

Population size shapes the type of nucleotide variations in humans

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Supplementary material

Supplementary Fig legends

Figure S1. Correlation between effective population size (N_e) and the ratio AT→GC to GC→AT (β) estimated for homozygous SNVs from whole genomes of the 1000 Genomes Project. The relationship was highly significant ($P < 10^{-6}$).

Figure S2. Relationship between the effective population size (N_e) and the ratio of nucleotide changes within the same types (β): **(A)** within strong types i.e. C→G/G→C **(B)** within weak types i.e. A→T/T→A. The ratios were estimated using the high frequency SNVs (DAF > 0.9) belonging to 27 populations obtained from the 1000 genome Project.

Figure S3. Relationship between the effective population size (N_e) and the ratio of nucleotide changes within the same types (β): **(A)** within strong types i.e. C→G/G→C **(B)** within weak types i.e. A→T/T→A. The ratios were estimated using the homozygous SNVs belonging to 126 populations obtained from the Simon Genome Project.

Figure S4. Relationship between the effective population size (N_e) and the normalized ratio of AT→GC to GC→AT (β') changes using equation 2 (see methods). We used A↔T and G↔C to normalize AT→GC and GC→AT changes respectively. **(A)** High frequency SNVs (DAF>0.9) and **(B)** Homozygous SNVs of the 1000 genome project **(C)** Homozygous SNVs from the Simons Genome Diversity project. The relationships were highly significant ($P < 10^{-6}$).

Figure S5. The relationship between effective population size (N_e) and the normalized ratio AT→GC/GC→AT (β) estimated for homozygous SNVs present in individual genomes

belonging to 126 distinct populations of the world. This is very similar to Fig 4A except that the nucleotide diversities of non-Africans were 5% reduced while calculating N_e in order to neutralize the difference in mutation accumulation rates between Africans and non-Africans as reported recently. The correlation was highly significant ($P < 10^{-6}$).

