

Rethinking Human Pigmentation

ALICE M. BRUES

Department of Anthropology, University of Colorado, Boulder,
Colorado 80302

KEY WORDS Pigmentation · Skin Color · Hair Color · Eye Color

ABSTRACT Though pigmentation has been of interest to anthropologists for a long time, its inheritance, and particularly the reasons for the incomplete correlation of skin, hair and eye, is poorly understood. It is suggested that this is largely due to a lack of genetically plausible hypotheses.

Taking into account racial and individual variation in pigment traits, and knowledge of pigmentation in other mammals, a minimum set of genetic factors for pigmentation in man is suggested. These include: (1) a set of polygenes affecting skin color only, (2) one locus for depigmentation of the eye, not affecting skin or hair, (3) one pleiotropic gene for reduction of pigment at all sites, and (4) one or more loci with multiple alleles producing blondness or rufosity of the hair in symmetrical patterns over the body.

I am very glad that Dr. Post has chosen human pigmentation for a renewal of interest, since it has been relatively neglected in recent years. For one thing, it has suffered by being an obvious and popularly recognizable correlate of human geographical variation, or to use that wicked word, *race*. We may not like to admit that political pressures, including those within our universities, can affect our choice of research topics, but it can be so. Fortunately at the present time we are receiving more political input from minority groups, who thoroughly approve of their own color, to somewhat outweigh that of their honky friends, who are still bravely trying to conceal the fact that skin color upsets them. A more soundly based reason is that pigmentation in man appears to be more complexly determined than that of the mammals generally used for genetic study, the difference probably being due to the presence in man of more modifier genes which produce intermediate and ambiguous phenotypes. Early work in human pigmentation was done without careful thought about these complexities and consequently resulted in interpretations which were clearly inadequate. Some of the naive pre-conceptions of this period are still with us. Anthropological genetics has tended to take the easier course towards study of traits with clear phenotypes and generally single-locus determination. That these traits were invisible to the human eye

was also a solace to workers sensitive on the subject of race.

A word is appropriate here about the significance of polygenetically determined traits in anthropology. I am sure we were all impressed by the recent demonstration (Szabo et al., '69) that the skin pigment in certain dark skins was not only present in greater quantity, but dispersed in smaller particles so as to increase its light-absorbing effect. The existence of a second mechanism for increasing light absorption in the skin fortifies the supposition that the dark phenotype has adaptive value. Similarly, the existence in certain areas of extreme phenotypes involving polygenic systems is more convincing of functional adaptation than is the existence of random gene frequency differences at single loci. Genetic drift may have produced much of this interpopulation variation. But the coincidence of alterations of gene frequencies at a number of loci, *all in the same phenotypic direction*, greatly strengthens the case for selection effect versus genetic drift. Thus the concentration of attention on single-gene, easily studied traits, which some of us have felt made us more respectable in the eyes of other biologists, may have tended to distract us from the study of functional, adaptive and evolutionarily significant traits which are still an important part of our proper subject matter and scientific responsibility. The non-adaptive (or supposedly non-adap-

tive) traits will always be of interest to those whose interest in human populations is a wholly historical one; but we need not be convinced that this is the only respectable form of physical anthropology.

A recent work which should be carefully read by anyone interested in human pigmentation is Searle's *Comparative Genetics of Coat Color in Mammals*, which appeared in 1968. This is a thoughtful summary and analysis of research on a large number of species, with particular emphasis on homologizing the various loci which influence hair color in different species and orders. (Incidentally, his 6-page summary of genetic knowledge of human pigmentation, about 2% of the entire volume, should make anthropologists blush.) Searle has pushed to the limits the process of homologizing the known loci for mammalian pigment determination, and makes it clear that some of his homologies may be forced. It is obvious, however, that the same mutations have occurred repeatedly in different lineages, on occasion under the very eye of the breeder, showing that the basic enzyme systems for pigment formation are similar over a wide taxonomic range. We have, therefore, a fair knowledge of the general potentialities of pigment variation in mammals, and may expect man to partake of the same genetic mechanisms. Of course hair color, which is for practical purposes *the* color of an ordinary mammal, is only one of the elements of human pigmentation which we generally consider. The skin color of most mammals is not conspicuous except in hairless areas like the nose-pad, which tends to correlate with the color of the furred areas. Eye color is also largely a separate problem, which has been little studied in other mammals. A perusal of the information available for mammals in general will make us well aware of the variety of genotype we may be faced with and the abundant possibilities of different genetic mechanisms lying behind what appear, at least superficially, to be identical phenotypes.

