

Deleterious Mutation Burden and its Association with Complex Traits in Sorghum

Ravi Valluru, Elodie E. Gazave, Samuel B. Fernandes, John Ferguson, Roberto Lozano,
Pradeep, Tao Zuo, Patrick J. Brown, Andrew D.B. Leakey, Michael A. Gore, Edward S. Buckler,
Nonoy Bandillo

Supplementary Information

Fig. S1

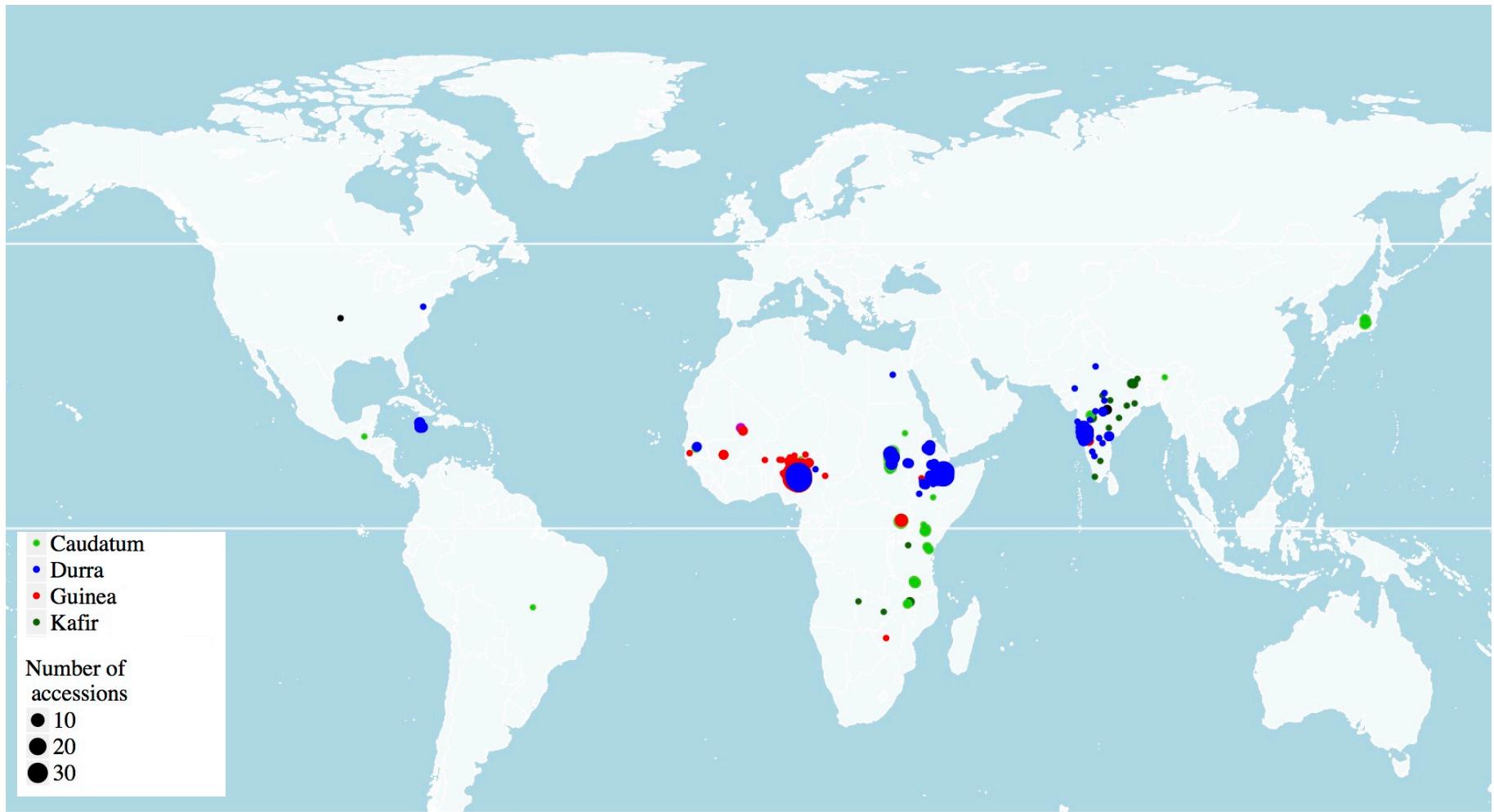


Fig S1 The geographical distribution of diverse sorghum lines (229) used in the study.

