

Germline copy number variants and endometrial cancer risk

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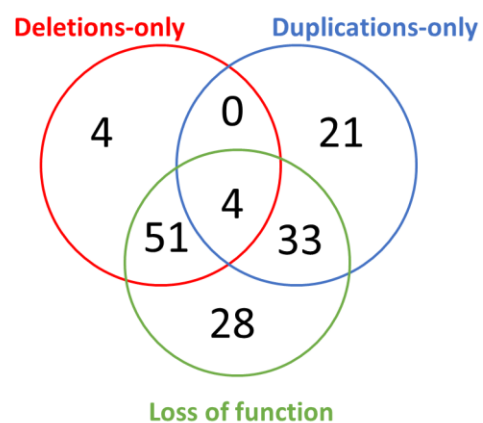
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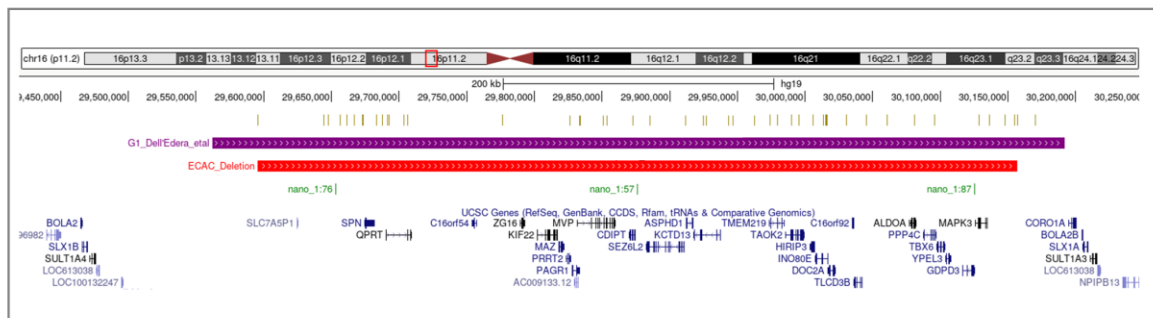
CNV-GWAS	Genes $p < 0.01$
Deletion-only	59
Duplication-only	58
Loss of function	116



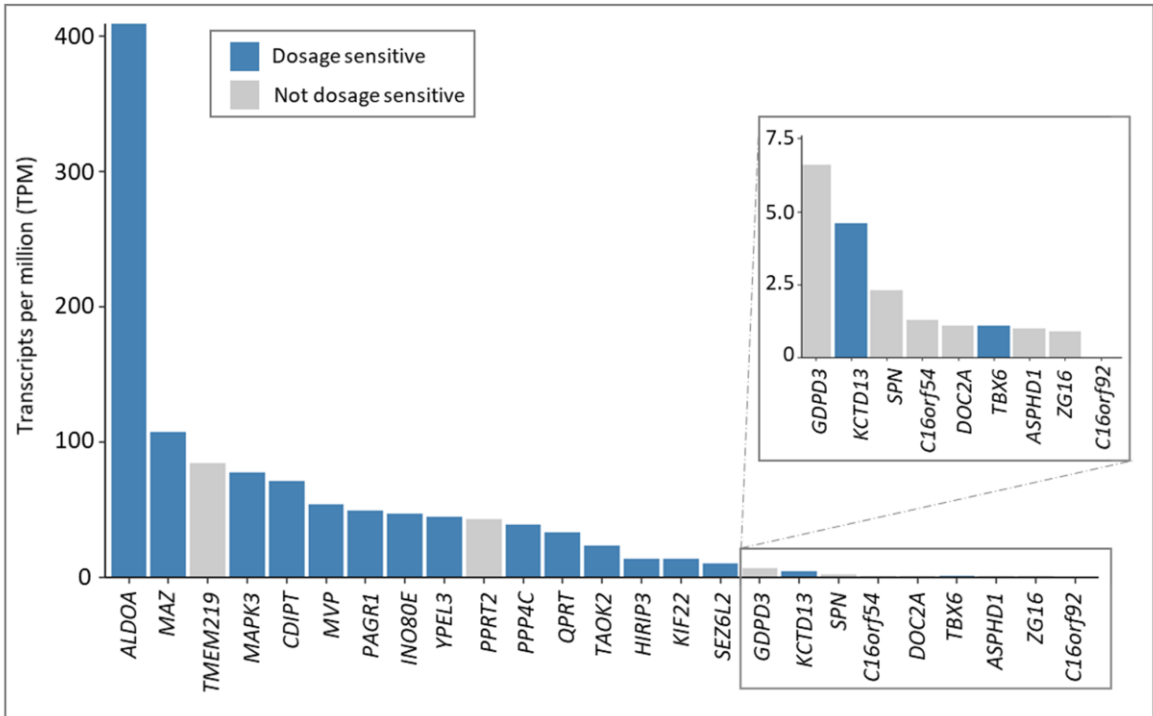
Supplementary Figure 1.

