# Human Skin Colour, Its Genetics, Variation and Adaptation: A Review

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## INTRODUCTION

Human skin colour is one of the most conspicuous human polytypic variations and has attracted probably more scholarly attention than any other aspect of human variability. It was the very first character used for racial classification and has served as a primary feature in most systems of racial classifications.

In the late nineteenth century, process of melanization in plants was discovered to be dependent on the enzyme tyrosinase. Subsequently the same pathway was found to be involved, by German histochemist Bruno Bloch, in pigmentation of human skin *in vitro*. By 1950 the melanocytes, the cells where these reactions took place, were identified and it was further found that these cells originate embryonically in the neural crest.

Edwards and Duntley (1939a) found that variation in human skin colour is mainly due to the presence of four pigments, namely, Melanin, Haemoglobin, Carotene and Melanoid. In addition to this, effect produced by the scattering of light from the skin surface is also involved in giving a particular skin colour to the person. G.A. Harrison (1957) demonstrated that there appears to be no significant difference between races in haemoglobin content provided there are no pathological and/ or malnutritive conditions. He further demonstrated that there appears to be constancy in the vascular supply under conditions of constant temperature and activity. He also found that the contributions of scattering of light and carotene appear to be respectively constant and in any case negligible. Further the melanoids are also found to be negligible even in dark skinned people particularly at the parts of the body protected from the sun.

No wonder that Edwards and Duntley (1939b) had suggested that "the coloured races owe their colours only to variations found in melanin content". No pigment other than the one present in the Whites, excepting an unusual rare red pigment among the New Guinea indigents (Walsh, 1971) is found in human skin. This red pigment is due to the accumulation of a red intermediary metabolite, namely, DOPACHROME.

## THE MELANIN PIGMENTATION

The melanin pigment granules are mostly found in the basal layer or the *stratum germinativum* inside the melanocytes. The *stratum basale* consists of columnar cells, the carotenocytes, with about 10% of the cells comprising melanocytes. This is the germinal layer of the skin which gives rise to the outer layer of cells and the melanin granules that pigment them. The *stratum spinosum* consists of several layers of irregular polyhedral cells, carotenocytes, flattened on their edges. Thus it can easily be inferred that the effect of haemoglobin of the blood flowing in the underlying papillary layer of the dermis on the skin colour will be masked by the dark brown melanin pigment present in the overlying basal layer of epidermis and particularly so in the darker populations.

The metabolic pathway involved in melanin synthesis is extremely complicated involving several intermediate steps. It starts with the amino acid tyrosine oxidized by the copper containing enzyme tyrosinase (TYR) to dihydroxyphenylalanine (DOPA) and then to dopaquinone. A mutation in the gene for enzyme tyrosinase (TYR) that produces a protein with decreased functionality will result in a reduced production of melanin and under extreme decrease in the functionality this produces a genetic form of albinism. Dopaquinone undergoes a series of non-enzymatic reactions and rearrangements forming the different molecules that are co-polymerized to make one of the two types of melanin: *Eumelanin*, which is the dark brown/ purple/ black compound found in skin/ hair, and *Phaeomelanin*, which is yellow to reddish brown pigment present in red hair and rarely in human skin. Both forms of melanin combine with other proteins to form the melanosome that is distributed from the melanocytes to the surrounding cells. There is a gradient of melanosome size and number from dark to intermediate to light skin colour

besides melanosomes of dark skins being more widely dispersed.

Activation of the melanocortin I receptor (MCIR) promotes the synthesis of eumelanin at the expense of phaeomelanin, although oxidation of tyrosine by tyrosinase (TYR) is needed for synthesis of both the types of pigments. The extent of pigment synthesis within melanosomes is affected by the membrane associated transport protein (MATP) and the pink-eyed dilution protein (P) which are the melanosomal membrane components.

Total amount of melanin produced is more important than the ratio of the two types of melanin. Both the light and the dark skinned individuals have similar number of melanocytes for the same body region (there being considerable variation between different body regions in number of melanocytes), but melanosomes that contain the pigment are more numerous and more pigmented in darker people than in light skinned people (Szabo et al., 1969; Toda et al., 1972).

