Genetic diversity and natural selection footprints of the glycine amidinotransferase gene in various human populations

Running Title: Diversity and Natural selection on GATM

Keywords: GATM, AGAT, Genetic diversity, Natural selection

Asifullah Khan^{1,2,§}, Lei Tian ^{1,§}, Chao Zhang ¹, Kai Yuan ¹, Shuhua Xu^{1,3,4,*}

¹Chinese Academy of Sciences (CAS) Key Laboratory of Computational Biology, Max Planck Independent Research Group on Population Genomics, CAS-MPG Partner Institute for Computational Biology (PICB), Shanghai Institutes for Biological Sciences, Chinese academy of Sciences, Shanghai 200031, China;

²Department of Biochemistry, Abdul Wali Khan University Mardan (AWKUM), Mardan, Khyber Pakhthunkhwa, Pakistan;

³ School of Life Science and Technology, ShanghaiTech University, Shanghai 200031, China;

⁴Collaborative Innovation Center of Genetics and Development, Shanghai 200438, China.

[§]These two authors have contributed equally to this work.

*Correspondence and requests for materials should be addressed to S.X. (xushua@picb.ac.cn).

Supplementary figures legends:

Figure S1: Estimation of pairwise F_{ST} values for the *GATM* gene among different human populations.

Figure S2: Distribution of F_{ST} differentiation ratios for the *GATM* gene and 68 statin response-associated genes. Black dots represent genes that contain no SNPs.

Figure S3: (A) Allele frequency plot of *GATM* eQTL experimentally proved to be associated with statin associated myopathy. (B) Population wide global *Fst* calculation for three eQTLs experimentally proved in association with statin response. Two of these eQTLs (i.e. rs1719247 and rs1346268) are showing allele frequency difference > 1 % whole genome *Fst* differentiation.

Figure S4: Estimation of *lnRsb*, ΔDAF , and *F*_{ST} around the *GATM* gene. The red and green lines indicate the respective top 1% and top 5% thresholds of these abovementioned parameters at the whole-genome level.

Figure S5: Recombination hotspots observed within the vicinity of the *GATM* gene in the CEU population. The *GATM* gene is indicated in red. Data on the CHB and YRI populations are also shown for comparison.

Figure S6: Linkage disequilibrium (LD) plot across the *GATM* gene in the CEU, CHB, and YRI populations.



Figure S1



Figure S2

Figure S3

A.



B.