Table shows number of genes with $p < 0.01$ from each of the three CNV-GWAS. Venn diagram shows the concordance between candidate genes.

A)



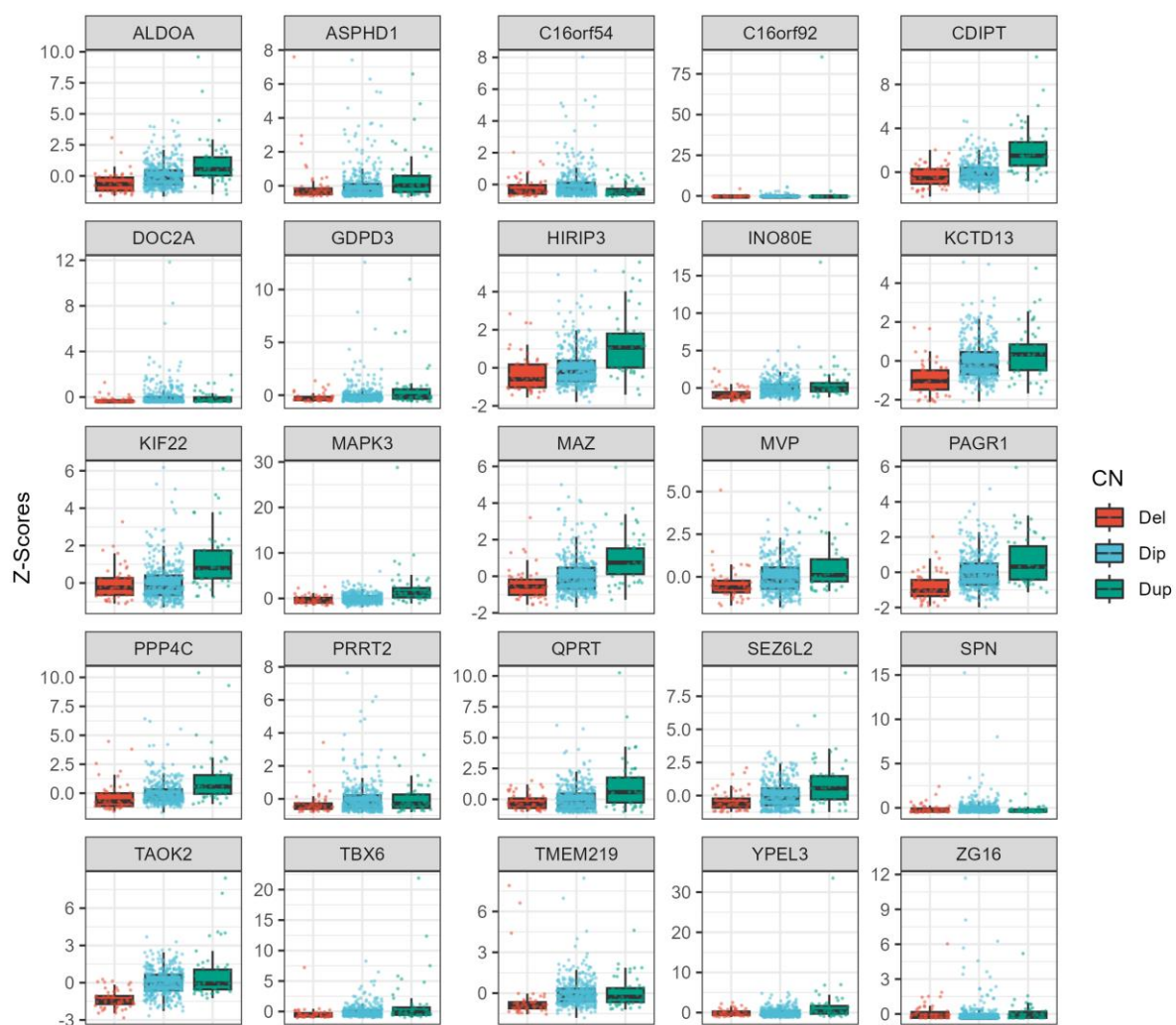
B)



Supplementary Figure 2.

A) Germline deletions represented by red bars, duplications by blue bars. NanoString probe location presented in dark green (track name: NanoString) Genes encompassed by the ECAC CNV (red) and published syndromic CNV (purple) are shown. Genomic positions according to hg19 genomic build.

B) Expression in normal endometrium tissue. Coloured bars represent those genes that show evidence ($p < 0.0001$) of dosage sensitivity in endometrial cancer tissue.



Supplementary Figure 3

Gene expression and dosage of 24 genes at the proximal 16p11.2 BP4-BP5 locus in 521 endometrial tumours from TCGA. Sample are grouped based on copy number status. Del, deletion (red); Dip, diploid (blue); Dup, duplication (green).