Table S1. Control CNV datasets used to construct the dbVAR control dataset.

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
|  | **Conrad et al (2010) Hapmap** | **Conrad et al (2010) Discovery** | **Park et al (2010)** | **Perry et al (2008)** | **de Smith et al (2007)** |
| Total Genes Mapped to CNV regions | 781 | 373 | 815 | 1212 | 883 |
| StMA Genes | 1 | 1 | 1 | 2 | 4 |
| StMA/1000 CNV Genes | 1.3 | 2.7 | 1.2 | 1.7 | 4.5 |
|  |  |  |  |  |  |
| CNV region total size (Mb) | 50.83 | 25.66 | 35.45 | 75.61 | 69.93 |
| CNV region mean size (Mb) | 0.28 | 0.29 | 0.22 | 0.41 | 0.55 |
| Number of CNV regions | 182 | 89 | 161 | 184 | 128 |
| CNVs containing StMA | 1 (0.5%) | 1 (1.1%) | 1 (0.6%) | 2 (1.1%) | 4 (3.1%) |
|  |  |  |  |  |  |
| Genes/Mb CNV region | 15.4 | 14.5 | 23.0 | 16.0 | 12.6 |
| StMA Genes/Mb CNV region | 0.020 | 0.039 | 0.028 | 0.026 | 0.057 |

Total gene content and StMA gene occurrence are shown for each data set. The Conrad et al (2010) control study consisted of two CNV lists – a CNV discovery dataset and HapMap based dataset as shown in the table.