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CONTEXT

Papua New Guineans are one of the most biologically and culturally diverse people of the world, resulting from complex demographic processes and the wide spectrum of geographical contexts. They settled all territories, from the high-altitude mountains to the coastal swamps on New Guinea Island as well as in the neighboring islands. Each of these populations adapted their culture and technologies to these environment, as did their biology.

A remarkable feature of the Papuan biological diversity resides in the large panel of skin pigmentation. Not only groups of Papuan ancestry show the darkest skin color worldwide, as in Bougainville, the variability between group is unmatched. This observation has long been made but so far no study focused on the genetic information that drives it in order to understand the anthropological mechanisms that led to this exceptional biological patrimony. Using whole genome sequences and multiple standardised skin pigmentation measurements, we conducted the first genome-wide analysis of Papuan skin pigmentation.

Sampling

106 participants for which four skin pigmentation variables were collected (using DSM II DermaSpectrometer,Cortex Technology) (Figure1):

-on the inner arm (baseline constitutive skin color) and on the top of the hand (most tanned region)
-melanin (main skin color pigment) and erythema (vascularisation in response to sunburn) index.

Anthropological questionnaires (language, genealogy) to access individual origins.
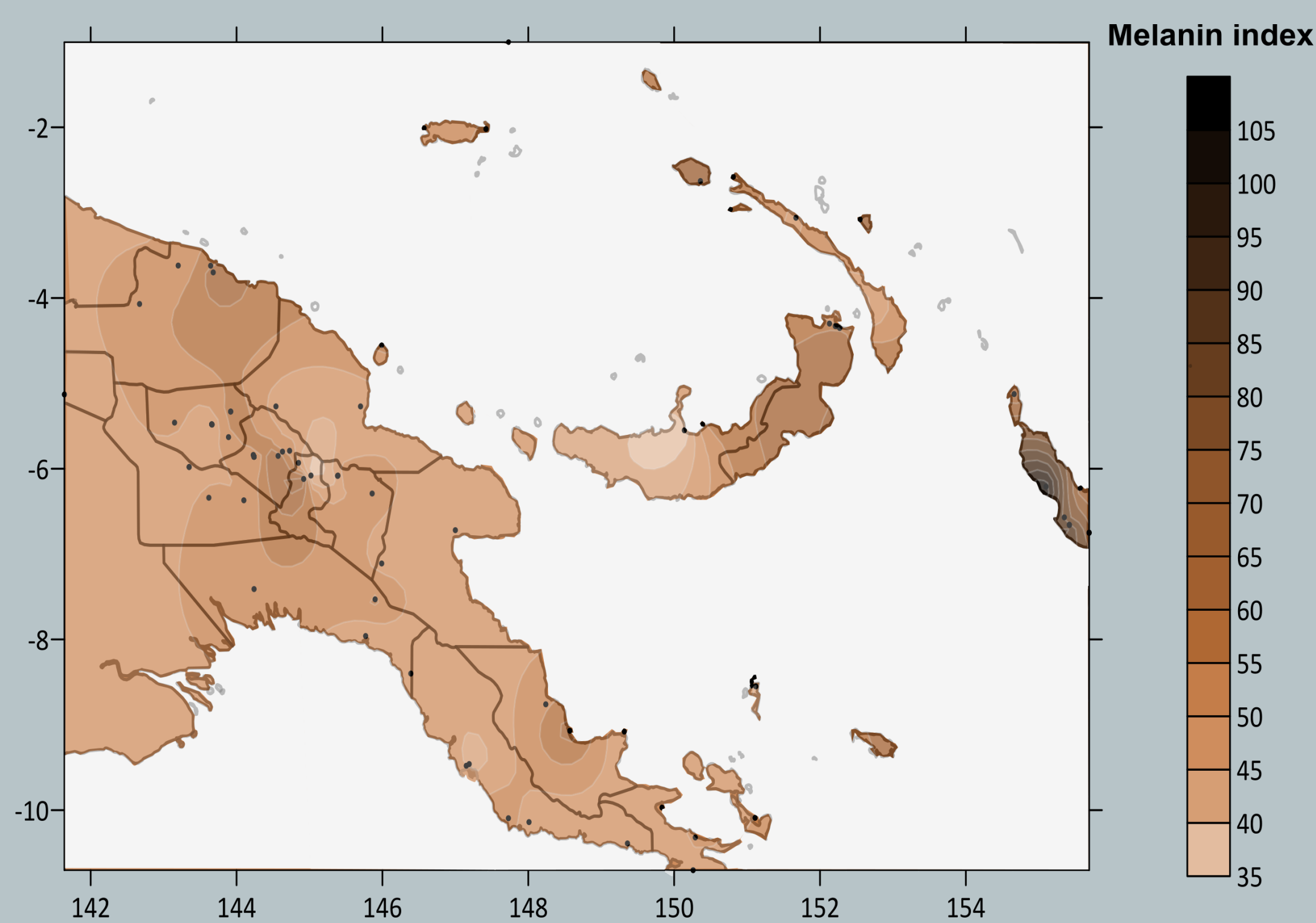
58 whole genome sequences (30X, Illumina X5).