The three elements of human pigmentation: that is, skin, hair and eye color, are often thought of collectively and generalized in population descriptions. Actually, the correlations between the three do not hold up well when examined on either an individual or a population basis. At one time anthropologists stumbled along with a

sort of hypothesis that the three were inherently correlated by some pleiotropic mutation, which later somehow "broke up" into parts. I know of no cytogenic mechanism by which any such strange thing could happen, and I suggest that we keep an open mind on the existence of any inherent correlation between these different aspects of pigmentation.

If we were to take all the human beings in the world who have dark brown eyes and black or dark brown hair, we would not only have the vast majority of the human species, but would have a group which shows virtually the complete range of human skin color, from black to almost completely depigmented. In fact millions of individuals, nearly monopolizing large areas of southern Europe, northern Africa, central and eastern Asia, and parts of the aboriginal New World, have the darkest possible eye and hair color combined with skin which falls within "white" limits. This skin color represents one end of a continuum, obviously determined by a polygenic system. We do not know the "original" skin color of man. In view of the fact that human evolution took place largely in tropical areas, and that tropical man now generally has relatively dark skin, I would guess that the original skin color of man was at least medium to dark brown. If this is the case, it is the lighter skins which represent the mutations of later origin; in any case, I shall refer to these alleles as *Leucoderm* polygenes. (The comparative rarity of truly *black* phenotypes in mammals suggests that the darkest human skins also may be regarded as involving mutations from a hypothetical original genotype.) The *Leucoderm* polygenes can be defined as being without substantial effect on either eye or hair color, which they leave in the heavily pigmented condition which I have assumed is basic for our species.

Eye color is a very complex matter. I made a try at analyzing it almost 30 years ago (Brues, '46) and I am still not satisfied with the answers. Human eyes exhibit almost as much diversity of color as you wish to look for, and many loci must be involved. Blue eyes, at least some of them, are apparently a sex-linked recessive, and the heterozygote at that locus is probably one of the mixed varieties; it cannot be the only mixed eye, since mixed eyes occur in males

also. Analogy suggests that a variegated color due to a heterozygous sex-linked gene may be a true mosaic, like the coat of a calico cat, due to random inactivation of one X chromosome. The world distribution of the blue or gray eye and its relation to other depigmentation is an interesting one. It is common, of course, only in Caucasians, with a strong bias towards the higher latitudes. (Interestingly, it appears in Arctic Canidae also, both wild and domestic.) But light eyes in man occur sporadically among African Blacks and in India; they are known among American Indians and not uncommon in some tribes. It is easy to attribute this to European mixture in some of these areas, though due to the recessive inheritance it would not be expected to appear often except where admixture was extensive. However, the most interesting point in these scattered occurrences of the light eye is that it appears in individuals who resemble their neighbors in skin and hair color. This means the same thing whether it is attributed to racial admixture or to local mutation: namely, that it is independent of general pigment reduction. I suggest that the black-hair blue-eye combination of western Europe, classically associated with Ireland, is the same mutation. So I call the independent blue eye, combined with dark hair, and with dark skin if that is characteristic of the local population, the *Irish* mutation.

Another pigment variation which appears most conspicuously in a remote place but which I suggest should not be dismissed as insignificant is the occurrence of blond hair with heavily pigmented skin and eye. This occurs classically among the aborigines of central Australia, where it has been referred to as the "tawny" trait (Birdsell, '50). There is a strong popular belief that all blondism must be European in origin, so that this phenomenon was once attributed to white admixture. For the same reasons the same phenomenon when it occurs in Melanesia is still frequently explained by artificial bleaching. No one has accused the Australian desert dwellers of this technological accomplishment. The occurrence of light hair in these two related peoples appears to be a distinct mutation, which I have called *Australian*. Perhaps the term "Tawny" should have priority. Must we assume that this mutation is present only in