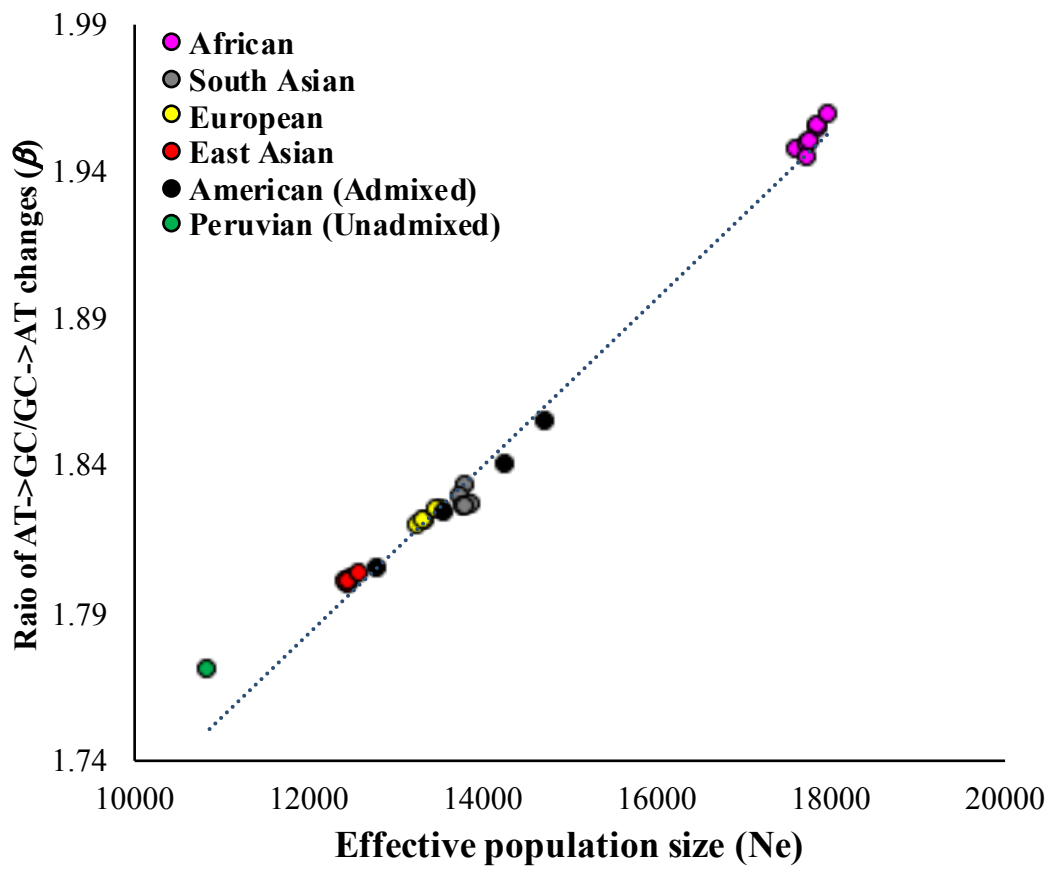


Figure S1