Figure 1: Skin color variation within Papua New Guinea. Melanin index was took from the upper inner left arm. black dots representindividuals's location. x and y axis represent respectively longitude and latitude

Biostatistical analysis

Clustering analysis of skin pigmentation variables.

Pigmentation variables corrected for gender and East Asian ancestry (inferred from ADMIXTURE analysis).

Multivariate genome-wide association with Plink.multivariate (10^8 pernutations).

GO term enrichment analysis with Enrichr.

Candidate gene univariate associations with Plink:
-new associations found
-previously associated genes.

POLYGENICITY OF SKIN COLOR

To identify loci significantly associated with skin pigmentation, we performed a genome-wide multivariate association test on pigmentation variables (Figure 2).

8 genes were found significantty associated with skin pigmentation ($p<10^{-8}$). None of these genes were previously associated in humans. However, GO term enrichment analysis revealed their biological role in skin cells and inflammatory response. Indeed, based on bibliography, these genes are known actors of the epidermis physiology, some even being influenced by UV-induction (like AKR1C1). Our results show that these 8 genes have specific variations in Papuan groups that significantly explain the large diveristy of skin pigmentation.

A more in-depth analysis is now necessary to determine if some of these SNPs are population-specific, which would explain the striking difference of skin pigmentation between Papuan groups.
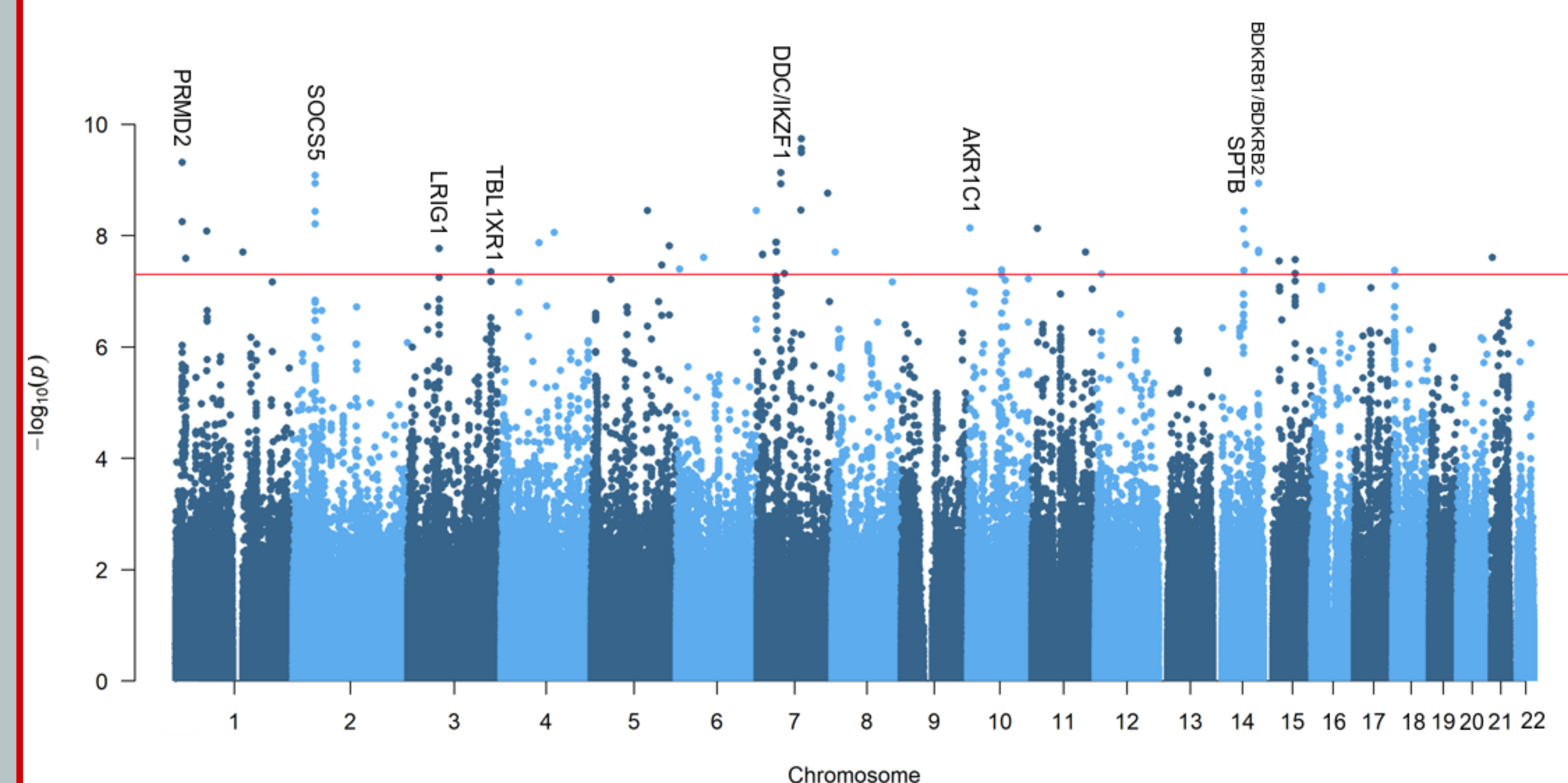

Figure 2: Multivariate genome-wide associations for skin pigmentation variables. Red line represents p-value = 1.0E-8. Top hits variants are noted with the closest gene. Multivariate associations were runon 10,000 permutations using Plink.multivariate.

A UNIQUE GENETIC PATTERN

Based on our multivariate association test and on bibliography, we selected 30 candidate genes related to skin pigmentation in diverse human groups. We ran univariate association tests on each candidate genes.

