

## SUPPLEMENTARY DATA

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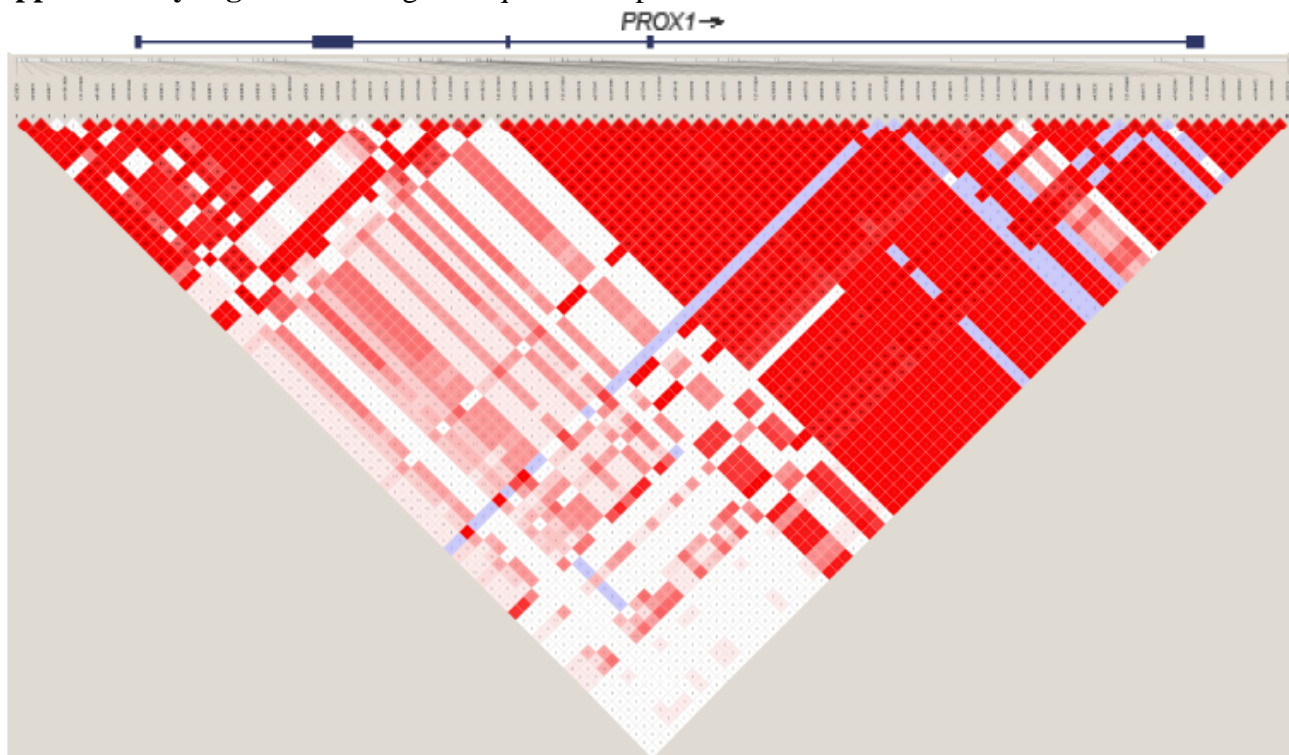
**Supplementary Table 1.** Genotype distributions of the 12 genotyped *PROXI* SNPs in the HELENA study.

| SNP        | Position in <i>PROXI</i> | Genotype | n (frequency) | MAF  | <i>P</i> HWE          |
|------------|--------------------------|----------|---------------|------|-----------------------|
| rs340877   | promoter                 | AA       | 304 (0.27)    | 0.47 | 0.20                  |
|            |                          | AG       | 579 (0.52)    |      |                       |
|            |                          | GG       | 237 (0.21)    |      |                       |
| rs340874   | promoter                 | CC       | 291 (0.26)    | 0.48 | 0.55                  |
|            |                          | CT       | 568 (0.51)    |      |                       |
|            |                          | TT       | 258 (0.23)    |      |                       |
| rs340837   | intron 1                 | TT       | 268 (0.24)    | 0.49 | 0.02                  |
|            |                          | TG       | 602 (0.54)    |      |                       |
|            |                          | GG       | 254 (0.22)    |      |                       |
| rs340835   | intron 1                 | GG       | 321 (0.29)    | 0.45 | 0.05                  |
|            |                          | GA       | 593 (0.52)    |      |                       |
|            |                          | AA       | 216 (0.19)    |      |                       |
| rs4655313  | intron 1                 | AA       | 768 (0.68)    | 0.18 | 0.19                  |
|            |                          | AG       | 316 (0.28)    |      |                       |
|            |                          | GG       | 42 (0.04)     |      |                       |
| rs11802122 | intron 2                 | GG       | 789 (0.70)    | 0.16 | 0.95                  |
|            |                          | GT       | 310 (0.27)    |      |                       |
|            |                          | TT       | 30 (0.03)     |      |                       |
| rs2289002  | intron 2                 | TT       | 597 (0.54)    | 0.27 | 0.99                  |
|            |                          | TC       | 437 (0.39)    |      |                       |
|            |                          | CC       | 80 (0.07)     |      |                       |
| rs3754138  | intron 3                 | GG       | 650 (0.57)    | 0.24 | 0.60                  |
|            |                          | GT       | 420 (0.37)    |      |                       |
|            |                          | TT       | 62 (0.06)     |      |                       |
| rs12748973 | intron 3                 | CC       | 1057 (0.95)   | 0.02 | 0.39                  |
|            |                          | CT       | 56 (0.05)     |      |                       |
|            |                          | TT       | 0 (0.00)      |      |                       |
| rs4655482  | intron 4                 | GG       | 1063 (0.94)   | 0.03 | 0.03                  |
|            |                          | GA       | 59 (0.05)     |      |                       |
|            |                          | AA       | 3 (0.01)      |      |                       |
| rs12092859 | intron 4                 | CC       | 1078 (0.95)   | 0.02 | 4.11x10 <sup>-6</sup> |
|            |                          | CG       | 43 (0.04)     |      |                       |
|            |                          | GG       | 4 (0.01)      |      |                       |
| rs10494972 | intron 4                 | TT       | 1034 (0.91)   | 0.05 | 4.73x10 <sup>-4</sup> |
|            |                          | TA       | 93 (0.08)     |      |                       |
|            |                          | AA       | 8 (0.01)      |      |                       |

MAF, minor allele frequency. HWE, Hardy-Weinberg equilibrium. Positions are based on the sequence NM\_002763 (chr1: 212,223,454...212,281,411).

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**Supplementary Figure 1.** Linkage disequilibrium pattern across *PROX1*.



Schematic representation of the linkage disequilibrium by measurements of  $D'$  values (red color scale) and  $r^2$  coefficients (values in the squares) for 80 *PROX1* SNPs based on the 1000 Genomes EUR reference panel.