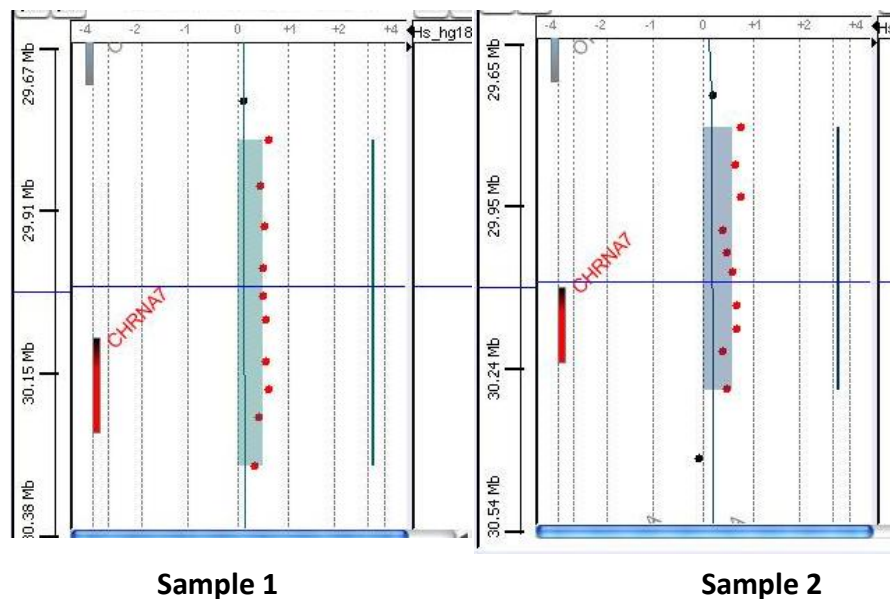
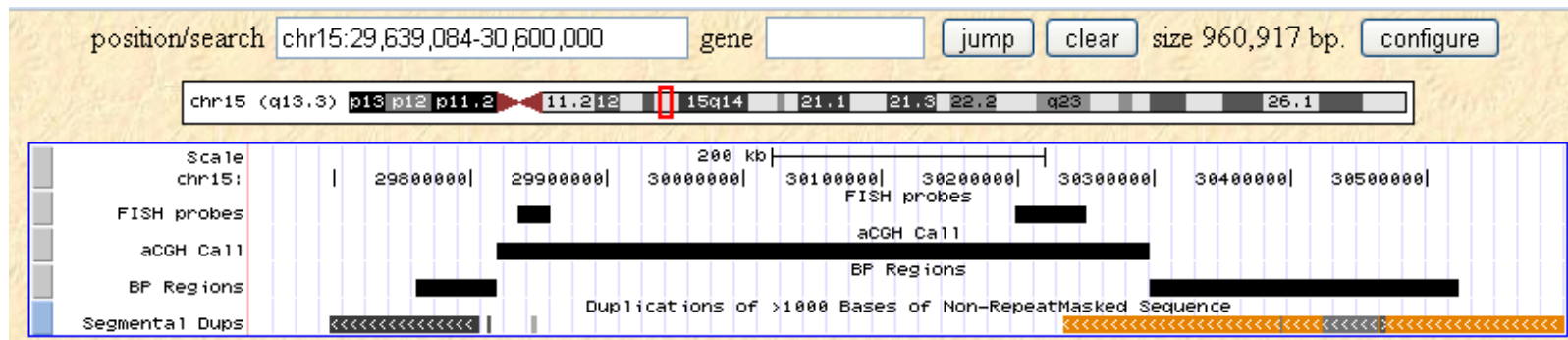


Supplemental Figure 1s. 6p25.3 (174,615-284,943) is a copy number variant region with a second locus at chromosome 16. The 6p25.3 probe was labeled in red/orange and the chromosome 6 pericentromeric region was labeled in green. a) metaphase cell from an individual with no chromosome 16 duplicated site, b) metaphase cell from an individual with a duplication on only one homolog 16 (arrow), and c) metaphase cell from an individual with a duplication on both homologs of chromosome 16 (arrows).

a



b



Supplemental Figure 2s. 15q13.3 duplication in two independent samples. a) Profiles of log<sub>2</sub> ratio data from array CGH on chromosome 15q13.3. The copy number gains called by ADM2 using DNA Analytics software (Agilent) are indicated by shaded bars, encompassing an identical set of 10 probes (red spots) spanning 479 kb in each individual. The red bar shows where the CHRNA7 gene is located. b) Genomic architecture surrounding the 15q13.3 duplication from the UCSC Browser. BP (breakpoint, second row from the bottom on the browser) regions marks the distance between the probe for which the aCGH call (third row from the bottom) was made and the neighboring probe. FISH probes (fourth row from the bottom on the browser) indicate where probes were designed to cover the 5' and 3' ends of the duplication.