Análisis de equilibrio de HWE

Lista de SNP analizados

|  |  |
| --- | --- |
| 'rs17599586' | 'HbF change, %' |
| 'rs2295644' | 'HbF change, %' |
| 'rs10483801' | 'HbF and HbF response to hydroxyurea' |
| 'rs2182008' | 'HbF and HbF response to hydroxyurea' |
| 'rs9319428' | 'HbF and HbF response to hydroxyurea' |
| 'rs3751395' | 'HbF and HbF response to hydroxyurea' |
| 'rs2387634' | 'HbF and HbF response to hydroxyurea' |
| 'rs10494225' | 'HbF and HbF response to hydroxyurea', |
| 'rs7977109' | 'HbF and HbF response to hydroxyurea' |
| 'rs816361' | 'HbF and HbF response to hydroxyurea' |
| 'rs7309163' | 'HbF and HbF response to hydroxyurea' |
| 'rs5006884' | 'Regulation of HbA2 level' |
| 'rs8002446' | 'HbF and HbF response to hydroxyurea' |
| 'rs826729' | 'HbF and HbF response to hydroxyurea' |
| 'rs765587' | 'HbF and HbF response to hydroxyurea' |
| 'rs9693712' | 'HbF and HbF response to hydroxyurea' |
| 'rs172652' | 'HbF and HbF response to hydroxyurea' |
| 'rs380620' | 'HbF and HbF response to hydroxyurea' |
| 'rs2693430' | 'HbF and HbF response to hydroxyurea' |
| 'rs12155519' | 'HbF and HbF response to hydroxyurea', |
| 'rs28384513' | 'Basal Levels of HBF' |
| 'rs9402686' | 'Basal Levels of HBF' |
| 'rs9399137' | 'Basal Levels of HBF' |
| 'rs4895441' | 'Basal Levels of HBF' |
| 'rs7581162' | 'Basal Levels of HBF' |
| 'rs10189857' | 'Basal Levels of HBF' |
| 'rs1427407' | 'Basal Levels of HBF' |
| 'rs7599488' | 'Basal Levels of HBF' |
| 'rs766432' | 'Basal Levels of HBF' |
| 'rs11886868' | 'Basal Levels of HBF' |
| 'rs4671393' | 'Basal Levels of HBF' |
| 'rs7557939' | 'Basal Levels of HBF' |
| 'rs10184550' | 'Basal Levels of HBF' |
| 'rs10128556' | 'Basal Levels of HBF' |
| 'rs2310991' | 'HBF levels in response to Hydroxyurea treatment' |
| 'rs61743453' | 'HBF levels in response to Hydroxyurea treatment' |
| 'rs334' | 'sickle cell anemia' |

Rs334 se incluye acá y no en el análisis de regresión logística

Poblaciones: "AFR", "AMR", "EAS", "EUR", "SAS"