those areas in which the contrast of hair and skin makes it conspicuous? I think not. In Europeans we sometimes see a combination of dark eye and light hair, though not as commonly as the reverse. In general (I base this on Coon's summary, 1939) this is more common in eastern than in western Europe, and therefore is less often seen in the United States than the Irish mutation. It would be a great diseconomy of explanation to assume that this dark-eye light-hair combination in Europeans was anything but the coincidence of the Australian mutation, with the Leucoderm polygenes, common in this area, quite independently producing light skin color. One characteristic of the light hair found in Australian Aborigines is that it is most common in children, largely disappears in adult males but often persists in adult females. It is also true in Europeans that sometimes the fair hair of childhood, particularly in males, darkens very dramatically at puberty; of course permanently blond hair occurs also in Europeans. It would be premature to tie this specifically to the Australian mutation; but it points up the fact that adult dark hair should not be accepted as a simple trait. Hair which turns dark under the influence of sex hormones may be an entirely different genotype from hair which is dark from birth.

Is there a true "blond" genotype, in addition to these independent factors for light skin, light eyes and light hair? Perhaps this is most clearly seen when such individuals appear within populations of generally dark pigmentation. Among the phenotypes which have sometimes been called "albino" in man are some with minimum pigmentation which would pass for blond if they were encountered in a Leucoderm population. Such phenotypes sometimes appear in Africa, Melanesia, and among American Indians. These cases show clearly a pleiotropic effect on skin, hair and eye simultaneously. In a population already light-skinned due to the Leucoderm polygenes, would this phenotype appear different from a coincidence of Irish and Australian mutation? Probably not. I pulled some old data from my files the other day and checked the light-hair blue-eye combination on about 300 white Americans. The number of "true blonds" was about 60% in excess of the number expected on the basis of random

combination of hair and eye color. This certainly leaves a very open possibility that there are two different genotypes here: some who are random combinations of Irish and Australian mutations, and some who result from a pleiotropic mutation. The latter hypothetical allele I have called the *Albinoid* mutation.

Do these three factors I have suggested represent an unnecessary multiplicity of causes? Given three partially but not completely independent variables, this is a pretty good economy of hypothesis. If we accept these three (leaving alone the many modifiers there must be as well) we are saying that our general concept of "blondness" which has strongly affected our conceptualization of human pigmentation in the past, is due to a pleiotropic gene which explains a fraction of the light skins in the world, some of the light hair and some of the light eyes, but falls far short of explaining all the varieties of partial depigmentation which we find. Another kind of scheme, in effect a mirror image of the one we have presented, would picture the *Albinoid* mutation as a primary one, which depigmented everything, and then secondary mutations which counteracted its effects in limited ways. (I do not even consider that an *albinoid* condition could have been the human original.) This would require only two more mutations, a reverse to dark hair and a reverse to dark eye, to explain Caucasian pigment combinations in a very broad way. However, it would demand either a coincidence of two secondary mutations, or a still different pleiotropic gene, to explain the very abundant light-skin dark-hair dark-eye type, which as we have said is the commonest single kind of human there is. A scheme of reverse mutations from a basic blondism would make it much easier to get a combination of dark skin with blue eyes and light hair — the one kind of human being that is virtually nonexistent. In any case, we have presented only the roughest outline of a system. Questions like "What on earth is *medium brown* hair and what do we do with it?" I leave to the happy ingenuity of future investigators.

A persisting problem in human hair color is the presence of more or less red pigment (phaeomelanin) in addition to black (eumelanin). At one time we liked to think that the two pigments were genetically inde-

pendent and could be treated as two separate traits. It is probably more likely that the independent traits are (1) amount of pigment substrate and (2) its differentiation into red and black forms. The dichotomy of color has analogies among other vertebrates: many species of both birds and mammals have black and red phases. There seem to be two types of genetic control involved. There may be a factor which causes a melanocyte to be capable of producing only one of the colors (as in cats). Or there may be genes which cause melanin to be formed in different quantity or selectively differentiated into various amounts of the black or red forms in different parts of the body, producing symmetrical patterns. (In many animals, not including man, variation in pigment formation may be intermittent, producing color-banded hairs.) Two loci are involved in this kind of color patterning, designated A (agouti) and E (extension), and authorities disagree as to which of these loci certain traits should be assigned to. Variation of this kind in man is conspicuously shown by males who have beards of different color from the head hair. Some of these color patterns have apparent homologies in dogs, the black hair-red beard pattern being similar to some terriers and hounds, and the blond hair-black beard pattern similar to the fawn-with-black-muzzle of boxers and bulldogs. Close inspection in many of these cases in man shows that there is often a mixture of differently colored hairs in a single area, a trait called "brindle" in other mammals and considered a part of the same allelic system. A further complication well known in man is that red or reddish hair often darkens with age, and sequential studies are needed to determine if this is due to a change in relative numbers of different colored hairs. This again emphasizes that hair color should be evaluated on a developmental basis. The inclusion of beard color, which anthropologists have traditionally ignored, adds to the complexity of hair color description. Black hair with a red beard may not be the same genotypically as black hair with a black beard. From the genetic point of view the prospect is discouraging. Not only are these combination phenotypes ascertainable only in one sex; if styles change they may cease to be ascertainable at all.