**Measurement of Skin Colour**

Reflectance spectrophotometry has been extensively used for measuring skin pigmentation (See Weiner, 1951; Feather et al., 1988) In this method a reflectance spectrophotometer shines a light of specific wavelength (using a filter), and measures the intensity of light reflected by the skin. The technique involves alcohol wash of the skin on the inner upper arm of the subject, allowing time for local circulation run to normal and then shines the light on the skin and measure the skin reflectance.

**Genetics of Skin Colour**

Skin colour is a polygenic trait and multiple genetic loci are involved in determining it besides the environmental factors. Multiple genes working together produce a continuous distribution in a bell shaped curve of skin colour varying from light to dark. Early models (Harrison and Owen, 1957; Stern, 1970; Kalla, 1968) suggested 2-5 additive/ equal/ unequal genes being responsible for it. Recent works (Sturm, 1998) suggest many genes working together in very complex additive and non-additive combinations to give rise to various shades of human skin colour. The non-enzymatic conversions of dopaquinone into eumelanin and phaeomelanin, and their combination into melanosomes, are affected by several genetic loci.

Of nearly 100 different genes (Bennett and Lamoreux, 2003; Jackson, 1994), 15 to 20 mutations of mouse coat colour genes are found to have human homologues in which null mutations cause albinism.

Loss of functional alleles in a single gene, the melanocortin I receptor (MCIR), causes the characteristic phenotype of fair skin coupled with freckling and carrot red hair because of large amounts of phaeomelanin and small amounts of eumelanin (Sturm et al., 1998; Rees, 2000; Barsh, 2003). However, the MCIR variation significantly affects pigmentation only in populations who commonly have red hair and fair skin and its primary effects – to promote synthesis of eumelanin at the cost of phaeomelanin or vice versa – contribute little to the variation of skin reflectance among or between major ethnic groups.

The TYR, P and MATP genes are well known causes of albinism and their primary effects are limited to pigment cells (Oetting and King, 1999); of these gene *P* is highly polymorphic but the phenotypical consequences of *P* gene polymorphism are not yet known. However, the variation in the above MCIR gene sequence does not contribute significantly to variation in the human skin colour in different populations of the world, but a functional MCIR is important for dark skin.

**Skin Tanning Potential and Its Genetics**

Under the influence of ultra violet radiations, the human skin tans i.e. additional melanin pigment formation takes place. This takes place in two steps, first, the pigment migrates from lower epidermal layers to upper epidermal layers and in the second step more pigment is formed.

Skin Tanning is a two-stage acclimatizational response of the skin to the increasing levels of UV exposure. Immediate tanning results in the transient brownish tan because of exposure of the skin to UV-A and visible light. It reaches a maximum within 1-2 hours after the exposure and is lost between 3-24 hours after exposure. In immediate tanning no new melanosomes are formed and the likely mechanism is the photo-oxidation of existing melanin or epidermal elements. Delayed and sustained tanning too occurs due to repeated or focused exposure primarily to UV-B while UV-A and visible light may also play a role. In this, skin darkening

gradually starts from 48-72 hours after irradiation. Initially this darkening is the result of migration of the melanosomes from lower epidermal layers to the upper epidermal layers. Subsequently melanocytes enlarge, the dendrite density increases, melanosomes increase in number and the maximum tanning reaches upto 19 days after an exposure whereafter peeling of the skin sets in. Normally the sustained tanning goes off by nine and a half months but in some cases it may sustain even after a year of exposure.

The skin tanning potential is also found to be inherited (Kalla, 1972) and it is further found to be under the control of fewer genes than the normal skin colour as indicated by the respective parent child correlation in the normal and induced skin pigmentation respectively.

**Variation in Skin Colour**

The worldwide distribution of skin colour suggests an association with environmental factors varying with latitude, ultra violet radiation, in particular, the quantity of UV rays striking the surface of the earth from the sun and temperature. The skin colour tends to become lighter in the low UV regions and it tends to be darker in the tropical and sub-tropical high UV regions. Relethford (1997) analyzed the skin reflectance data from more than 100 populations all over the world and found that skin reflectance is lowest at equator and then gradually increases by about 8% per 10 degree of latitude in the northern hemisphere and about 4% per 10 degree of latitude in the southern hemisphere. This pattern clearly showed inverse correlation of skin reflectance with levels of UV radiations which are greater in the southern hemisphere than in the northern hemisphere. As Templeton (2002) rightly suggested we do not know how patterns of UV radiation have changed with time in the two hemispheres and that we also do not know when skin colour is likely to have evolved along with multiple emigration fro Africa and extensive genetic interchange over the last 500,000 years.

