Reconstructing the evolutionary history of human skin pigmentation

Íngrid Palomino Pérez
Grau de Biologia

INTRODUCTION

Skin colour is one of the most variable phenotypes of human race and is primarily determined by the pigment "melanin". Melanin is produced by melanocytes in the basal layer of epidermis inside intracellular organelles called melanosomes, which are subsequently transferred to adjacent keratinocytes that eventually will migrate to the upper layers of the epidermis. Melanin functions as a broadband UV absorbent, but it also has antioxidant and radical scavenging properties.

Pigmentation differences among human groups are mainly determined by the amount and type of melanin produced by melanocytes, as well as the size and distribution of melanosomes inside the keratinocyte cytoplasm:

- **Dark skin**: enriched in black eumelanin contained in bigger, more dense and more numerous melanosomes, distributed as single units (see African in Fig. 1).
- **Light skin**: enriched in light-brown eumelanin and yellow/red pheomelanins contained in smaller, less dense and less numerous melanosomes, clustered in groups (see Asian and European in Fig.1).

PURPOSE

In this paper I have intended to provide the reader with a fairly complete overview of the evolution of human skin pigmentation, from the main evolutionary explanations for the existing skin colour variation among human groups to the underpinning loci involved in the expression of this character that show signatures of selection, particularly focussing on how these loci have contributed to infer the evolutionary history of this distinctive trait.

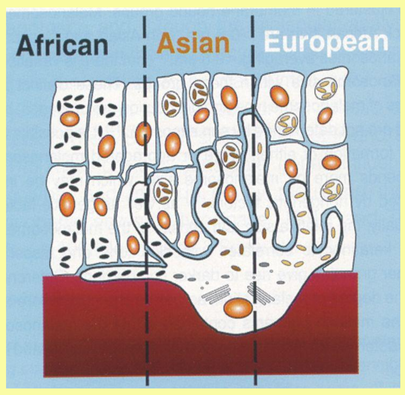


Fig. 1. Melanosomal characteristics and distribution within epidermal keratinocytes in three human populations. Source: Barsh, 2003.

MATERIAL AND METHODS

An extensive search in online scholarly databases, through search engines like "Pubmed" or "Google Scholar", provided the necessary information for the development of this project. Key words that have been used include "skin pigmentation" , "variation", "UVR", "evolution", "selection" and "genes" . The selection of the articles and reviews was carried out by applying a criterion of relevance based on the number of citations received together with the year of publication. Additional material such as printed journals, monographs and textbooks was also used.

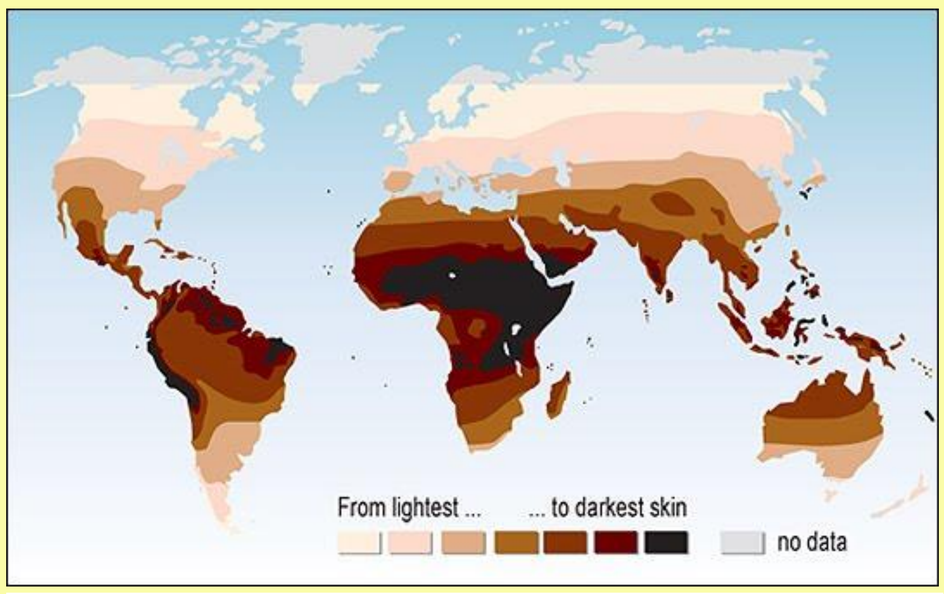
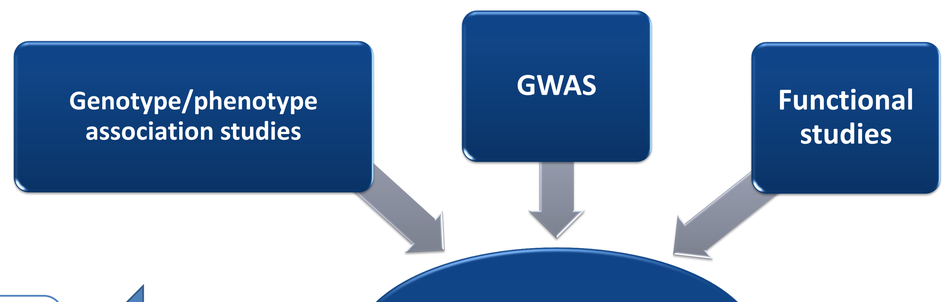