Redness, which can be so conspicuous in

the hair, is possibly present in the eye also. A whole class of relatively light eye colors, which I have previously called Pale Mixed (Brues, '46), have areas of yellow or orange tint which appear qualitatively different from the brown pigment of many mixed and most dark eyes. These eye colors, incidentally, show no strong correlation with hair redness. There are some cases of intense red-orange eye color in non-human primates, and these probably would be the most easily studied for evaluation of the pigment involved. Hair rufosity is known to have correlates in the skin. Red-haired individuals are particularly liable to sunburn, even more so than ordinary blondes, many of whom tan rather effectively. Is their problem due to a lack of eumelanin and inadequacy of pheomelanin to absorb ultra-violet radiation? Or does the skin not produce pigment at all in the genotype which develops red pigment in the hair? To evaluate the presence of pheomelanin in the skin we need spectroscopic readings well controlled for the effect of red-reflecting blood pigments and the presence of carotene. Then, what are freckles? They show an indubitable, if inconsistent, correlation with redness of the hair. Are they analogous to the darker hairs of the red-brown brindled beard? Are they due to single hyperactive melanocytes, perhaps the product of somatic mutation? Do they represent some mosaic condition? Mosaicism is a most intriguing genetic phenomenon and it would be a pity to overlook the possibility.

One last note on hair color: I would not think it too unlikely that what I have called "Australian mutation" is another allele at the A or E locus, along with other patterned hair color variations. The tawny hair of the Pitjandjara shares with various red-hair phenotypes the liability to age change and sex difference, and is actually somewhat reddish in tone.

I have not really said much about genetics in these pages, though I have used genetic terms. I have discussed what might more accurately be called the natural his-

tory of human pigmentation. I think past failures in the field have resulted from inadequate thinking about the phenotypes and lack of meaningful hypotheses. The field of pigmentation may not even be a suitable one for the student who wants a good quick dissertation project, or the faculty member who wants a publication in time for the next swindle sheet he sends to the dean. Perhaps it is a study only for the tenured. I guarantee you moments of madness when you try to decide how to classify medium brown hair and various other ambiguous phenotypes which human flesh, at least human keratinized epidermis, is heir to. You will find a fertile field for speculation about the geographical distribution of some of the mutations which have occurred in man. Depigmentation involving several different forms and several different mutations appear to have been favored in certain northern latitudes. Is this entirely coincidence? What are the selective factors acting on pigmentation, either directly or through still unknown pleiotropic effects?

On every successive page of this paper I find myself making fewer statements and asking more questions. Sometimes we tend to believe that if a question has not been answered for 50 years, it is no longer a question. But the questions about human pigmentation are still there, and the longer we look the more we see. If any of you feel that I have not provided enough, I will be glad to supply some more privately.

LITERATURE CITED

- Brues, A. M. 1946 A genetic analysis of human eye color. *Am. J. Phys. Anthropol.*, 4: 1-36.
- Birdsell, J. B. 1950 Some implications of the genetical concept of race in terms of spatial analysis. *Cold Spring Harbor Symposia on Quantitative Biology*, 15: 259-314.
- Coon, C. S. 1939 *The Races of Europe*. MacMillan, New York.
- Searle, A. G. 1968 *Comparative Genetics of Coat Colour in Mammals*. Academic Press, London.
- Szabo, G., A. Gerald, M. A. Pathek and T. B. Fitzpatrick 1969 Racial differences in the fate of melanosomes in human epidermis. *Nature*, 222: 1081-1082.