Fig. S2

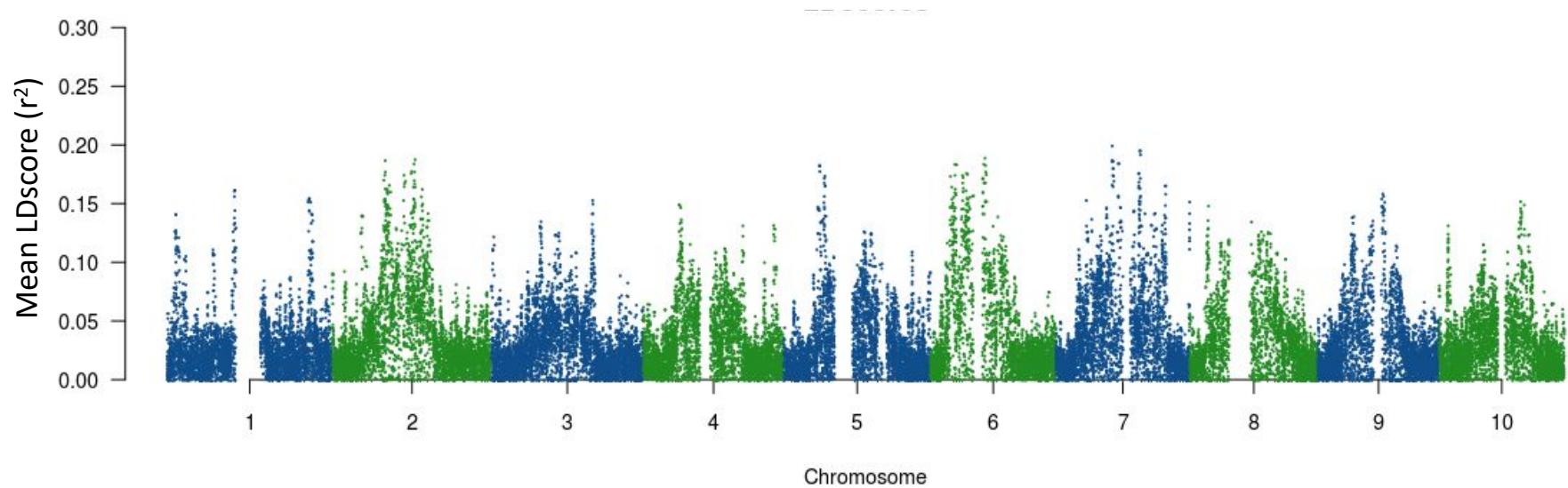


Fig S2 Mean linkage disequilibrium (LD) scores estimated for all chromosomes.

Fig. S3

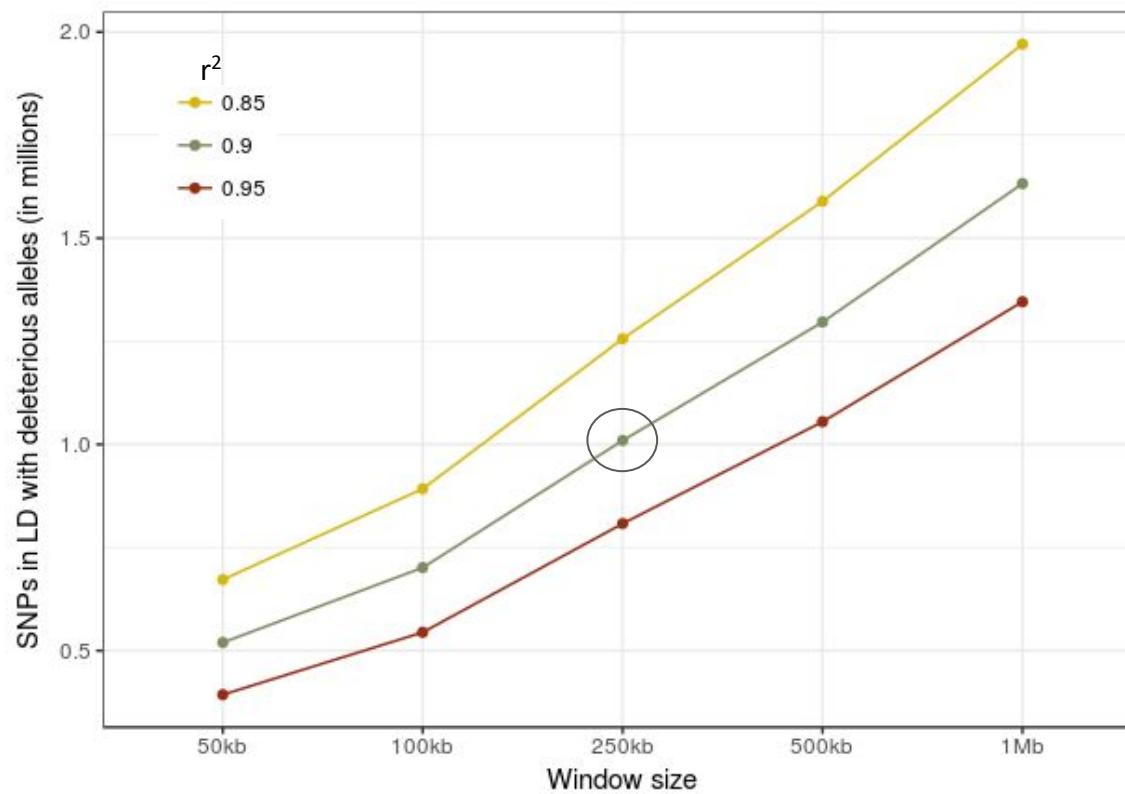


Fig S3 The number of single-nucleotide polymorphisms (SNPs) estimated under different parameters of window size and r^2 .

