Table S1. Characteristics of the SNPs studieda.

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| **Gene** | **SNP ID** | **Localization** | **Number of exons** | **Mutation** | **Probable allele function** | **Studied populations** | **Reference (s)** |
| *ADAM17*b | rs1524668 | 2p25.1 | 19 | -968C>A - 5'UTR | - | Admixed Brazilian populations | Present study |
| *AFG3L1* | rs4785763 | 16q24.3 | pseudogene | A>C | A = red hair, freckles/ C = not-red hair, no freckles | Europeans | [1] |
| *ASIP* | rs6058017 | 20q11.22 | 4 | 8818G>A - 3'UTR | G = more melanin, no freckles/ A= less melanin, freckles | African-Americans from Washington, Europeans, East Asians | [1-4] |
| *HERC2* | rs1129038 | 15q13.1 | 93 | G>A - 3'UTR | A = blue eyes/ G = not-blue eyes | Europeans | [5, 6] |
| *MC1R* | rs1805009 | 16q24.3 | 1 | Asp294His - G>C | G = not-red hair, not-fairer skin/ C= red hair, fairer skin | Europeans, Caucasians | [7-9] |
|  | rs1805008 |  |  | Arg160Trp - C>T | T = blonde or red hair, freckles, skin sensitivity to sun, fairer skin/ C = not-blonde or not-red hair, no freckles | Europeans, Caucasians | [1,8-10] |
|  | rs1805007 |  |  | Arg151Cys - C>T | T = blonde or red hair, freckles, skin sensitivity to sun, fairer skin/ C = not-blonde or not-red hair, no freckles | Europeans, Caucasians | [1, 8-10] |
| *OCA2* | rs1800407 | 15q13.1 | 24 | Arg419Gln - G>A | G = not-green or brown or black eyes/ A = green or blue eyes | Caucasians of Pennsylvania, Europeans | [11, 12] |
|  | rs1800401 |  |  | Arg305Trp - C>T | C = blue eyes, lighter pigmentation/ T = brown/black eyes and darker pigmentation | Caucasians of Pennsylvania | [11] |
|  | rs1800414 |  |  | His615Arg - A>G | A = darker skin/ G = fairer skin | East Asian ancestry living in Canada | [13] |
| *SLC24A5 (NCKX5)* | rs1426654 | 15q21.1 | 9 | Ala111Thr - G>A | G = more melanin/ A = less melanin | Europeans, Mixed populations, African–Americans, Asians | [4, 14, 15] |
| *SLC45A2 (MATP)* | rs6867641 | 5p13.3 | 7 | -1169C>T - 5'UTR | C = fairer skin/ T = darker skin | Caucasians, Asians, African Americans, Australian Aborigines, Spanish Basques | [16] |
|  | rs26722 |  |  | Glu272Lys - G>A | G = fairer skin, lighter brown hair, not-brown eyes/ A = darker skin, hair, and eye color | Caucasians, Asians, African-Americans, Australian Aborigines | [17] |
|  | rs16891982 |  |  | Phe374Leu - C>G | C = darker skin, brown or black eyes/ G = blue eyes, lighter or medium skin, darker skin, hair, and eye color | Caucasians, Asians, African-Americans, Australian Aborigines, Europeans, Mixed populations | [4, 12, 14, 15, 17] |
| *TPCN2* | rs3750965 | 11q13.2 | 25 | Lys376Arg - A>G | A = blonde hair / G = brown hair | Europeans | [1] |
|  | rs3829241 |  |  | Gly734Glu - G>A | A= blond hair, skin sensitivity to sun/ G= brown hair, darker skin | Europeans | [1] |
| *TYR* | rs1042602 | 11q14.3 | 5 | Ser192Tyr - C>A | C = skin sensitivity to sun, freckles, brown hair/ A = no freckles, blonde hair | Europeans | [1, 10] |
|  | rs1126809 |  |  | Arg402Gln - G>A | G= not-blue eyes, brown hair/ A=blue eyes, blonde hair, skin sensitivity to sun | Europeans | [1] |

aKey to abbreviations: A = Adenine, *ADAM17* = a disintegrin and metallopeptidase domain 17, *AFG3L1* = ATPase family gene 3-like 1 (pseudogene), Ala = alanine, *ASIP* = agouti signalling protein, Arg = arginine, Asp = aspartic acid, C = Cytosine, G = Guanine, Gln = glutamine, Glu = glutamic acid, Gly = glycine, *HERC2* = HECT and RLD domain containing E3 ubiquitin protein ligase 2, His = histidine, Leu = leucine, Lys = lysine, *MATP* = membrane associated transporter protein, *MC1R* = melanocortin 1 receptor, *NCKX5*= Na/Ca/K exchanger 5, *OCA2* = oculocutaneous albinism II, Phe = phenylalanine, Ser = serine, *SLC24A5* = solute carrier family 24, member 5, *SLC45A2* = solute carrier family 45 member 2, T= Thymine, Thr = threonine, *TPCN2* = two pore segment channel 2, Trp = tryptophan, *TYR* = tyrosinase (gene), Tyr = tyrosine (amino acid), UTR = untranslated region.

b*ADAM17* was mentioned by Norton et al. [4] as an ‘a priori pigmentation candidate gene’, with a positive selection signal in East-Asians. We selected the *ADAM17*rs1524668 SNP because the derived allele is in high frequency in Amerindians (HGDP-CEPH, http://www.cephb.fr/en/hgdp/diversity.php/).