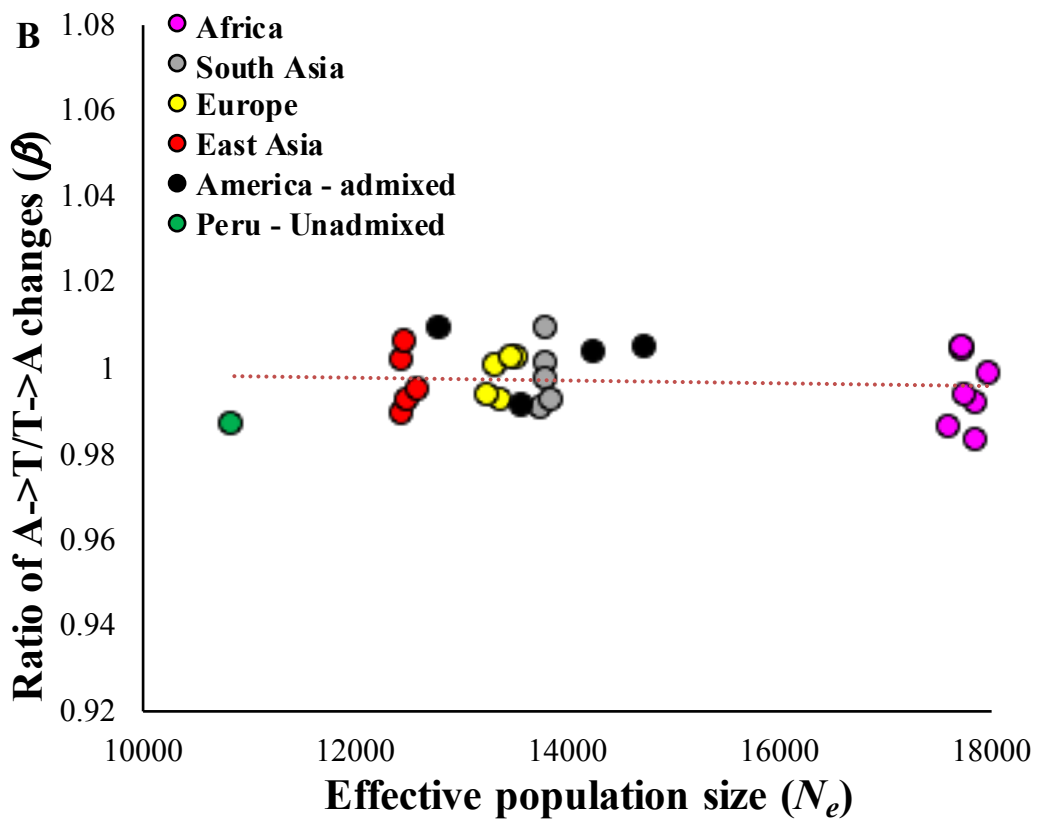
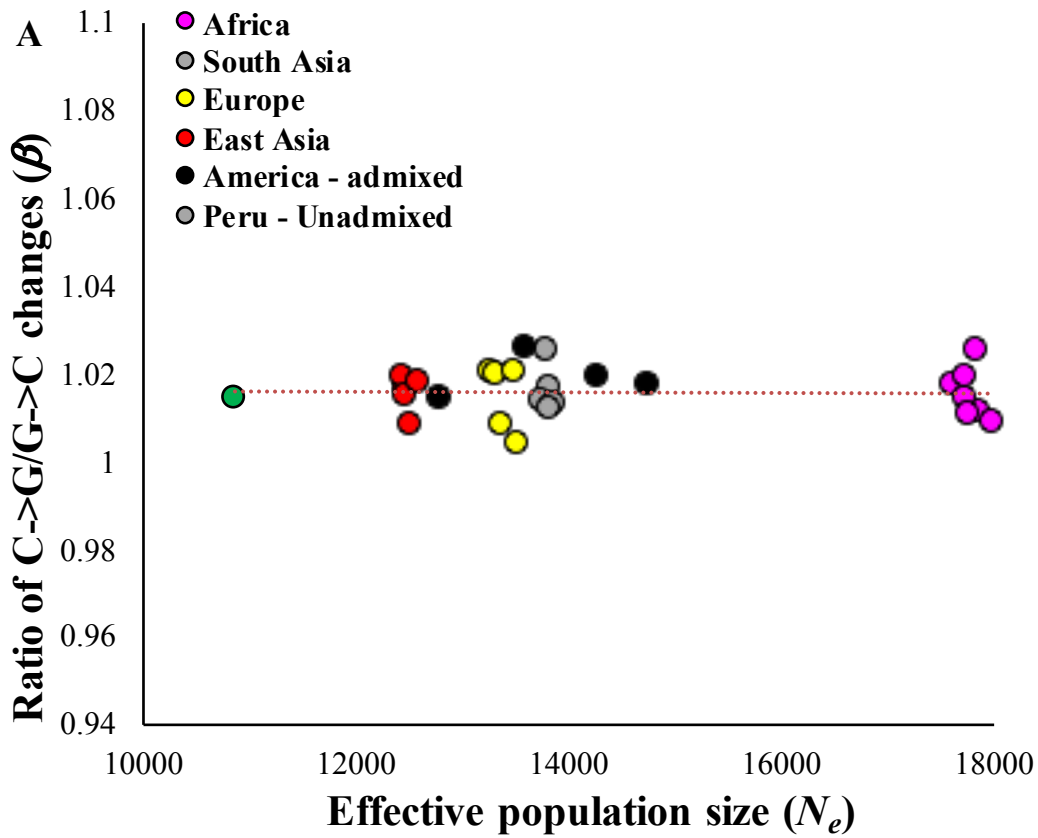