Fig. S4

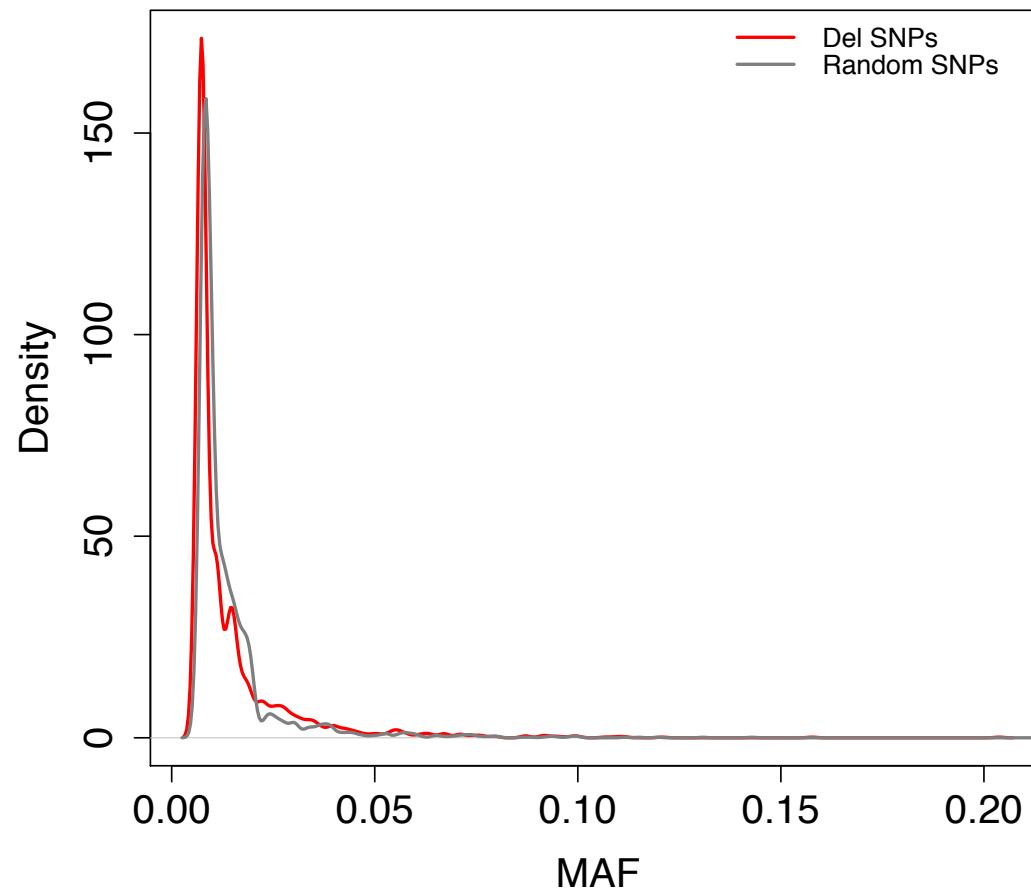


Fig S4 Allele frequency distribution comparison for high conserved deleterious variants (HGERP_{DEL-SNPs}, red) and nondeleterious variants (grey).

Fig. S5

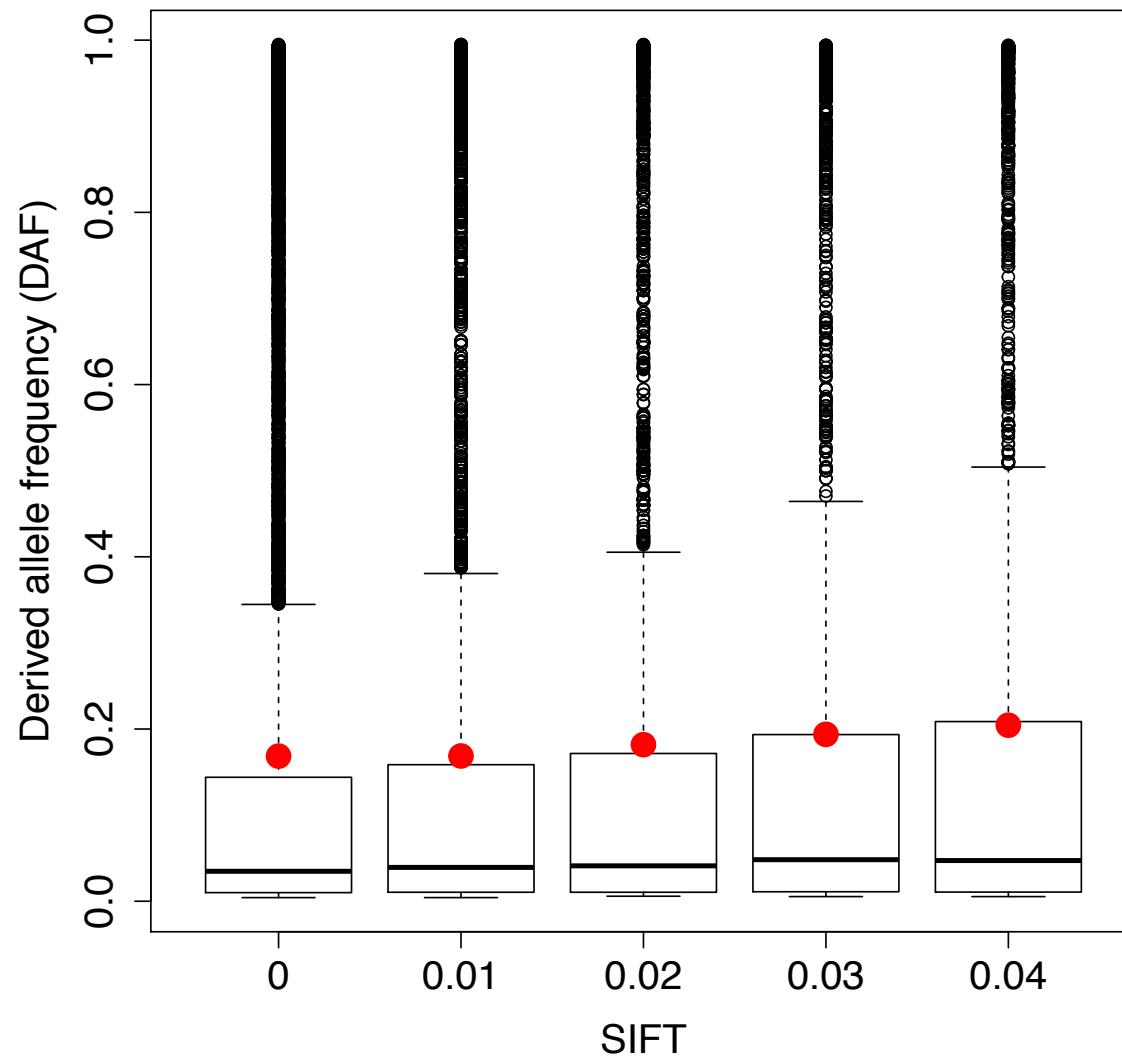


Fig S5 Derived allele frequency (DAF) association with Sorting Intolerant From Tolerant (SIFT) scores, Derived allele was defined as a minor allele from a multi-species sequence alignments. SIFT was estimated using Vaser et al. (2016).

