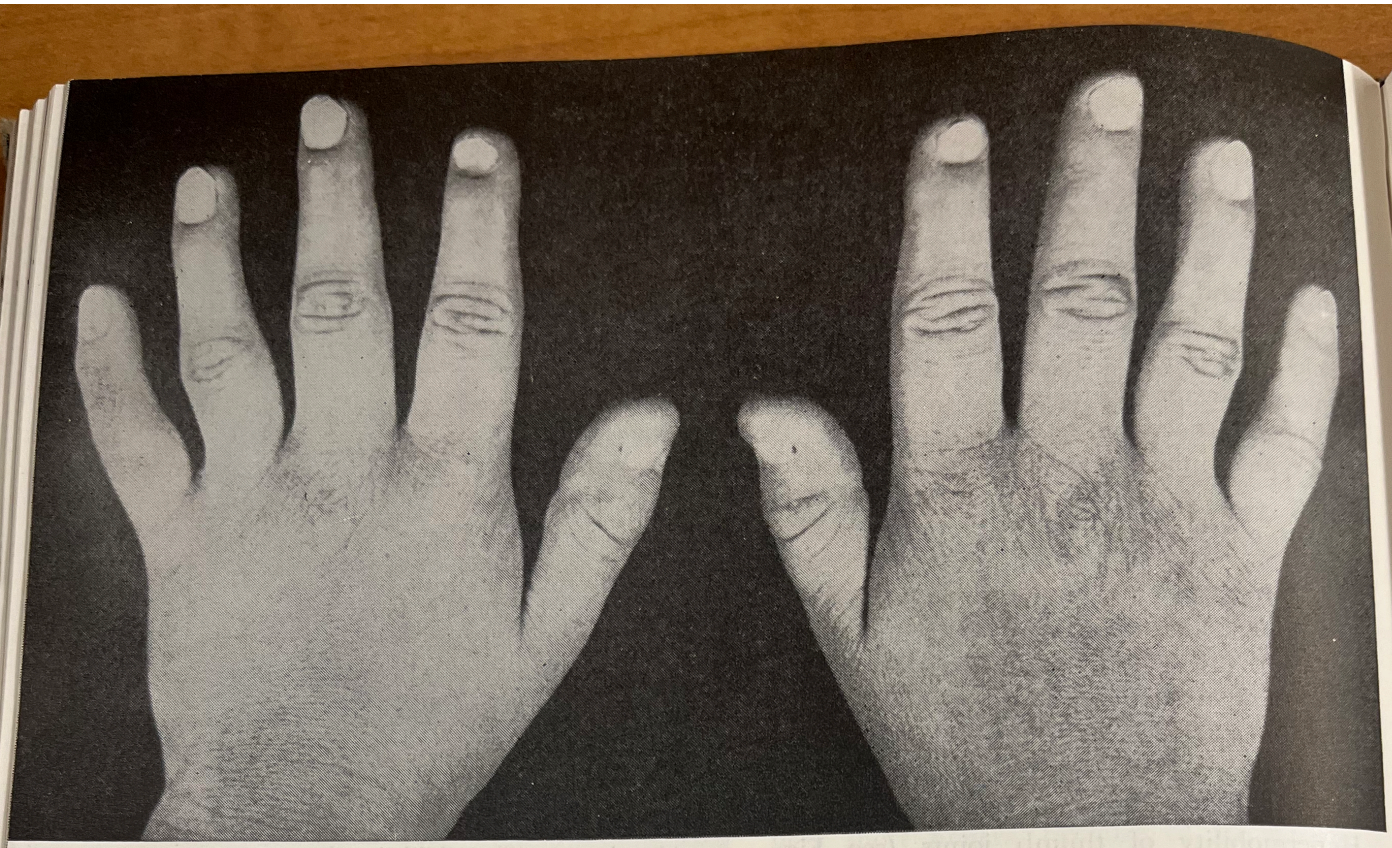


Fig. 220 Absence of thumb nails. Although there is a depression in the region of the thumb nail, there is no nail of any kind. The trait is probably inherited as an autosomal dominant.

carrying this gene may have three or four digits, and in a few cases it may be expressed only in the deformity or the absence of a nail of a single finger. This gene causes such an extreme physical handicap that it should be considered dysgenic for an afflicted person to have children. A gene mutation causing monodigital hands and feet is illustrated in Figure 219.