Table S2. Genotype and allele frequencies for 18 SNPs in the *Gaucho* (N = 352) and *Baiano* (N = 148) samples**.**

|  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
|  | rs1042602 | | rs1126809 | | rs6867641 | | rs26722 | | rs16891982 | | rs1800407 | |
|  | 1 = C/2 = A | | 1 = G/2 =A | | 1 = C/2 = T | | 1 = C/2 = T | | 1 = C/2 = G | | 1 = G/2 = A | |
| Genotypes/alleles | Gaucho | Baiano | Gaucho | Baiano | Gaucho | Baiano | Gaucho | Baiano | Gaucho | Baiano a | Gaucho | Baiano |
| 11 | 0.367 | 0.483 | 0.667 | 0.752 | 0.484 | 0.000 | 0.826 | 0.815 | **0.070** | **0.302** | 0.880 | 0.875 |
| 12 | 0.460 | 0.392 | 0.286 | 0.234 | 0.410 | 0.750 | 0.171 | 0.178 | **0.321** | **0.388** | 0.120 | 0.125 |
| 22 | 0.173 | 0.126 | 0.047 | 0.014 | 0.106 | 0.250 | 0.003 | 0.007 | **0.609** | **0.309** | 0.000 | 0.000 |
| 1 | 0.597 | 0.678 | 0.810 | 0.869 | 0.689 | 0.375 | 0.911 | 0.904 | **0.230** | **0.496** | 0.940 | 0.938 |
| 2 | 0.403 | 0.322 | 0.190 | 0.131 | 0.311 | 0.625 | 0.089 | 0.096 | **0.770** | **0.504** | 0.060 | 0.062 |
|  | rs1800401 | | rs1800414 | | rs1129038 | | rs6058017 | | rs3750965 | | rs3829241 | |
|  | 1 = C/2 = T | | 1 = A/2 = G | | 1 = G/2 = A | | 1 = G/2 = A | | 1 = A/2 = G | | 1 = G/2 = A | |
| Genotypes/alleles | Gaucho | Baiano | Gaucho | Baiano | Gaucho | Baiano | Gaucho | Baiano | Gaucho | Baiano | Gaucho | Baiano |
| 11 | 0.883 | 0.799 | 0.994 | 0.993 | **0.327** | **0.653** | **0.012** | **0.095** | 0.437 | 0.514 | 0.487 | 0.587 |
| 12 | 0.117 | 0.188 | 0.006 | 0.007 | **0.472** | **0.326** | **0.228** | **0.361** | 0.452 | 0.363 | 0.398 | 0.371 |
| 22 | 0.000 | 0.013 | 0.000 | 0.000 | **0.201** | **0.021** | **0.760** | **0.544** | 0.111 | 0.123 | 0.115 | 0.042 |
| 1 | 0.941 | 0.893 | 0.997 | 0.997 | **0.563** | **0.816** | **0.126** | **0.276** | 0.663 | 0.695 | 0.686 | 0.773 |
| 2 | 0.059 | 0.107 | 0.003 | 0.003 | **0.437** | **0.184** | **0.874** | **0.724** | 0.337 | 0.305 | 0.314 | 0.227 |
|  | rs1426654 | | rs1524668 | | rs1805009 | | rs1805008 | | rs1805007 | | rs4785763 | |
|  | 1 = G/2 = A | | 1 = G/2 = T | | 1 = G/2 = C | | 1 = C/2 = T | | 1 = C/2 = T | | 1 = A/2 = C | |
| Genotypes/alleles | Gaucho a | Baiano | Gaucho | Baiano | Gaucho | Baiano | Gaucho | Baiano | Gaucho | Baiano | Gaucho | Baiano |
| 11 | **0.026** | **0.097** | 0.407 | 0.432 | 0.971 | 0.986 | 0.914 | 1.000 | 0.945 | 0.993 | 0.045 | 0.056 |
| 12 | **0.147** | **0.366** | 0.462 | 0.480 | 0.029 | 0.014 | 0.086 | 0.000 | 0.055 | 0.007 | 0.364 | 0.347 |
| 22 | **0.827** | **0.538** | 0.131 | 0.088 | 0.000 | 0.000 | 0.000 | 0.000 | 0.000 | 0.000 | 0.591 | 0.597 |
| 1 | **0.100** | **0.279** | 0.638 | 0.672 | 0.986 | 0.993 | 0.957 | 1.000 | 0.973 | 0.997 | 0.227 | 0.229 |
| 2 | **0.900** | **0.721** | 0.362 | 0.328 | 0.014 | 0.007 | 0.043 | 0.000 | 0.027 | 0.003 | 0.773 | 0.771 |

aGenotype frequencies deviate from Hardy-Weinberg Equilibrium;

Allele and genotype frequencies which differ (*p* < 0.001) between *Gaucho* and *Baiano* are in bold.

Table S3. Mean and range values for the ancestry proportion (%) in the *Baiano* and *Gaucho* samples.

|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
|  | ***Baiano* subsample** | | |  | ***Gaucho* subsample** | | |  |
|  | Mean (SD) | Mininum | Maximum |  | Mean (SD) | Mininum | Maximum | *p* |
| European ancestry | 61.47 (22.51) | 2.42 | 100 |  | 84.86 (18.06) | 0 | 100 | < 0.001 |
| African ancestry | 29.91 (20.30) | 0 | 88.49 |  | 6.99 (10.49) | 0 | 91.13 | <0.001 |
| Native American ancestry | 8.62 (6.91) | 0 | 36.80 |  | 8.15 (9.07) | 0 | 55.99 | 0.017 |

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