Fig. S6

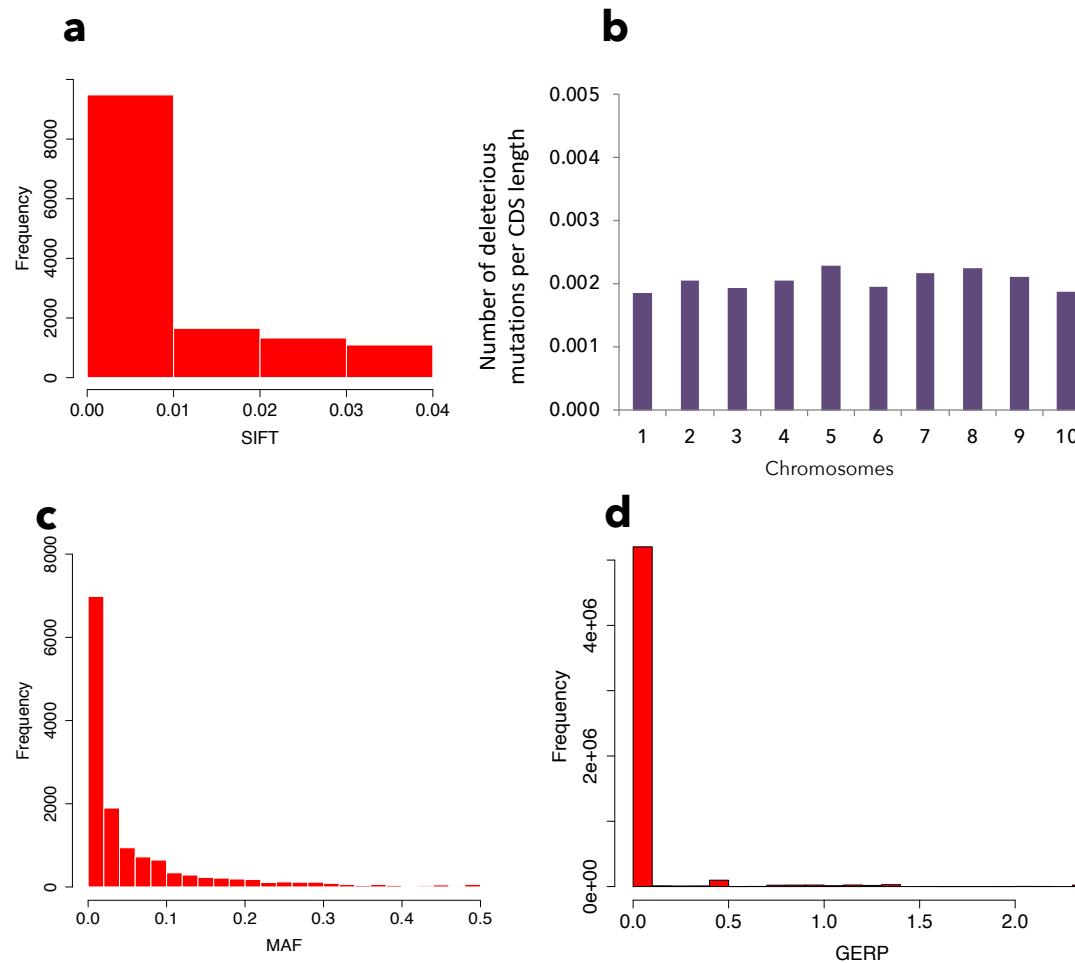


Fig S6 Frequency distributions of highly conserved deleterious mutations ($\text{SIFT} < 0.05$ and $\text{GERP} > 2$). Sorting Intolerant From Tolerant (SIFT) distributions of (a) all deleterious mutations, (b) number of deleterious mutations estimated per coding regions in all chromosomes, (c) minor allele frequency distribution of deleterious mutations, and (d) GERP distributions of deleterious mutations.

Fig. S7

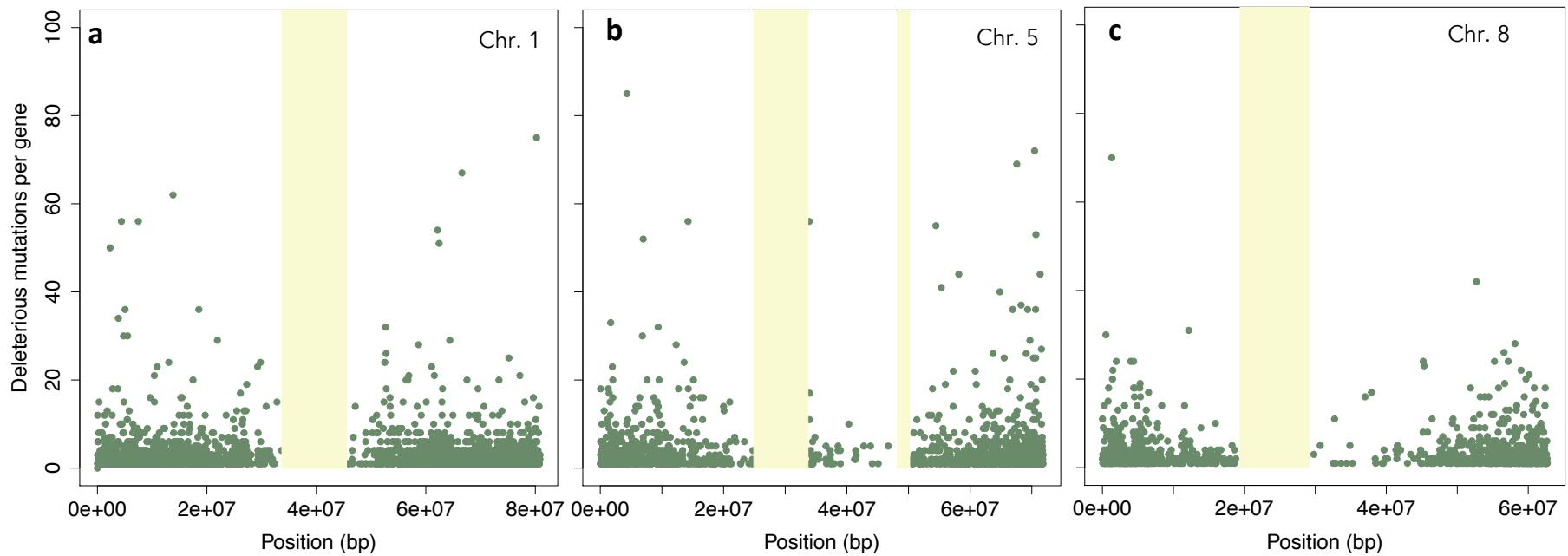


Fig S7 Gene deleterious mutations distribution in chromosomes 1 (a), 5 (b), and 8 (c). The yellow color vertical bar indicates a centromeric regions showing absence of genes or deleterious mutations.

