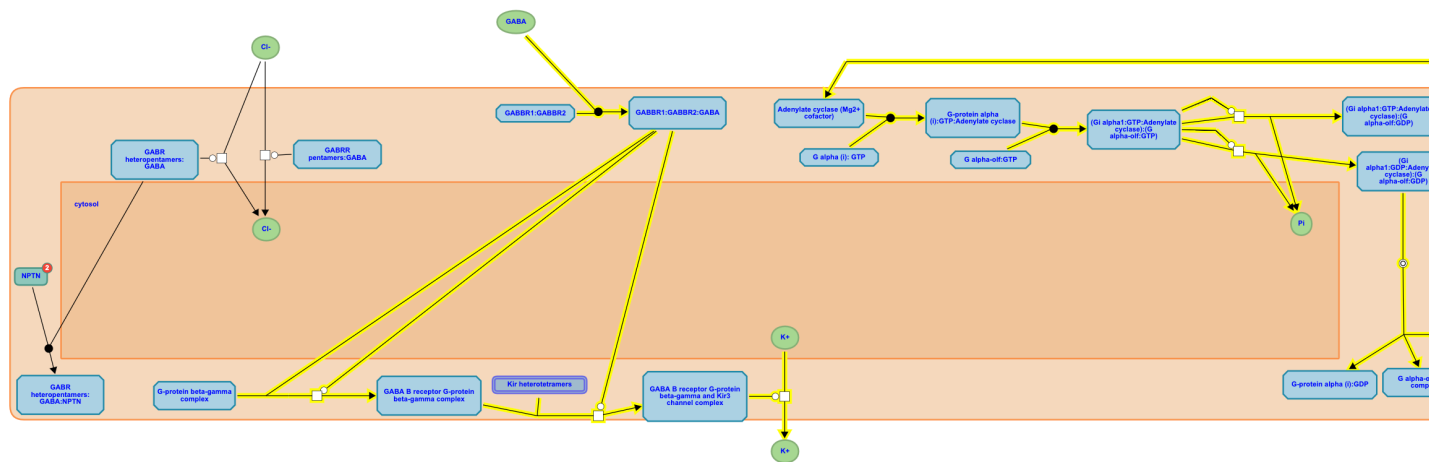


Supplementary material

Dosage effect of Copy Number Variation in Epilepsy and ten regions of the human brain