Figure S2

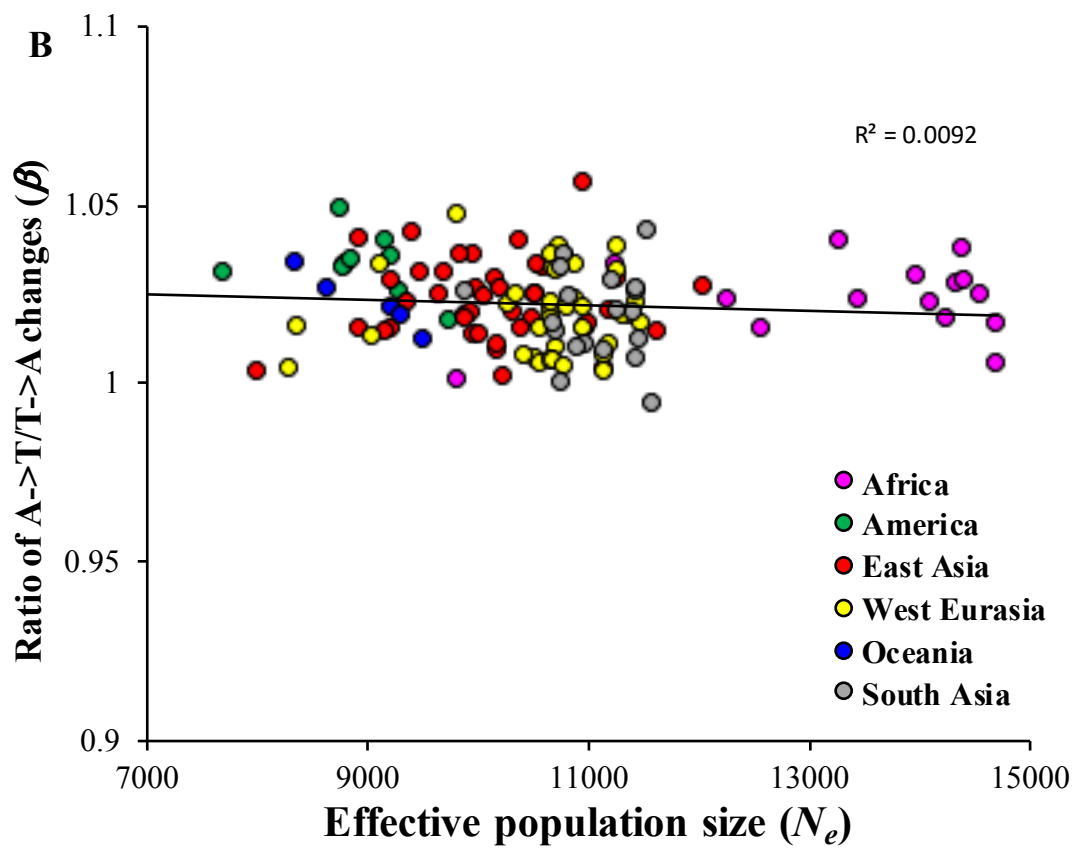
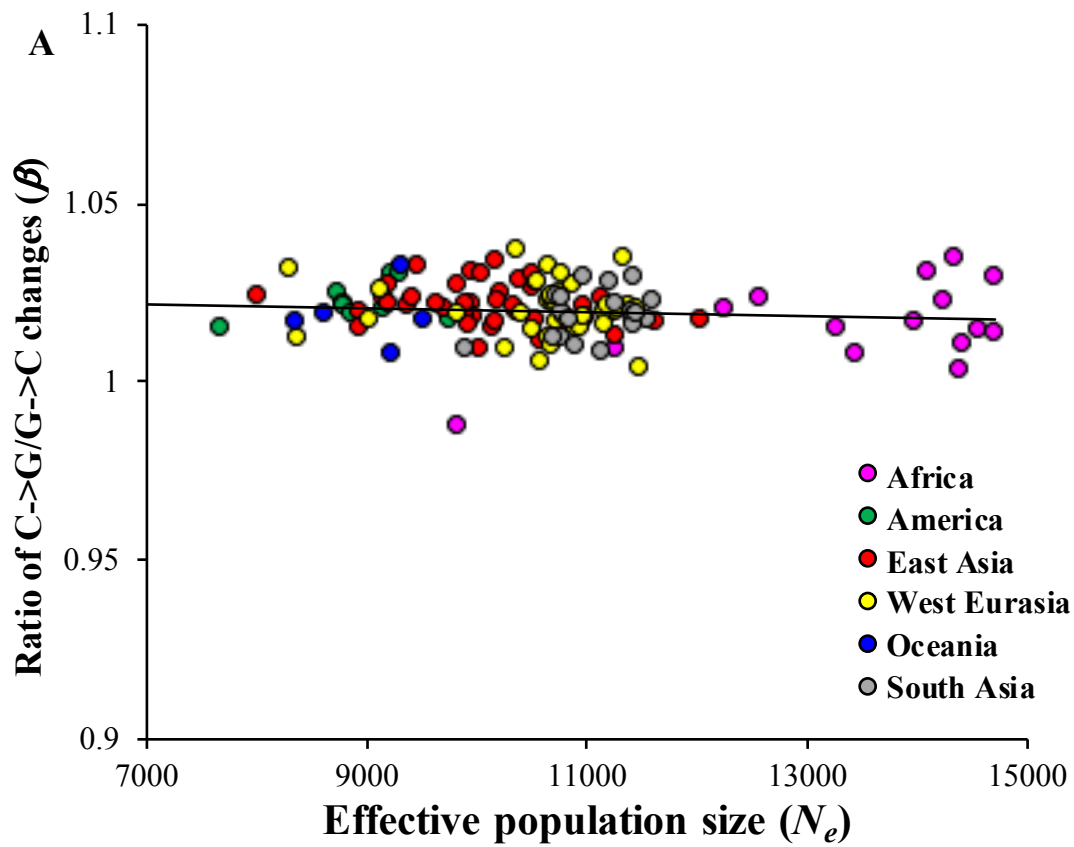