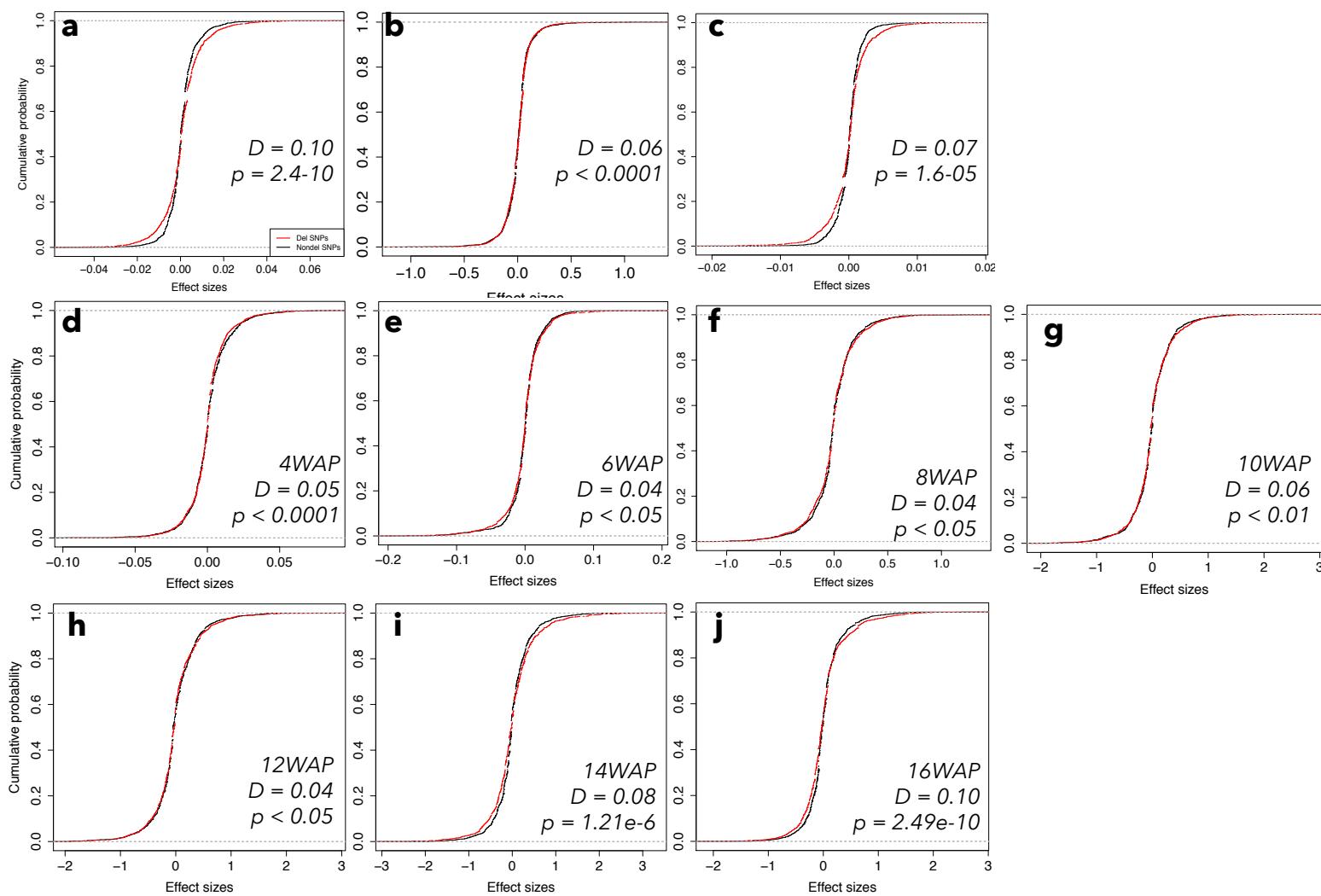
Fig S8

Fig S8 Empirical cumulative distributions (ECD) of the effect sizes for both high conserved deleterious variants ($\text{HGERP}_{\text{DEL-SNPs}}$) and nondeleterious variants for four phenotypic traits, biomass (a), specific leaf area (SLA, b), tissue starch content (TSC, c), and plant height 4 (d), 6 (e), 8 (f), 10 (g), 12 (h), 14 (i), and 16 (j) WAP, following a two-sample Kolmogorov-Smirnov (KS) test.

