

# A genetic study on South Asians helps to understand human skin color variation

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Though genetics of skin pigmentation has shown recent advancements in the last decade, studies involving populations of South Asia, one of the major hot spots of pigmentation diversity, is still in its infancy. In a recent study publishing in *PloS Genetics*; an international team of scientists, led by researchers from the University of Tartu and the University of Cambridge, took skin color measurements from local residents in India to quantify the range and extent of variation in skin pigmentation phenotype and found that one of the important pigmentation genes; SLC24A5, plays a key role in skin pigmentation variation among South Asians. The comprehensive map of the genetic variant associated with light skin further revealed that it is quite wide spread in the subcontinent.

"It was interesting to see that the effect of geographical, linguistic, socio-cultural boundaries further shaped by strict endogamy which forms the backbone of the South Asian genetic diversity was so strongly reflected in the complex patterning of this [light skin](#) allele" explains Chandana Basu Mallick, lead author from University of Tartu, Estonia. She further adds, "This study helps us to understand the other possible mechanisms that could have contributed in shaping the existing biological spectrum of human [skin color](#) besides natural selection driven by UV rays and in further understanding of this complex phenotypic trait".

Another element of the study involved resequencing of SLC24A5 using diverse set of samples which helped to unveil an important fact; that Indians share the same mutation of SLC24A5 for their light skin as

Europeans and belong to the same haplotype background. Though evidence of positive selection in SLC24A5 has been well demonstrated in the previously reported genome-wide scans, the fact that South Asians have been underrepresented in the world-wide panels brought forward another question: whether there has been any evidence of positive selection for this gene in South Asians? To their surprise, they found that a differential pattern of selection revealed evidence of positive selection in North India, but not in South India.

"The variable presence of this light skin mutation across India suggests an intriguing interplay between the forces of natural selection and the unique demographic history and structure of the populations inhabiting the Indian subcontinent" says Mircea Iliescu, co-lead author from the University of Cambridge. This study also provides the first comprehensive estimate of the coalescence time of this allele, which is crucial in the understanding of the evolutionary history of light skin in humans.

Provided by Public Library of Science

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