Skin Tanning potential also seems to vary from population to population. It tends to increase with the increase in normal skin pigmentation upto a particular level of normal skin colour beyond which the skin tanning potential declines with the further increase in the level of skin pigmentation as reported by Kalla (1969b).

**Variation due to Age and Sex in Skin Colour**

Is the so-called "Fair Sex" really fairer to man? No consistent inference in this regard can be drawn from the data on various populations. Data collected by Barnes (1929), Mazess (1967), Kalla (1968), Kalla and Tiwari (1972), Conway and Baker (1972), found no sex difference in skin colour. Significant sex differences in the skin colour were observed by Weiner et al. (1964), Daiz Ungria (1972), Harrison and Salzano (1966) with females being significantly lighter in skin colour. But Leugebe (1964) and Sunderland (1967), found the adult male to be significantly less pigmented than the adult females at the unexposed skin. Age seems to intervene in a big way with the sex differences in the skin colour. At pre-pubertal age, males are found to be lighter in skin colour than females, but the relationship changes after puberty (Kalla, 1973). Most of the studies show a pre-pubertal increase in the melanin pigment in both males and females, which is attributed to the pre-pubertal increase in the melanocyte stimulating hormone (MSH) (Marshall, 1960).

**Selection Operative on Skin Colour**

Selection favouring high levels of melanin pigmentation in areas of high UV radiations may involve several selective agents. Sun burn can cause skin lesions and subsequent infections and it may also predispose the skin to skin cancer. Highly pigmented skin would not allow ultra violet to penetrate into the underlying papillary layer of the dermis and thus protect the dermis from being damaged. But in the absence of high pigmentation and in high UV areas the papillary layer of the dermis would be damaged by the UV radiations. Thus selection seems to favour dark skins in high UV areas. In the low UV areas, if the skin has high level of melanin pigmentation then no vitamin D synthesis may take place in the dermis as the weak UV would be cut off from penetrating into the papillary layer of the dermis to be able to manufacture vitamin D.

One of the important functions of vitamin D is to actively cause calcium absorption across the walls of small intestine into the blood stream and calcium is used for the bone and tooth developments as well as for nervous and muscle action. The skeleton serves as a calcium reservoir. If calcium levels in intracellular fluid drops, hormones are released to cause resorption of bone

placing calcium into circulation. Such a continued resorption as may be evident in infants and children suffering from deficiency of vitamin D may cause rickets and in adults it may cause osteomalacia. Rickets refers to a defect in the calcification of growing bone so that the bones are structurally weak and unable to withstand mechanical pressure. It leads to muscle weakness, deformity of the long bones including bowed legs, Knuckle like projections around the rib cage (rachitic rosary), deformities of the pelvis that are often permanent, etc. While long bone deformity impairs locomotion, pelvic deformation can make childbearing dangerous and is potential enough to kill mother and baby. This could ultimately lead to much reduced fertility and enhanced mother and child mortality. Here it would be interesting to report that prior to widespread vitamin D supplementation in the 1930s, the black women living in the USA had nearly eight times greater pelvic deformities than the white women. It is thus quite evident that the populations not getting vitamin D supplementation, having high melanin content in skin and living in low ultra violet regions could suffer great viability loss on account of deficiency of vitamin D caused because of their skin pigmentation.

Further UV light is also found to cause denaturization or chemical breakdown of folic acid circulation in the blood. This can induce folic acid deficiency in the body even if the diet supplies adequate folic acid. This folic acid deficiency may lead to anaemia, infertility and birth defects particularly, neural tube defects. High melanin content in the epidermis of the persons living under high ultra violet regions can also protect the circulating folic acid and this may be an important selective force for dark skin people in low latitude areas.

Skin cancer is found among light skinned individuals living in tropical latitudes, for example, in Nigeria and Tanzania no albino over the age of 20 years was found to be free of pre-malignant skin lesions. Further chronic skin damage was found in every albino by the end of first year of life in Tanzania. This high rate and early (prior to age of reproduction) onset of skin damage suggests that cancer of skin may have been a strong selective pressure in tropical areas.