Nails

Extremely short nails, **brachymegalodactyly,** seem to be inherited as a dominant. The complete absence of a thumb nail also seems to be inherited as a dominant (see Fig. 220). In some families there may be an excessive growth of the nail-forming tissue to give very thick nails, **hyperkeratosis subungualis,** another trait which is inherited as a dominant. Again, when viewed from the side nails may show a convex curvature or may be straight. The curved condition seems to be dominant over the straight.

Other features of hands and feet

In some families there is a fusion of the skin, and sometimes of the bone also, between some of the toes, usually the second and third toe. This condition, **syndactyly,** does not extend to the tip of the toe, however. In some families this trait shows Y-chromosome inheritance, since it is passed from a father to his sons generation after generation, and the indication is that the gene is on the nonhomologous portion of the Y-chromosome. Another gene, which may cause this same condition in either sex, seems to be an autosomal recessive. In hammertoe, another inherited condition of the feet, one of the toes, usually the big toe, overlaps the others. The condition appears to be inherited as a dominant.

Swollen tips of the fingers and toes (clubbed fingers and toes) is another inherited condition, an example of which is shown in Figure 221. X-ray photographs show that there is a broadening of the bones of the terminal

phalanges in this condition. Similar conditions arise as a result of pulmonary or heart trouble.

Clubfoot results in serious deformity if corrective measures are not taken early in life. The most common type causes an inward turning of the foot so that the person must walk on the outer side of the foot. This is due to a shortening of the muscles or the tendons attached to the muscles on the inner side of the leg. Other types of club-foot cause the afflicted person to walk on the front part of the foot, on the heel, or on the inner edge of the foot. Abnormal pressure on the embryonic foot will cause clubfootedness, but the frequency of the condition in some family pedigrees shows that it may result from heredity also. In some families it is inherited as a recessive, in others as a dominant with reduced penetrance. About one baby out of every 1300 is born clubfooted. Some authorities estimate that over half of these cases are due to heredity.

Flat feet may result from environmental conditions, but in some families babies are born with flat feet, so that obviously the defect cannot be due to undue pressure during walking or standing. Since man is the only animal that has a definite arch to the foot, the flat foot represents a condition which is closer to that of other animals. It is apparently inherited as a recessive trait.

Handedness

The hand which we use the most is determined not by the greater development of the hand itself, but by the functional dominance of one side of the brain over the other. Since the nerve pathways cross in the brain-stem, a person who is right-handed would be left-brained, in the sense that the left side of his brain would dominate the right. Just how completely heredity influences handedness is not clear, but the evidence indicates that right-handedness may be due to a dominant gene and left-handedness to its recessive allele. The fact that in many pairs of monozygotic twins one is left-handed while the other is not, shows that some other factors may also be involved. However, in twins there is a tendency for mirror-imaging, so that the right side of one person will be very much like the left side of his twin. The common occurrence of left-handedness in one member of a pair of monozygotic twins is therefore usually ascribed to this influence, but cases of left-handedness in other individuals may, in part at least, result from a pair of recessive genes.

The Skin

Color

Skin color, like so many other human characteristics, is inherited through multiple genes working in conjunction with environmental agents such as sunlight. The differences in pigmentation which distinguish the white race and the Negro, however, seem to rest upon two pairs of genes, both of which are intermediate in their expression. The typical mulatto condition, with an intermediate shade of skin, results in the children of a marriage between members of the two races, though of course there is some degree of variation in the mulatto coloration due to various modifying genes which may be present in either of the two parent races. Marriages between two mulattoes produce variation from the full white to the full Negro pigmentation in the rather unusual ratio of 1:4:6:4:1.