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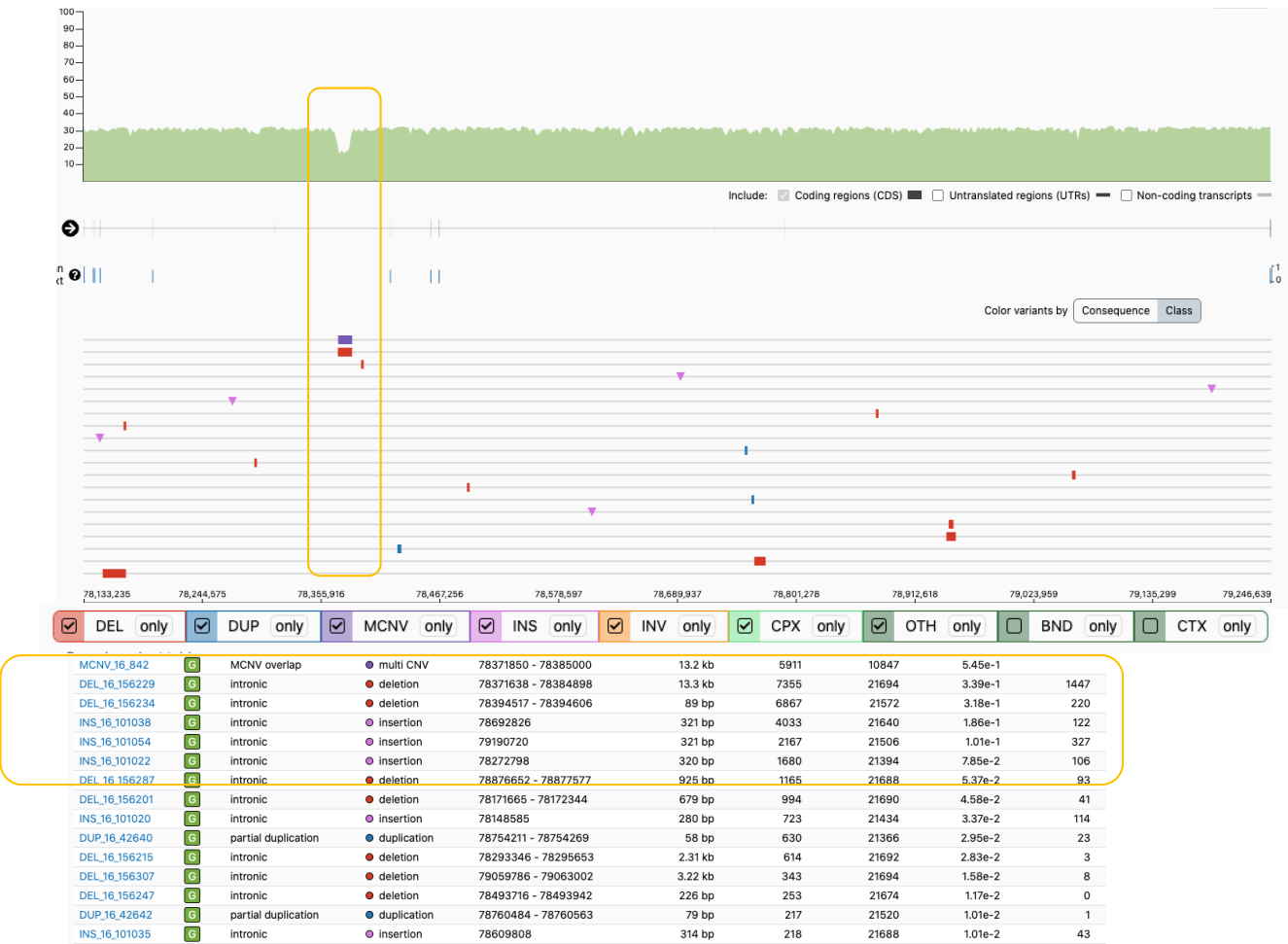
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Supplementary figure 2. GABRB3 interaction with GNB1. Reactome pathway details showing GABA B receptors interacting with G protein alpha-I, 0 beta and gamma subunits which leads to the activation of GIRK (KIR3) potassium channels.