Ultra violet light is also known to suppress immune function. It has been hypothesized that increase in melanization could shield the immune system by protecting the blood borne components of the system from ultra violet (Jeevan and Kripke, 1993). However, recent study (Sturm, 1998) demonstrated that the differences in skin colour were not associated with differences in UV induced immune deficiency.

Besides the selection favouring pigmentation it also favours depigmentation. In ecosystems where there are low levels of UV radiations (primarily in the high latitudes) selection favours low level of melanin which may involve different factors:

1. The regulation of vitamin D synthesis (as mentioned above)
2. Frost bite sensitivity and cold tolerance

In view of the molecular evidence which traces human ancestry to tropical region of Africa, if we assume that our ancestors had dark skin and as they moved into high latitudes there would have been substantial selection favoring lower melanin content in the skin to improve vitamin D synthesis in order to sustain the lineage by protecting it against rickets and osteomalacia. However, there are counter arguments to the above hypothesis. For instance, Robins (1991) maintains that there is no evidence of rickets in northern zones of North Africa where skin colour was presumably dark – including among the Eskimos. It is further argued that skin lightning in response to lower UV is unlikely with the substantial storage of vitamin D. Besides one also needs to consider the effects of clothing of populations living in northern latitudes where selection against dark skin would be lessened and diet may contribute more vitamin D.

As our ancestors moved from the tropical/ sub-tropical lower latitudes into temperate/ sub-temperate higher latitudes, they also would have been subject to colder temperatures. The medical record evidences (mostly from the Korean war) appear to suggest that the people with heavily pigmented skin were more susceptible to frost bite. The direct evidence, however, comes from the animal studies that demonstrate that melanocytes are more easily destroyed by freezing than the rest of the skin cells. Cooling both light and dark skinned areas of the same Guinea pig showed that the dark skin of the guinea pig was more susceptible to frost bite. Frost bite is known to cripple hands and feet causing viability problems; besides it also makes the body part more prone to infections including gangrene, which may be fatal.

Social selection also seems to play a part in skin pigmentation in some populations (as in India)

where skin colour is regarded to be important from the point of view of mate selection. While dark males are avoided by fair skin girls, the males, irrespective of the shade of their skin color, opt more often for the lighter skinned females; this leads to what is called as assortative mating positive among the females and negative as well as positive among the males. Such a situation is sufficient to offset the Mendelian mating pattern with respect to genes controlling skin pigmenta-tion and also the other genes linked to the skin colour genes; this may have far-reaching consequences on the population's genetic composition.

### Selection - Relaxation in Human Skin Colour

While in the pre-civilized societies people were exposed to natural selection vis-a-vis their skin colour and might have run the high risk for their reproductive survival on this account, in the civilized world the effect of natural selection was relaxed particularly because of clothing, housing, air-conditioning, lifestyle, skin protective cosmetics, vitamin D therapy and improved health care system, etc. It is thus obvious that under these conditions natural selection against dark/ light skin, as the case may be, is likely to be substantively reduced and minimized to a great extent.

### Social Implications of Variation in Human Skin Colour

Human skin colour has also been the most controversial human characteristic as sensitivity to its variation (noticeable with naked eye) aroused "racial" feelings and the subsequent discrimination between the people – The White and The Black; though going by its classification, a third "race" namely, The Yellow too exists. The characteristic had acquired a lot of political overtone, though unfortunate and rationally avoidable. Despite the best efforts of the right thinking people the skin colour based discrimination, more commonly known as racial discrimination, still persists and at times becomes a big problem in certain parts of the world.

No wonder that the skin colour still remains one of the most important characteristic of mankind for his honourable survival.

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**ABSTRACT** Human skin colour is the most studied characteristic of man and was responsible for the first racial classification. Variation in it is chiefly due to variation in the melanin pigment in the melanocytes while haemoglobin, carotene and melanoid marginally contribute to it. It is a polygenic trait and recent works suggest many genes working together in very complex additive and non-additive combinations to influence its phenotypic expression. However, skin tanning potential seems to be under the influence of fewer genes. Pre-pubertal increase in the melanin is a consistent observation in almost all the populations studied so far. Selection favours high levels of melanin in areas of high UV radiations. The political overtones of variation in skin colour have been devastating.

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