Figure S3

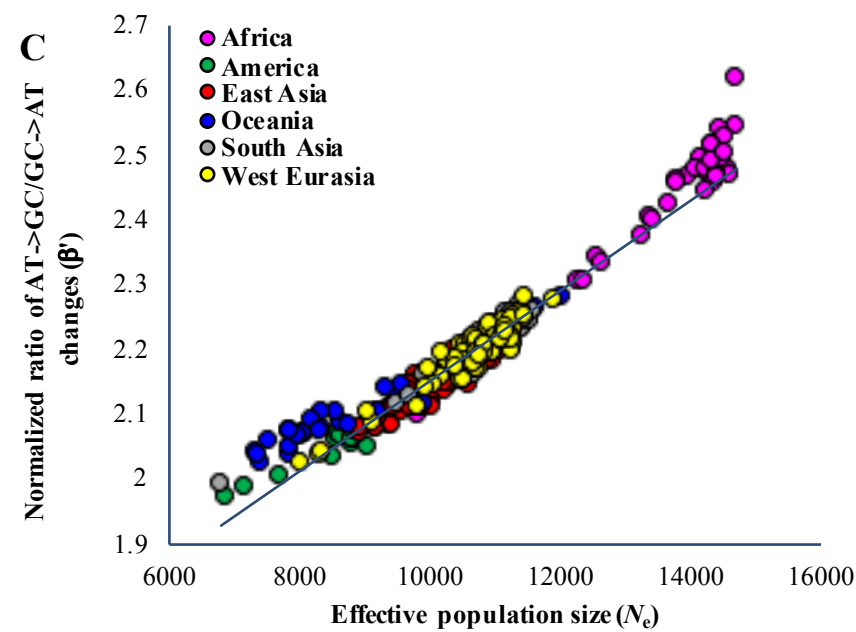
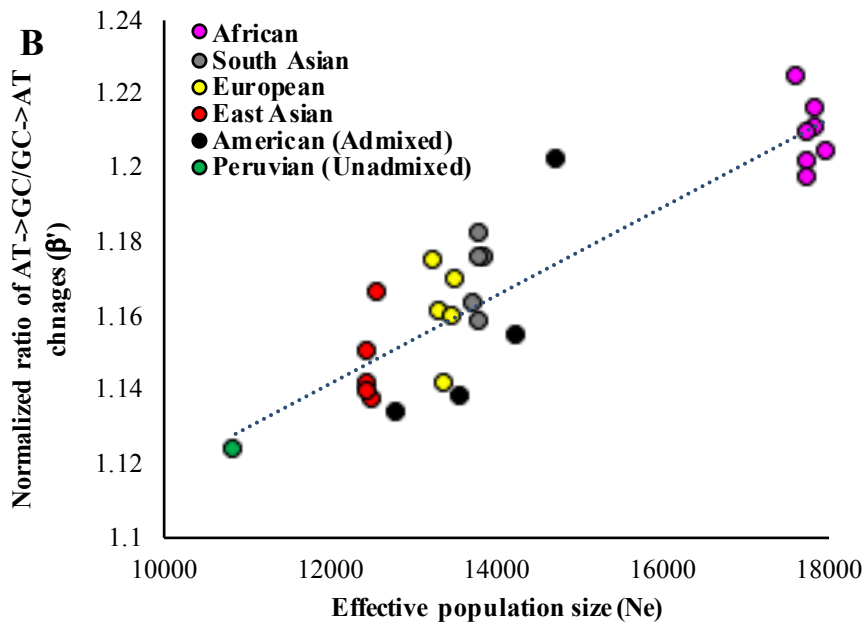
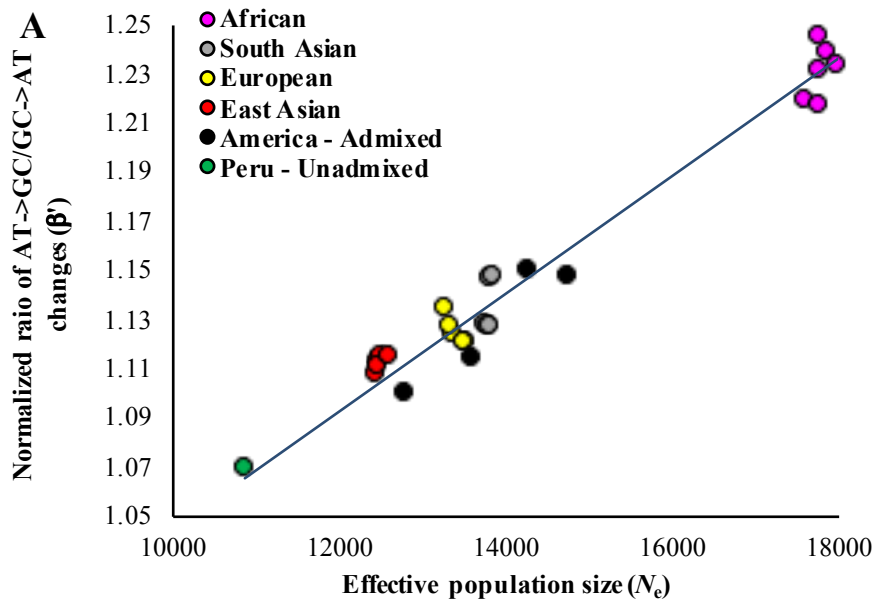