Fig. 2. Skin colour map for indigenous people showing darker phenotypes close to the Equator and a progressive depigmentation while we move to the poles. Source: Map updated in 2007 from Chaplin, 2004.

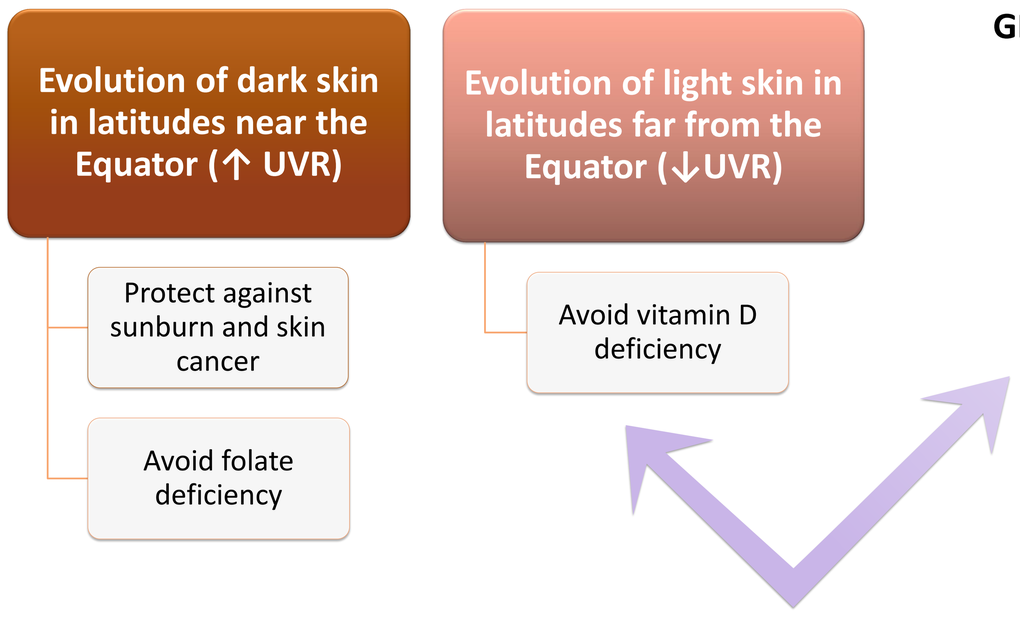
In Fig. 2 we can observe a clear latitudinal cline for skin pigmentation across world indigenous populations. This phenotype distribution has been attributed to the local UVR levels experienced by our ancestors.