Cálculo de HWE para cada SNP en cada una de las poblaciones, los siguientes SNP no se encontrarían en equilibrio de Hardy- Weinberg.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **snp** | **Population** | **description** | **chi\_value** | **p\_value** |
| rs10128556 | SAS | Basal Levels of HBF | 354,4552 | 1,07E-77 |
| rs10128556 | AMR | Basal Levels of HBF | 246,7468 | 2,63E-54 |
| rs10128556 | AFR | Basal Levels of HBF | 566,0709 | 1,2E-123 |
| rs10128556 | EAS | Basal Levels of HBF | 462,1858 | 4,3E-101 |
| rs10128556 | EUR | Basal Levels of HBF | 316,4231 | 1,95E-69 |
| rs10189857 | EAS | Basal Levels of HBF | 20,06321 | 4,4E-05 |
| rs10189857 | AMR | Basal Levels of HBF | 92,1718 | 9,66E-21 |
| rs10189857 | EUR | Basal Levels of HBF | 179,5613 | 1,02E-39 |
| rs10189857 | AFR | Basal Levels of HBF | 447,1257 | 8,09E-98 |
| rs10189857 | SAS | Basal Levels of HBF | 7,021048 | 0,029881 |
| rs10494225 | EUR | HbF and HbF response to hydroxyurea | 517,4806 | 4,3E-113 |
| rs10494225 | AMR | HbF and HbF response to hydroxyurea | 327,2085 | 8,86E-72 |
| rs10494225 | SAS | HbF and HbF response to hydroxyurea | 432,2749 | 1,36E-94 |
| rs10494225 | AFR | HbF and HbF response to hydroxyurea | 448,2387 | 4,64E-98 |
| rs10494225 | EAS | HbF and HbF response to hydroxyurea | 528,2404 | 2E-115 |
| rs11886868 | AMR | Basal Levels of HBF | 80,34456 | 3,58E-18 |
| rs11886868 | AFR | Basal Levels of HBF | 28,48313 | 6,53E-07 |
| rs11886868 | EAS | Basal Levels of HBF | 511,0362 | 1,1E-111 |
| rs11886868 | SAS | Basal Levels of HBF | 210,5475 | 1,91E-46 |
| rs172652 | AFR | HbF and HbF response to hydroxyurea | 49,54167 | 1,75E-11 |
| rs172652 | SAS | HbF and HbF response to hydroxyurea | 312,4771 | 1,4E-68 |
| rs172652 | EUR | HbF and HbF response to hydroxyurea | 207,1368 | 1,05E-45 |
| rs172652 | EAS | HbF and HbF response to hydroxyurea | inf | 0 |
| rs172652 | AMR | HbF and HbF response to hydroxyurea | 171,1094 | 6,98E-38 |
| rs17599586 | EAS | HbF change, % | inf | 0 |
| rs17599586 | SAS | HbF change, % | 477,6657 | 1,9E-104 |
| rs17599586 | EUR | HbF change, % | 481,9601 | 2,2E-105 |
| rs17599586 | AFR | HbF change, % | 615,3077 | 2,4E-134 |
| rs17599586 | AMR | HbF change, % | 356,733 | 3,44E-78 |
| rs2182008 | EUR | HbF and HbF response to hydroxyurea | inf | 0 |
| rs2182008 | SAS | HbF and HbF response to hydroxyurea | inf | 0 |
| rs2182008 | AFR | HbF and HbF response to hydroxyurea | 201,9845 | 1,38E-44 |
| rs2182008 | AMR | HbF and HbF response to hydroxyurea | 283,9491 | 2,19E-62 |
| rs2295644 | SAS | HbF change, % | 205,8865 | 1,96E-45 |
| rs2295644 | EUR | HbF change, % | 224,6866 | 1,62E-49 |
| rs2295644 | EAS | HbF change, % | 252,7167 | 1,33E-55 |
| rs2295644 | AMR | HbF change, % | 201,1632 | 2,08E-44 |
| rs2295644 | AFR | HbF change, % | 384,9919 | 2,51E-84 |
| rs2310991 | EAS | HBF levels in response to Hydroxyurea treatment | 9,728914 | 0,007716 |
| rs2387634 | EAS | HbF and HbF response to hydroxyurea | inf | 0 |
| rs2693430 | EUR | HbF and HbF response to hydroxyurea | 369,1575 | 6,89E-81 |
| rs2693430 | AFR | HbF and HbF response to hydroxyurea | 52,07308 | 4,93E-12 |
| rs2693430 | AMR | HbF and HbF response to hydroxyurea | 227,9695 | 3,14E-50 |
| rs2693430 | EAS | HbF and HbF response to hydroxyurea | 235,4426 | 7,49E-52 |
| rs2693430 | SAS | HbF and HbF response to hydroxyurea | 188,1194 | 1,41E-41 |
| rs334 | AMR | sickle cell anemia | inf | 0 |