Fig. 221 Clubbed fingers. This condition may arise as a result of certain cardiac diseases, but it may also arise from inheritance without any cardiac disease, as in the case illustrated below.

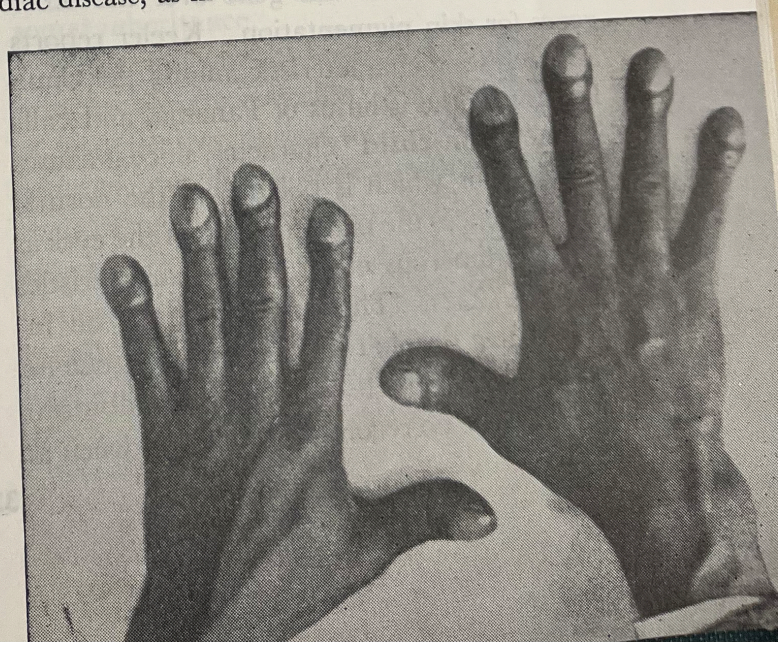




Fig. 1 Musical aptitude is influenced by heredity. This young lady is an accomplished pianist; she has inherited great musical ability and has had good training to bring out her musical potentialities.

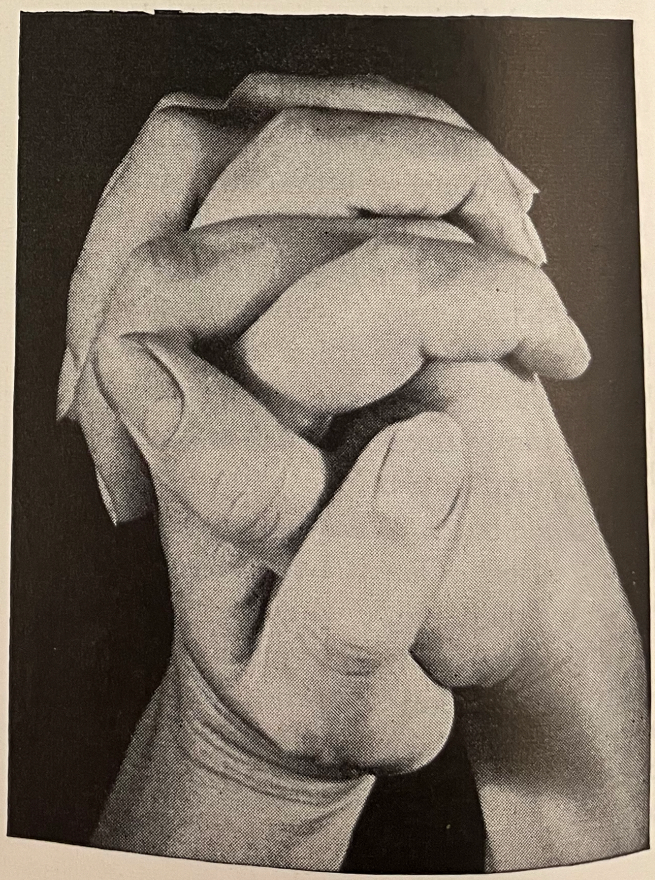
print upon our being as long as we live.