Each of the genes found in our multivariate analysis show more significant signal of association with the variable related to the melanin index of the inner part of the arm.
None of the genes previoulsy identified to be associated to skin pigmentation in Europeans and East Asians are implicated in skin color in Papuans. Only SMARCA2-VLDLR, previously found to be responsible for skin pigmentation in an African population, show a significant association (Figure 3). This could be due to the shared ancient ancestry or to an evolutive convergence caused by a similar environment.
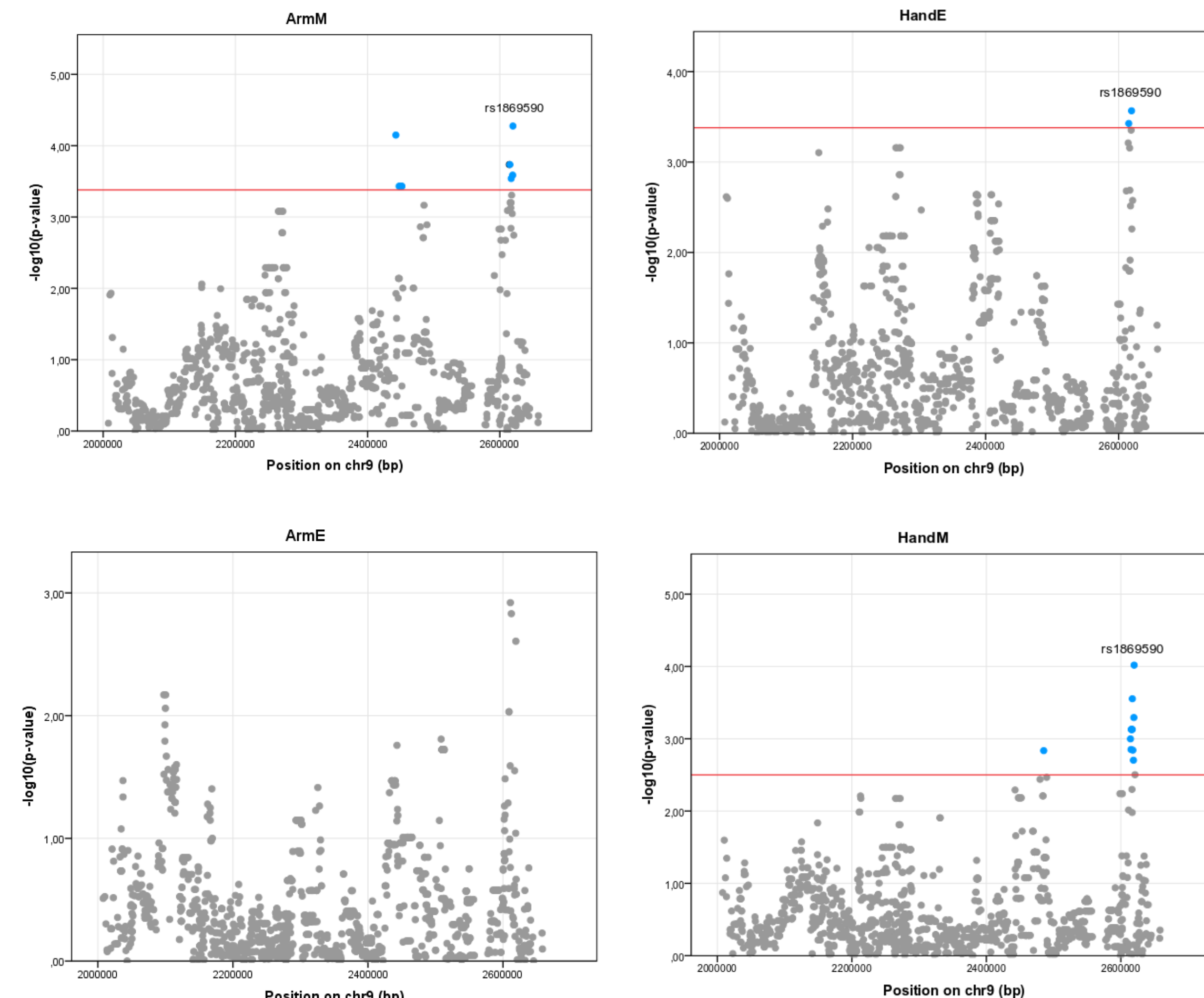

Figure 3: Univariate association of SMARCA2-VLDLR to each of the four skin pigmentation variables. Red line represents p-value = 4,2E-4. Significantly associated SNPs are represent by blue dots. ArmM: arm melanin, HandM: hand melanin , ArmE: arm erythema, HandE: hand erythema.Associations were runon 1.0E-8 permutation using Plink.

The uniqueness of Papuan skin pigmentation diversity is directly inherited by a set of genes with population-specific variants.

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More information on https://papuanpast.hypotheses.org/

PAPUAN PAST PROJECT