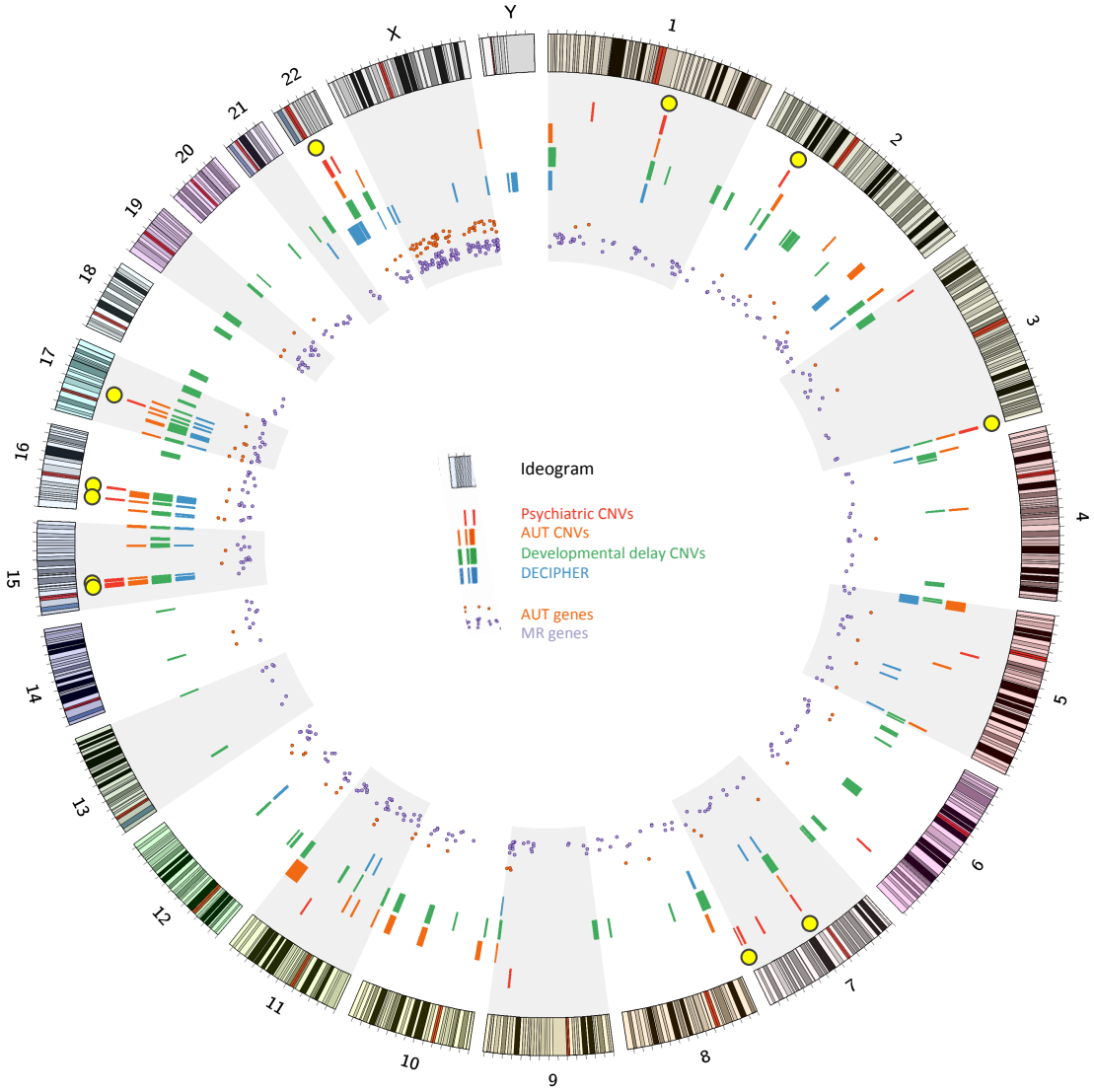
Figure S3: Variants of strong effect on psychiatric disorders



All of these variants are uncommon or rare, and have high genotypic relative risks (ranging from ~5 to deterministic). The outer ring shows the human genome as an ideogram, from 1ptel to Yqtel clockwise from the top. Odd numbered chromosomes are shaded. Five sets of data are depicted. (a) SV with replicated associations for ADHD, ASD, and SCZ (dark red). (b) SV from a comprehensive review of syndromic forms of autism (dark orange).[1] (c) SV associated with developmental delay (dark green).[2] (d) SV from DECIPHER associated with intellectual impairment (dark blue). (e) The innermost ring shows genes identified for syndromic forms of ASD (dark orange)[1] and genes from reviews of the medical genetics of MR (purple).[13-15] SV included in ***Table 2*** are noted with yellow circles.

There are ~110 loci associated with a clinical presentation that can include ASD features,[1] >430 genes for MR,[3–5] and sizable numbers of structural variants associated with ASD features and/or MR.[2,6] We do not mean to imply that all loci in ***Figure S3*** can be considered Mendelian and deterministic, and, indeed, many of the ASD loci have incomplete/uncertain penetrance or

assessments that relied on clinical impression rather than formal structured review. However, for many loci in ***Figure S3***, the *prima facie* evidence for a causal association is strong (e.g., a deleterious exonic mutation in a brain-expressed gene with perfect correlation between genotype and a distinctive clinical phenotype correlation in a large pedigree).

References

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