The principles of heredity apply not only to the more readily distinguished physical characteristics, but also to the less noticeable characteristics of the nervous and glandular systems which influence the more intangible qualities such as intelligence, aptitudes, temperament, and even the minor body mannerisms which contribute to one's total personality. A fond mother may be determined to make a musical genius out of her daughter, yet no amount of study and practice can produce an outstanding musician unless the child has inherited an aptitude for music as the young lady shown in Figure 1. One would hardly think that heredity had anything to do with the way a person folds his hands, yet the placement of the fingers in this simple gesture (Fig. 2) can be traced through heredity just as the shape of the nose or the height of the cheek bones can be. A group of students with similar backgrounds may receive the same amount of training in a course of mathematics, yet there will be some in the group who will have the greatest difficulty in comprehending the subject while others grasp the same material with comparative ease. Again, heredity plays an important part in such variations in aptitude. In the course of this book we shall learn a good deal about why all this is true.

Practical Application of Genetics

Moreover, a knowledge of heredity has a wide variety of practical uses. To the **animal breeder** or the **horticulturist,** who is concerned with the improvement of domesticated animals or cultivated plants, the science of heredity is of utmost importance. The fat, beef-producing cattle that roam our western plains today are a far cry from the scrawny

Fig. 2 Heredity influences such minor body mannerisms as the method of folding the hands. The young lady who owns this pair of hands has inherited the tendency to place the left thumb on top of the right.



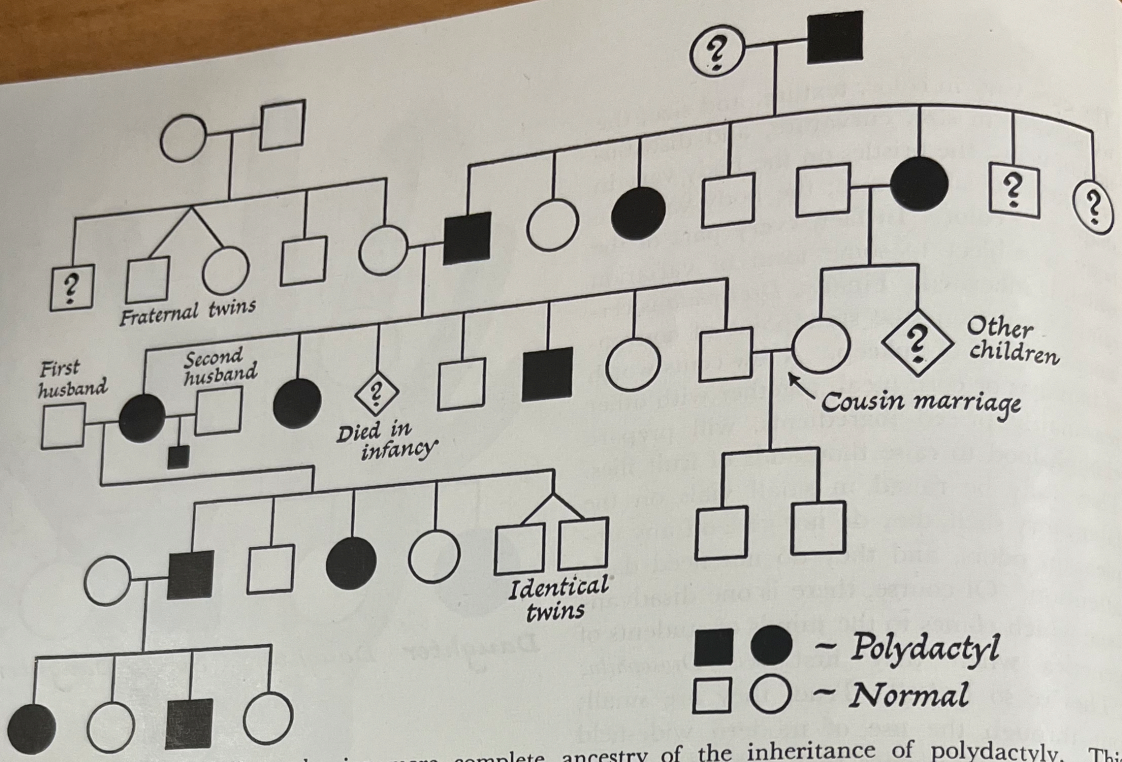
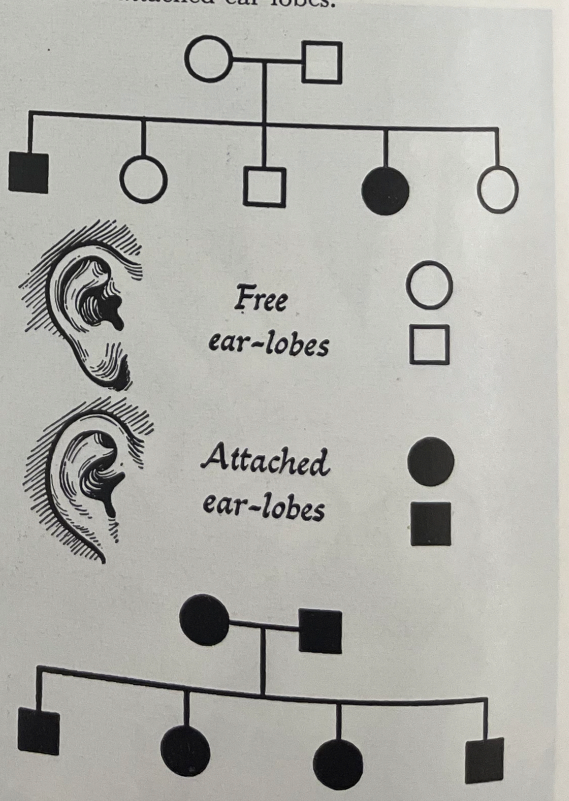