Figure S4

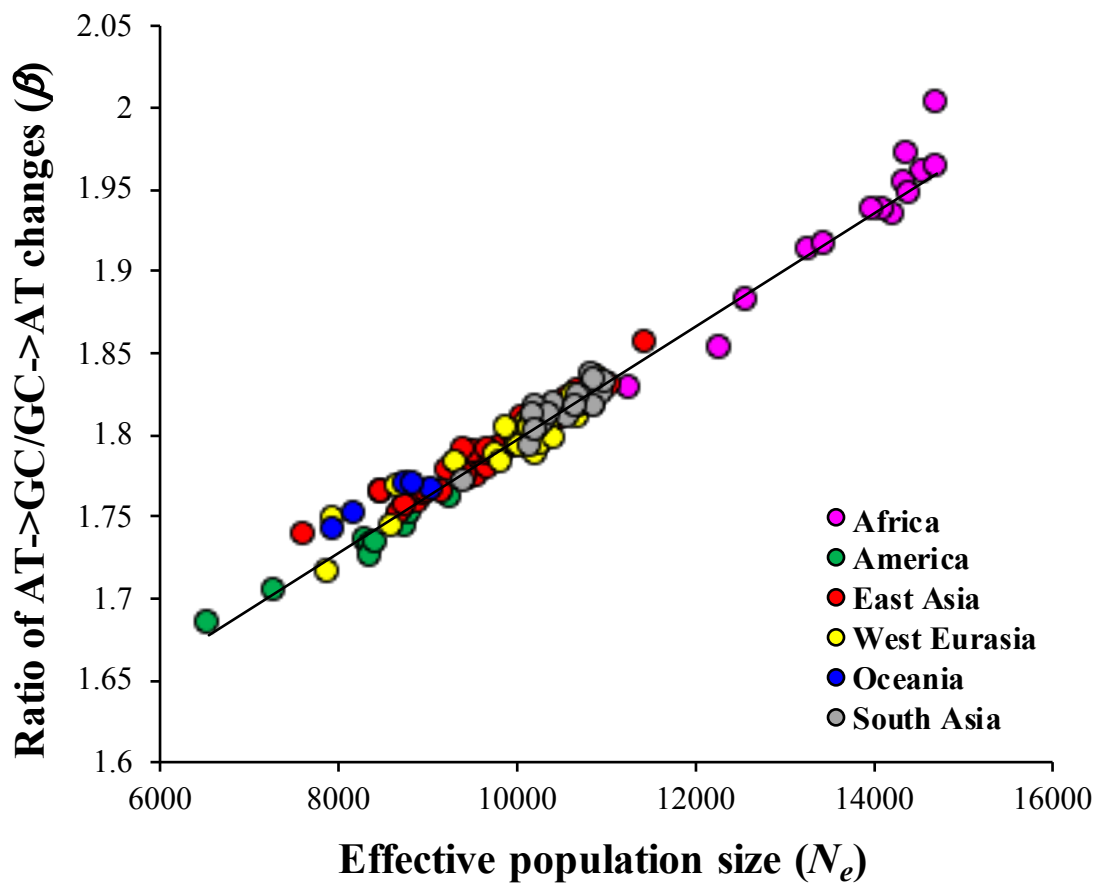


Figure S5