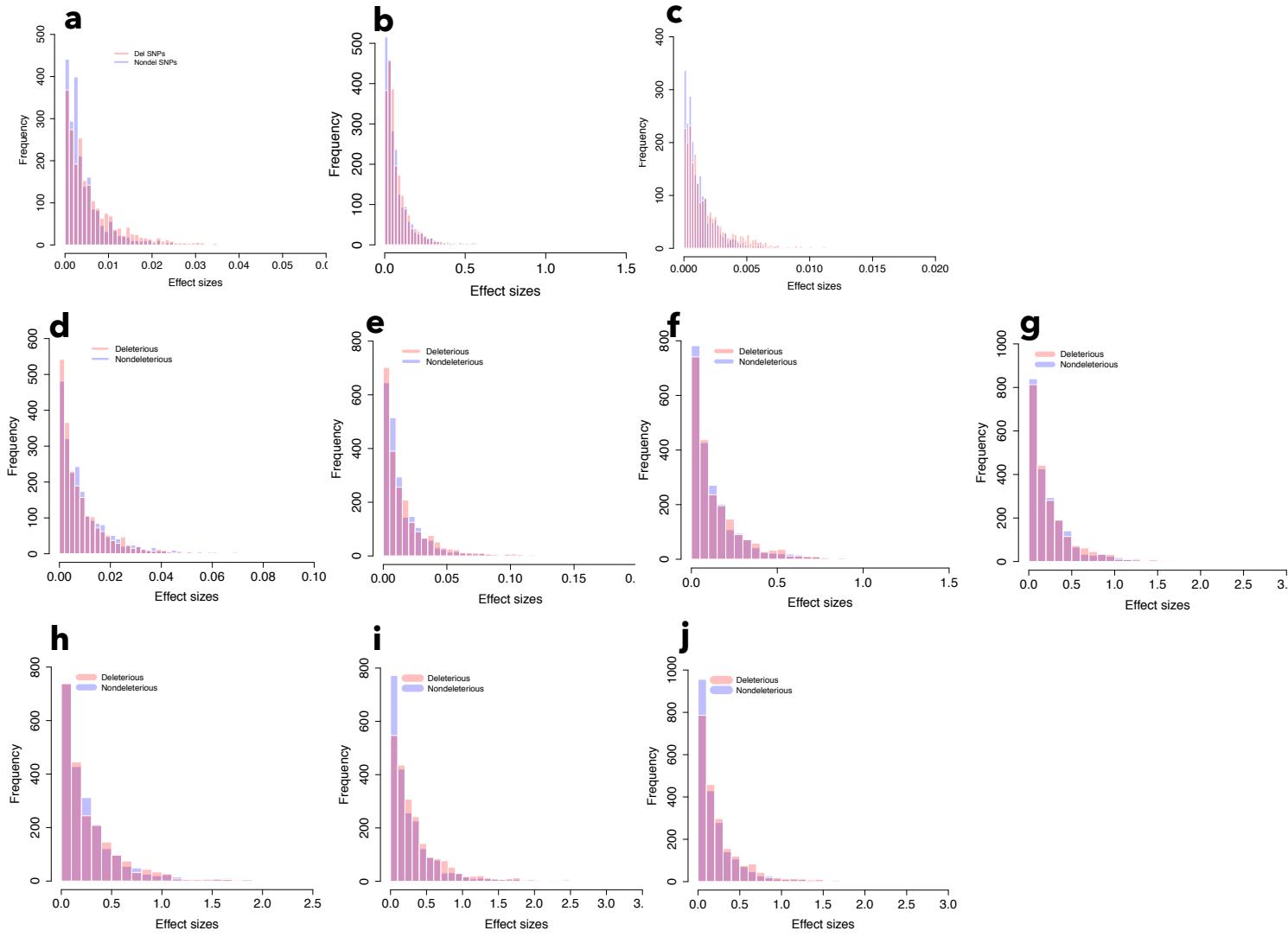
Fig S9

Fig S9 Folded distributions of the effect sizes for both high conserved deleterious variants ($HGERP_{DEL-SNPs}$) and nondeleterious variants for four phenotypic traits, biomass (a), specific leaf area (SLA, b), tissue starch content (TSC, c), and plant height 4 (d), 6 (e), 8 (f), 10 (g), 12 (h), 14 (i), and 16 (j) WAP.