| rs334 | AFR | sickle cell anemia | 669,7165 | 3,7E-146 |
| rs380620 | SAS | HbF and HbF response to hydroxyurea | 29,99445 | 3,07E-07 |
| rs380620 | EUR | HbF and HbF response to hydroxyurea | 53,55173 | 2,35E-12 |
| rs380620 | AFR | HbF and HbF response to hydroxyurea | 248,4805 | 1,1E-54 |
| rs380620 | AMR | HbF and HbF response to hydroxyurea | 106,0345 | 9,44E-24 |
| rs4671393 | EAS | Basal Levels of HBF | 27,43307 | 1,1E-06 |
| rs4671393 | AFR | Basal Levels of HBF | 24,83807 | 4,04E-06 |
| rs4671393 | SAS | Basal Levels of HBF | 179,2484 | 1,19E-39 |
| rs4671393 | AMR | Basal Levels of HBF | 32,2236 | 1,01E-07 |
| rs4671393 | EUR | Basal Levels of HBF | 136,5117 | 2,27E-30 |
| rs4895441 | EAS | Basal Levels of HBF | 351,4358 | 4,86E-77 |
| rs4895441 | EUR | Basal Levels of HBF | 367,4829 | 1,59E-80 |
| rs4895441 | SAS | Basal Levels of HBF | 463,6068 | 2,1E-101 |
| rs4895441 | AMR | Basal Levels of HBF | 305,5343 | 4,51E-67 |
| rs4895441 | AFR | Basal Levels of HBF | 661,9941 | 1,8E-144 |
| rs5006884 | SAS | Regulation of HbA2 level | 354,6249 | 9,87E-78 |
| rs5006884 | AFR | Regulation of HbA2 level | 407,6082 | 3,08E-89 |
| rs5006884 | EUR | Regulation of HbA2 level | 372,5312 | 1,28E-81 |
| rs5006884 | AMR | Regulation of HbA2 level | 268,4789 | 5,02E-59 |
| rs5006884 | EAS | Regulation of HbA2 level | 461,0498 | 7,7E-101 |
| rs61743453 | AFR | HBF levels in response to Hydroxyurea treatment | 728,4883 | 6,5E-159 |
| rs61743453 | AMR | HBF levels in response to Hydroxyurea treatment | inf | 0 |
| rs7309163 | AMR | HbF and HbF response to hydroxyurea | 225,011 | 1,38E-49 |
| rs7309163 | AFR | HbF and HbF response to hydroxyurea | 44,29208 | 2,41E-10 |
| rs7309163 | EUR | HbF and HbF response to hydroxyurea | 268,9595 | 3,95E-59 |
| rs7309163 | SAS | HbF and HbF response to hydroxyurea | 234,6203 | 1,13E-51 |
| rs7309163 | EAS | HbF and HbF response to hydroxyurea | 272,7217 | 6,01E-60 |
| rs7557939 | EAS | Basal Levels of HBF | 511,0362 | 1,1E-111 |
| rs7557939 | AMR | Basal Levels of HBF | 14,76837 | 0,000621 |
| rs7599488 | AMR | Basal Levels of HBF | 87,10243 | 1,22E-19 |
| rs7599488 | AFR | Basal Levels of HBF | 451,5556 | 8,8E-99 |
| rs7599488 | EUR | Basal Levels of HBF | 184,4137 | 9,02E-41 |
| rs7599488 | EAS | Basal Levels of HBF | 18,26966 | 0,000108 |
| rs816361 | AFR | HbF and HbF response to hydroxyurea | 508,6557 | 3,5E-111 |
| rs816361 | AMR | HbF and HbF response to hydroxyurea | 251,5265 | 2,41E-55 |
| rs816361 | EAS | HbF and HbF response to hydroxyurea | 316,4722 | 1,9E-69 |
| rs816361 | SAS | HbF and HbF response to hydroxyurea | 378,6624 | 5,95E-83 |
| rs816361 | EUR | HbF and HbF response to hydroxyurea | 339,2567 | 2,14E-74 |
| rs9319428 | AFR | HbF and HbF response to hydroxyurea | 6,132478 | 0,046596 |
| rs9693712 | EAS | HbF and HbF response to hydroxyurea | 323,1362 | 6,79E-71 |
| rs9693712 | AFR | HbF and HbF response to hydroxyurea | 470,8223 | 5,8E-103 |
| rs9693712 | AMR | HbF and HbF response to hydroxyurea | 104,695 | 1,84E-23 |
| rs9693712 | SAS | HbF and HbF response to hydroxyurea | 48,71858 | 2,64E-11 |
| rs9693712 | EUR | HbF and HbF response to hydroxyurea | 13,89978 | 0,000959 |

23 de los 37 SNP analizados se encuentran en desequilibrio de HW

* En 4 poblaciones: rs11886868, rs2182008, rs380620, rs7599488
* En una o dos poblaciones: rs2310991 (EAS), rs2387634 (EAS), rs334 (AMR, AFR), rs61743453(AMR, AFR), rs7557939 (EAS, AMR), rs9319428 (AFR)