GENETIC BASIS OF SKIN PIGMENTATION



MAJOR EVOLUTIONARY HYPOTHESIS

TO EXPLAIN HOW NATURAL SELECTION SHAPED THE DISTRIBUTION OF SKIN PIGMENTATION IN RESPONSE TO UVR LEVELS



GENETIC SIGNATURES OF POSITIVE SELECTION

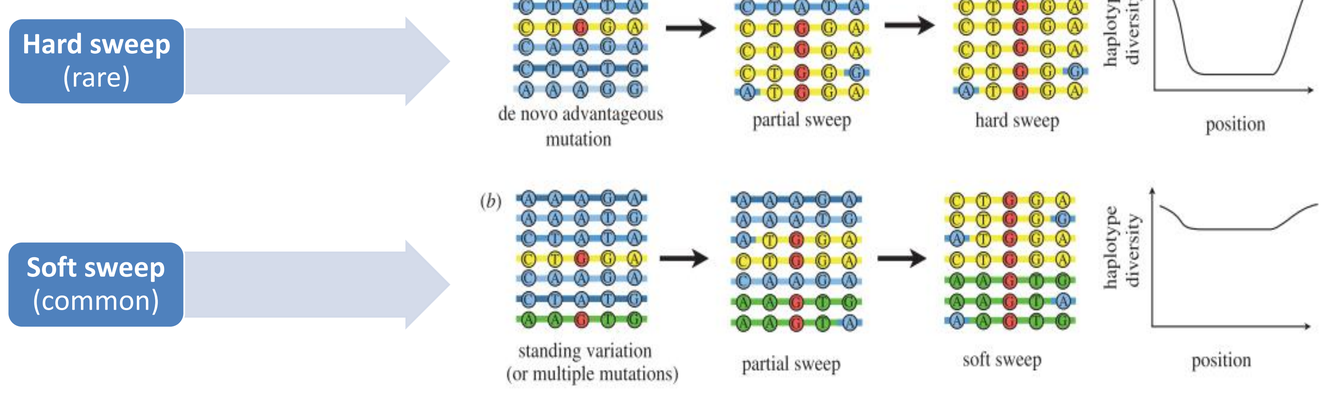
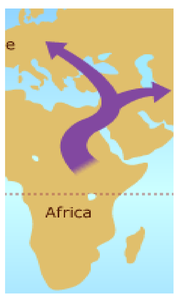


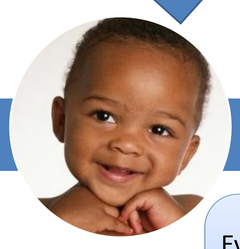
Fig. 3. Types of selective sweeps. Source: Novembre & Han, 2012.

Evolutionary history of skin pigmentation based on candidate gene studies



Derived phenotype

Evidence:
- Derived alleles from several genes that have shown signatures of positive selection in specific populations:
  - Eurasian: ASIP and KITLG → "Out of Africa"
  - European: SLC24A5 and SLC45A2 → Light skin evolution in Europeans and East Asians populations is partially independent (convergent evolution).



Ancestral phenotype

Evidence:
- MC1R (melanocyte receptor regulating eumelanin/pheomelanin synthesis): gene's function is highly conserved in African populations (purifying selection) but not in European and East Asian populations, where this gene is very polymorphic (relaxation of purifying selection).

CONCLUSIONS

- Skin pigmentation varies greatly among human populations and this differences are thought to be adaptive.
- The biological reason behind this differences is the type, amount and distribution of the melanin pigment.
- The character follows a latitudinal gradient and correlates with measured local UVR levels, supporting the different hypothesis proposed to explain the evolution of dark (sunburn/skin cancer protection and reduce folate photolysis) and light (promote vitamin D photosynthesis) skin colours.
- Several genetic approaches have been used in order to shed light on the evolutionary genetic history of this trait, with several evidences suggesting that dark skin appeared in our African ancestors after the loss of hair, whereas light skin evolved soon after the "Out of Africa" and separately after the European and East-Asian divergence, despite leading to similar phenotypes in both populations.

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