Fig. S10

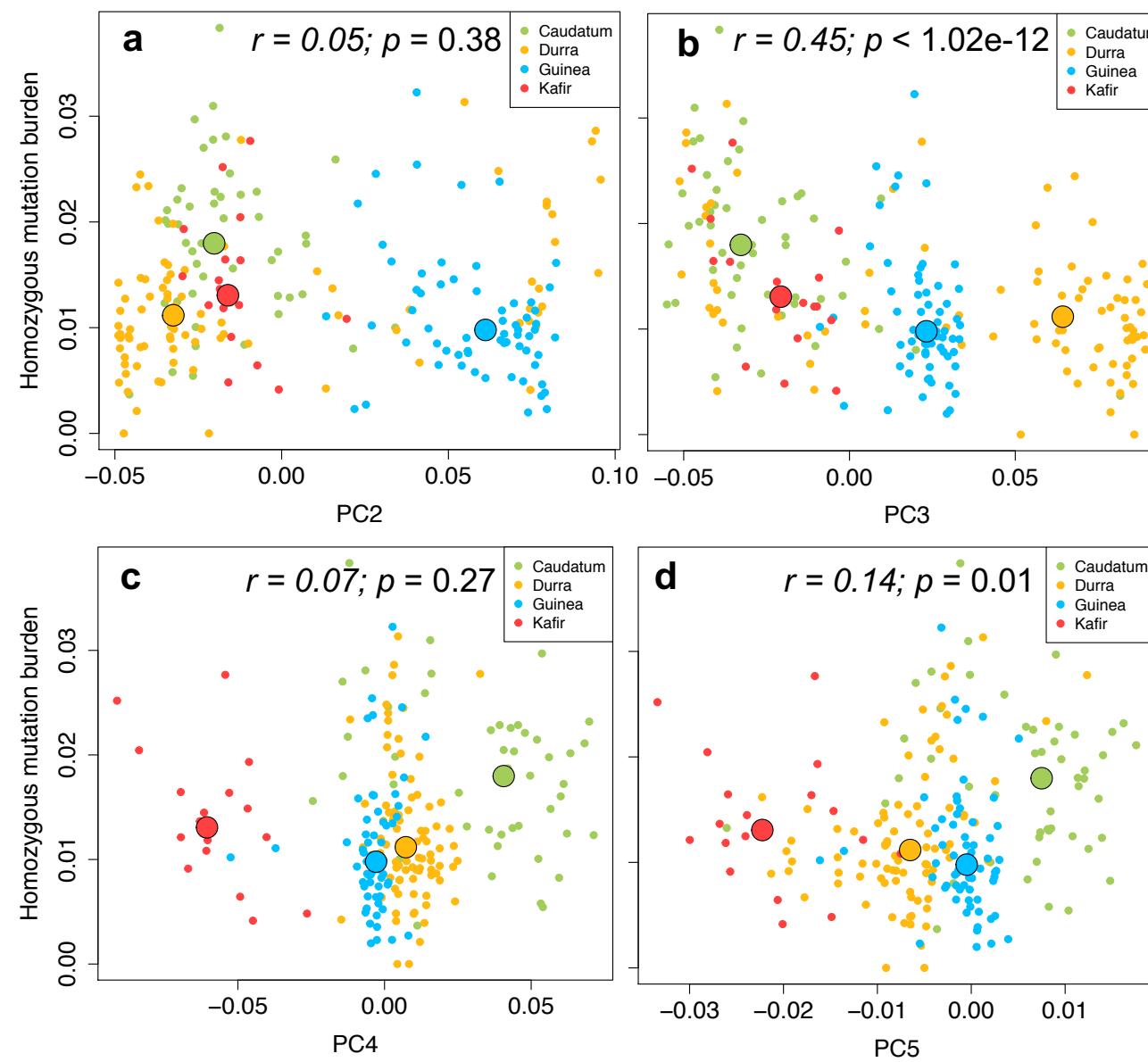


Fig S10 Scatterplots of the relationship between homozygous mutation burden and genetic principal components (PC2-5).

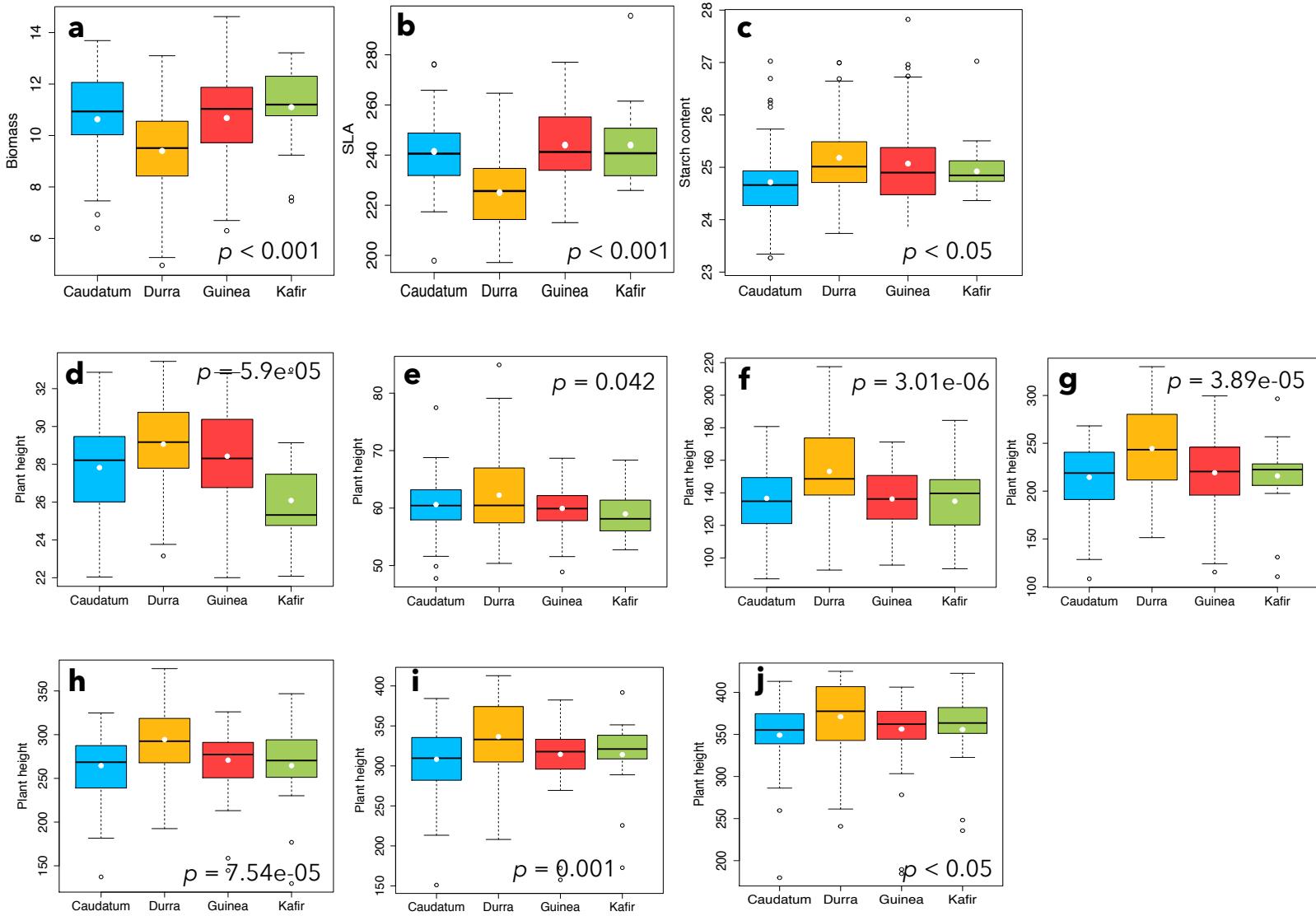
Fig S11

Fig S11 Boxplots of phenotypic data for biomass (a), specific leaf area (SLA, b), tissue starch content (TSC, c), and plant height 4 (d), 6 (e), 8 (f), 10 (g), 12 (h), 14 (i), and 16 (j) WAP under different racial groups of sorghum.