Fig. 9 Pedigree showing more complete ancestry of the inheritance of polydactyly. This chart also indicates the method used in preparing such family histories. Squares represent males; circles, females.

curate information. In the case of man, we now clearly understand the mechanism of inheritance for hundreds of characteristics which have been studied by this method.

In order to conduct such a study, however, it is necessary that a standard method be used to represent the family pedigrees in a concise, easily understood form. A case of a condition known as *polydactyly* (Fig. 7) gave rise to the form of pedigree illustrated in Figure 8, which has become standard usage among geneticists. This pedigree was discovered when a woman brought her young daughter to a doctor for examination. The girl had an extra finger on one hand and an extra toe on one foot. This relatively uncommon condition is known to be inherited. Upon further investigation it was found that the child's father showed this character (though the extra finger had been removed surgically), and that her brother also showed the condition, whereas two other children in the family had the normal number of fingers and toes. One can visualize this entire family history by a glance at the chart in Fig. 9. In charts of this kind men are customarily represented by squares and women by circles. Marriage is indicated by a connecting horizontal line and children are shown by attachment to a vertical line extending downward from the horizontal one.

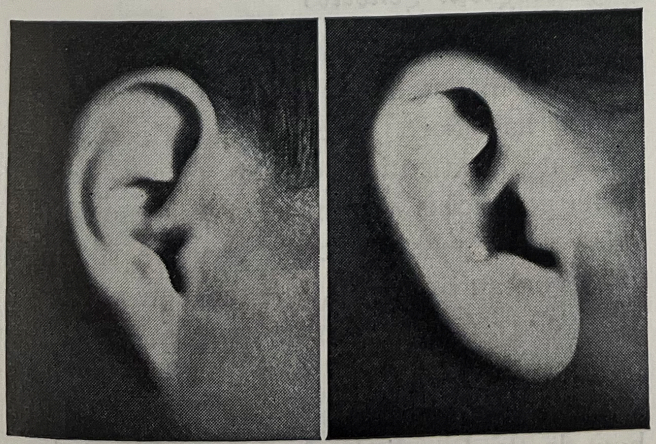
Fig. 10 Pedigree of two families showing inheritance of attached ear lobes.



