



**Figure S4.** Non-standard alleles at the 7q11.23 A-CNV and their population distribution. A) Diagram of the most common form of the “partial” copy number variants. Top, the reference version of duplicon A with three full copies of the CNV followed by the stable partial copy. Bottom, an allele of duplicon A containing two full copies of the CNV, one new partial copy, two more full copies, and the stable partial copy. Full copies are represented with yellow arrows, the new interspersed partial copy is represented with a red arrow, and the stable partial copy is represented by an arrow filled with diagonal yellow lines. The grey bar indicates the critical region used for genotyping. B) Diagram of the most common form of the downstream variant allele. Top, the reference version of duplicon A with three full copies of the CNV. Bottom, the variant containing no full copies of the CNV, with a different label pattern than seen in the reference. The grey bar indicates the critical region used for genotyping. C) Population-based prevalence of the three classes of variants at this locus: containing only full copies of A-CNV, containing one or more partial copies, or containing the downstream variant. \*  $p < 0.005$ , pairwise Fisher’s exact test with Benjamini-Hochberg multiple testing correction. Labels on the bars show the number of times a configuration was detected in each population.