

| Dataset | UCEs | Observed | | Random ¹ | Expected (bp) | | | P |
|---------|------------|----------|-------|----------------------------|---------------|-------|-------|--------|
| | | N | bp | | Avg | StDv | Min | |
| CNVs | Combined | 13 | 3,191 | Genome ² | 13,548 | 1,928 | 4,143 | < 2E-9 |
| | Exonic | 13 | 3,191 | Genome | 2,463 | 792 | 0 | 0.8229 |
| | | | | Exons ³ | 3,409 | 923 | 0 | 0.4239 |
| | | | | GO exons ⁴ | 3,295 | 907 | 0 | 0.4728 |
| | | | | | | | | |
| | Intronic | 0 | 0 | Genome | 4,834 | 1,201 | 439 | < 1E-6 |
| | | | | Introns ³ | 2,695 | 911 | 0 | 7E-5 |
| | | | | GO introns ⁴ | 2,111 | 807 | 0 | 0.0005 |
| | Intergenic | 0 | 0 | Genome | 6,248 | 1,284 | 936 | < 1E-6 |
| | | | | Intergenic ³ | 7,307 | 1,385 | 1,694 | < 1E-6 |
| | | | | GO intergenic ⁴ | 4,281 | 1,074 | 201 | < 1E-6 |
| | Combined | 4 | 1,102 | Genome | 4,942 | 1,188 | 206 | 4E-5 |
| | | | | Genome | 898 | 488 | 0 | 0.3163 |
| | | | | Exons | 1,185 | 559 | 0 | 0.1589 |
| | | | | GO exons | 968 | 505 | 0 | 0.2683 |
| | Intronic | 0 | 0 | Genome | 1,762 | 741 | 0 | 0.0019 |
| | | | | Introns | 1,454 | 674 | 0 | 0.0057 |
| | | | | GO introns | 1,644 | 715 | 0 | 0.0029 |
| | Intergenic | 2 | 456 | Genome | 2,279 | 792 | 0 | 0.0031 |
| | | | | Intergenic | 2,465 | 823 | 0 | 0.0016 |
| | | | | GO intergenic | 1,801 | 705 | 0 | 0.0139 |

Supplementary Table 1 Intronic and intergenic^{tr} (but not exonic) UCEs are depleted among SDs and CNVs. Overlaps of exonic, intronic and intergenic^{tr} UCEs with pooled SDs and CNVs (excluding those of Sebat et al.¹⁶) are shown. Only intronic and intergenic^{tr} UCEs show significant depletion, even in comparison with corresponding conserved regions of genes with similar functional annotations. ¹Set from which random sequences were drawn: ²entire genome, ³all exons, introns or intergenic^{tr} regions, or ⁴exons, introns or intergenic^{tr} regions of genes^{tr} whose Gene Ontology (GO) annotations were the same as those containing UCEs (for exonic or intronic UCEs) or flanking UCEs (for intergenic^{tr} UCEs; Supplementary Methods).