GRCh37 / hg19

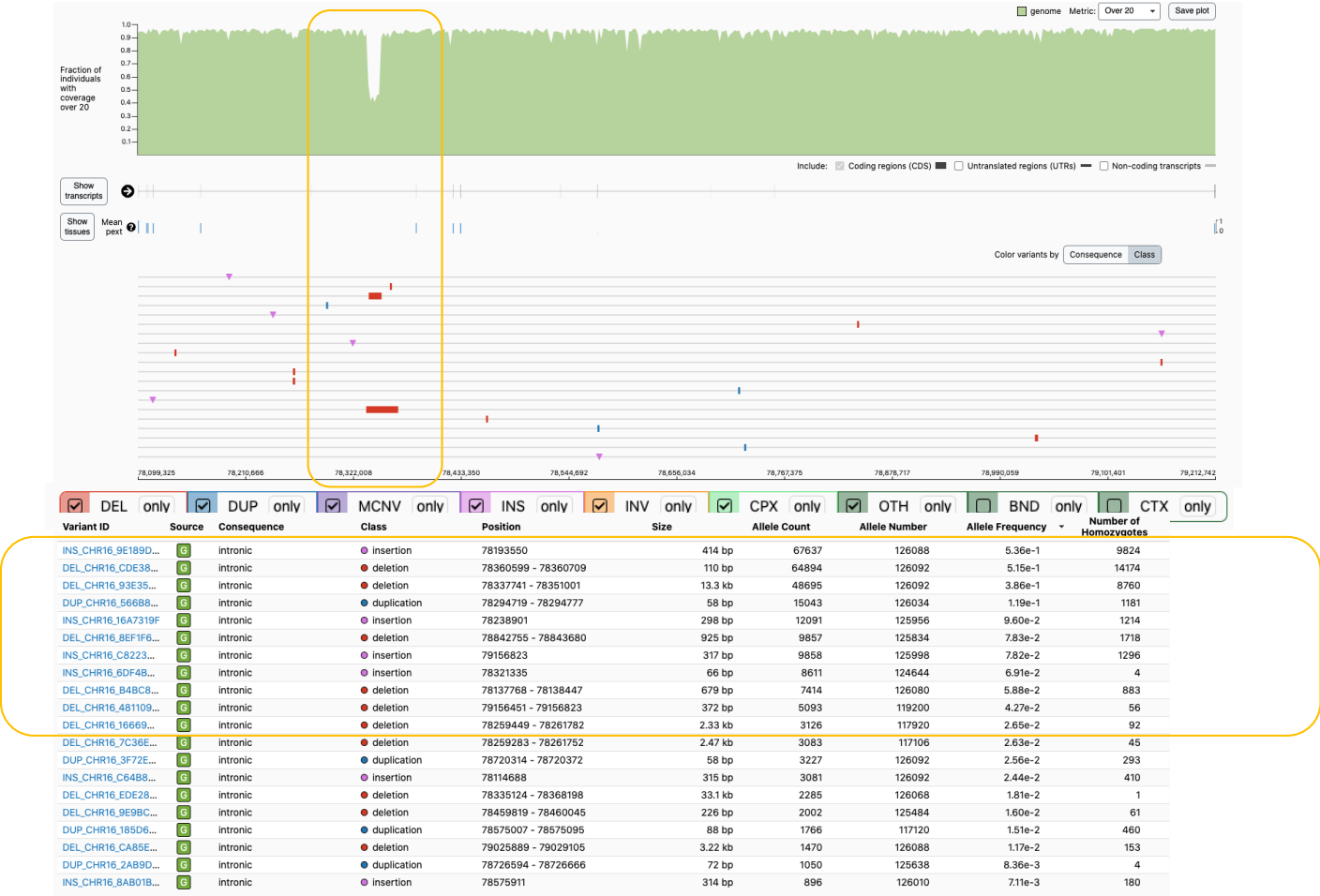
WVOX WW domain containing oxidoreductase



Supplementary figure 3. CNVs in WVOX. Figure shows the CNVs in the WVOX gene from the gnomAD database (GRCh37 / hg19)

GRCh38 / hg38

WWOX WW domain containing oxidoreductase



Supplementary figure 4. CNVs in WWOX. Figure shows the CNVs in the WWOX gene from the gnomAD database (GRCh38 / hg38)

Supplementary table legends

Supplementary table 1. Top results from all analyses. Table showing the top rank 1 results for SANAD, Australian cohort, Meta-analysis, UKBEC and NABEC studies.

Supplementary table 2. Clinvar database results for GNB1. CNV and SNP annotations for the GNB1 gene in the clinvar database.

Supplementary table 3. Association results in SANAD for chromosome 1p36. Association results for seizure phenotypes in SANAD in the chromosome 1p36 region.

Supplementary table 4. Association results for the WWOX gene. Association results for the drug-response phenotype in SANAD for CNV genotypes and LRR using univariate and multivariate models.

Supplementary table 5. NMF results for chromosome 1p36 in UKBEC. Frequency of exons in different gene programs uncovered through the NMF analysis of UKBEC dataset for NMF ranks 10 and 20 for gene expression matrices a) average all and b) full set.

Supplementary table 6. LRR results for SCN1B. Results for association of Log R Ratio with gene expression in different regions of the brain from the UKBEC dataset. Only three brain regions had p values < 1e-03 namely putamen, white matter and occipital cortex.

Supplementary table 7. Transcriptional sense of nearby genes. Table showing the transcriptional sense and the NMF analysis (rank 2 to 6) of genes located close to each other in the genome. Top exon refers to the most frequent exon in the NMF gene programs. NMF analysis was done for every gene separately with a 5 kilo base window around the gene boundary (see methods).