

Supplementary data:

Analysis of *MC1R* variants in Indian oculocutaneous albinism patients: highlighting the risk of skin cancer among albinos

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Table 1. Sequences of primers used for PCR and sequencing.

MC1R_3F	5'-GACACCTGGAGGGGAAGAACTGT-3'
MC1R_5R	5'-GCACACACTTCCACCCAGAGACT-3'
MC1R_4F	5'-TGCACTCACCCATGTACTGCTTC-3'
MC1R_N1R	5'-GGCGCCAGGAAGCAGAGGC-3'
MC1R_N2R	5'-TTGACAAACGGGGACCAGGG-3'

The first two primers have been used for PCR as well as sequencing where as the other three have been used for sequencing only.

Table 2. *MC1R* variants identified in Indian OCA patients.

	Variant		Reported status	No. of samples (H_e/H_o)	Remarks
	Nucleotide change	Amino acid change			
1	c.1-5G>A	NA	Novel	1/0	None detected in 25 control individuals
2	c.274G>A	p.Val92Met	rs2228479	2/0	Associated with both melanoma and nonmelanoma skin cancer
3	c.444C>T	p.Tyr148Tyr	Novel	1/0	None detected in 25 control individuals
4	c.451C>T	p.Arg151Cys	rs1805007	1/0	Loss of function variant; associated with both melanoma and nonmelanoma skin cancer
5	c.488G>A	p.Arg163Gln	rs885479	9/1	Associated with melanoma, nonmelanoma skin cancer, especially basal cell carcinoma
6	c.515G>T	p.Ser172Ile	Reported mutation	1/0	None detected in 25 control individuals
7	c.699G>A	p.Gln233Gln	rs146544450	2/0	Not expected to have any biological effect
8	c.942A>G	p.Thr314Thr	rs2228478	24/2	Not expected to have any biological effect

H_e , heterozygous, H_o , homozygous; *MC1R* cDNA positions and amino acid residues have been used in accordance to NM_002386.3 and NP_002377.4, respectively.