Those individuals which show the particular character which is being traced are represented by a solid square or circle while those not showing it are indicated in outline.

From such a chart it is easy to obtain a statistical ratio of the appearance of this trait in an attempt to analyze the method by which it is inherited, but the numbers are far too small to have much significance statistically. We may obtain larger numbers through a study of the ancestors of this family and can construct a more extensive family history chart for this purpose. We now begin to have sufficient numbers to have some statistical significance. As we survey the chart (Fig. 9) we note that no person shows the trait unless at least one parent shows it. This gives us one clue to the method of inheritance. Among the children of marriages where one parent showed the trait there are 11 children who are normal and 10 who are polydactyl. This is a ratio of about 1:1 (that is, $\frac{1}{2}$ show it and $\frac{1}{2}$ do not). In the group which shows the trait are four boys and six girls, so there seems to be no influence of sex on the development of the character. After other family histories showing this character have been studied, we may determine the method of inheritance through comparison with results obtained on lower animals through experimental breeding of various characters. It may then become possible to predict the chances of polydactyly appearing in the future children of this family.

Fig. 11 The ear lobes of some individuals are attached directly to the head (left), while in others the lobes hang free (right).



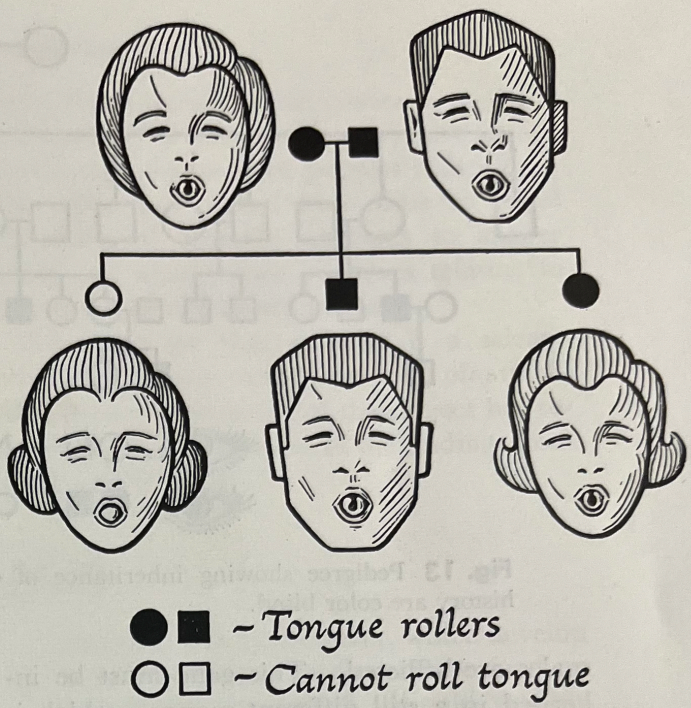


Fig. 12 Pedigree showing inheritance of ability to roll the tongue.

Two other family pedigrees are shown in Fig. 10 for **attached ear lobes.** (Fig. 11 shows the physical appearance of attached and free ear lobes.) From the chart it appears that this character must be inherited in a different manner, for two parents with free ear lobes produce two children with attached ear lobes in a family of five children. And in another family, both parents have attached ear lobes and so have all four of their children. This type of inheritance is typical of characters which are known as **recessive**.

Some persons have the power to roll the tongue, while others are not so gifted. A couple, both of whom are **tongue rollers,** have three children, two of whom can roll their tongues and one who can not, as shown in the family pedigree, Fig. 12. This seems to be a different method of inheritance than that for attached ear lobes.

Finally, the pedigree in Fig. 13 shows the inheritance of **red-green color blindness,** a condition which causes a person to have difficulty in distinguishing red and green. An examination of this chart shows that only