Fig S12

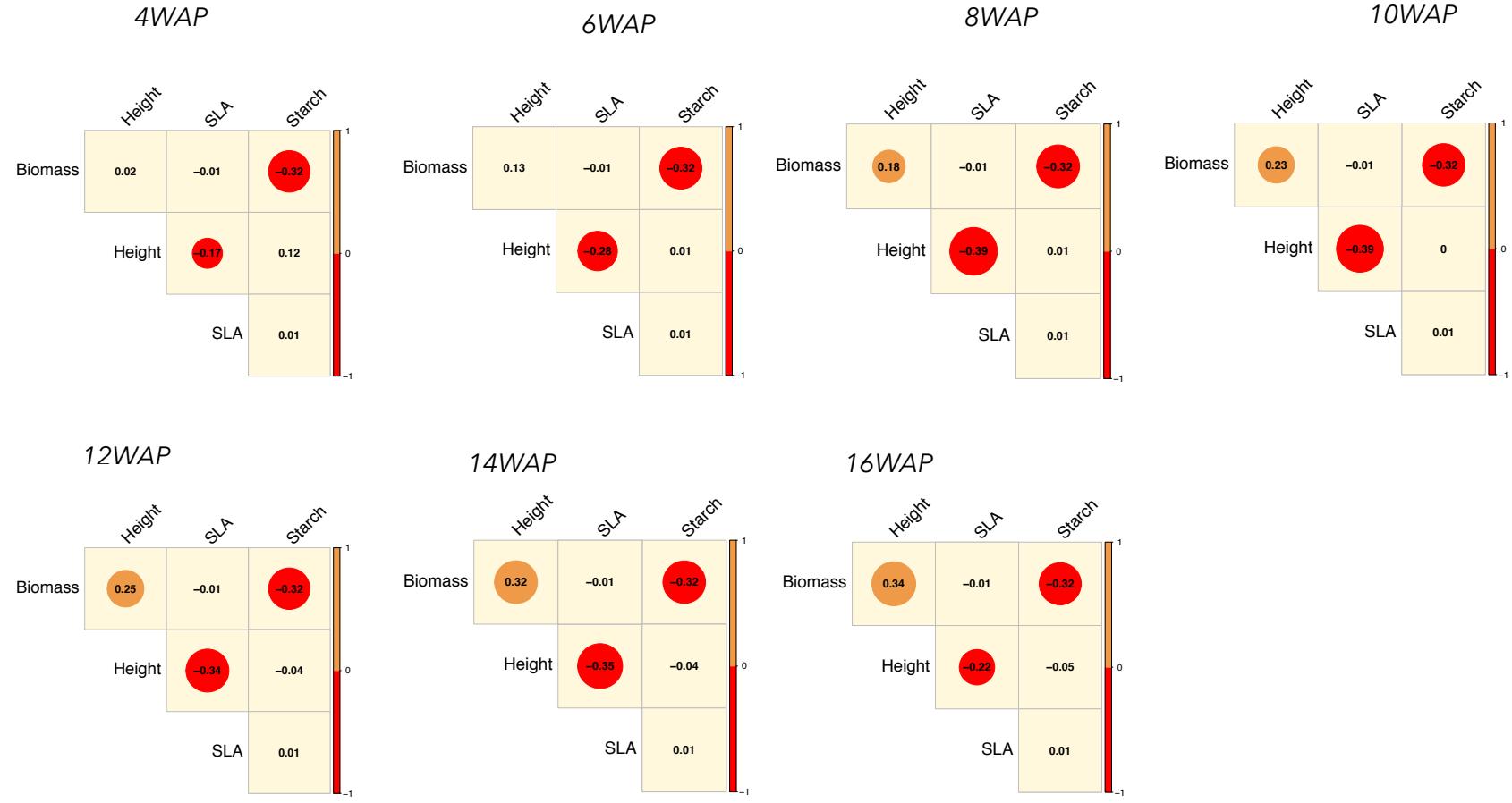


Fig S12 Correlations among traits either across all subpopulations using plant height 4, 6, 8, 10, 12, 14, and 16 WAP. All circles around values indicates significance at $p < 0.05$. Orange circles indicate positive correlation while red circle represents a negative correlation.

Fig S13



Fig S13 Heritability estimates for all traits using a two-kernel model in non-flowered lines.

Table. S1

Table S1 Slopes estimated between mutation burden and phenotypic traits using simple linear regression and grouped regression.

| Phenotype | Across all races | | | Grouped regression | | |
|------------------|-------------------------|----------------------|----------|---------------------------|----------------------|----------|
| | Estimate | r² | P | Estimate | r² | P |
| Biomass | -17.22 | 0.004 | 0.367 | -48.8 | 0.04 | 0.151 |
| PH (4WAP) | -32.64 | 0.008 | 0.199 | -58.28 | 0.03 | 0.058 |
| PH (6WAP) | -6.79 | 0 | 0.904 | -131.65 | 0.03 | 0.195 |
| PH (8WAP) | -15.67 | 0 | 0.945 | -468.2 | 0.03 | 0.207 |
| PH (10WAP) | -213.45 | 0.001 | 0.598 | -1326.8 | 0.07 | 0.056 |
| PH (12 WAP) | -329.48 | 0.003 | 0.422 | -1301.5 | 0.06 | 0.084 |
| PH (14 WAP) | -472.26 | 0.005 | 0.283 | -1552.5 | 0.05 | 0.006** |
| PH (16 WAP) | -716.27 | 0.014 | 0.081 | -1592.1 | 0.07 | 0.002** |
| SLA | -237.98 | 0.009 | 0.148 | -166.09 | 0 | 0.520 |
| Starch | -11.6 | 0.01 | 0.153 | -46.92 | 0.16 | 0.102 |