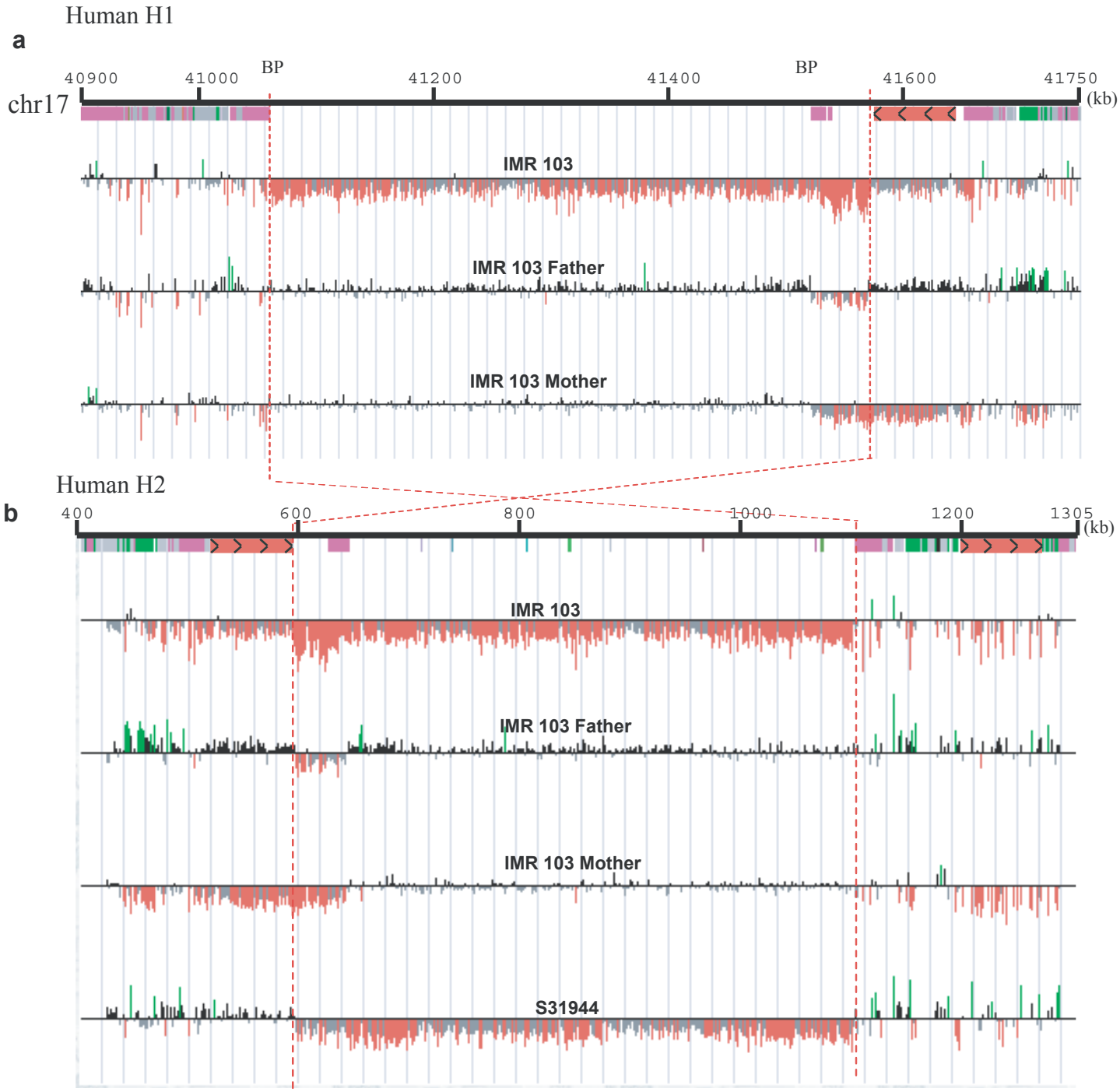

Supplementary Figure 2: Chr17q21.31 microdeletion breakpoints.
We remapped the oligonucleotides used microarray comparative genomic hybridization experiments (Sharp et al, 2006) to both the H1 and H2 haplotypes. Array CGH data are shown for parents and a child (IMR103) with mental retardation against both an a) H1 and b) H2 sequence haplotype. The microdeletion was shown to have occurred on the H2 chromosome of the father of IMR103. The pair of "H2-specific" duplications (red color with internal arrows) are shown with respect to the microdeletion breakpoints (red dashed lines). Precise localization of breakpoints is confounded by segmental duplications and copy-number polymorphism, but an examination another de novo microdeletions consistently places one of the breakpoints in or within 1 kbp of the H2-specific segmental duplication (e.g. patient S31944). Although we have fairly clear evidence that the microdeletion breakpoint occurs within proximal breakpoint near the "H2-specific" segmental duplication, we have less mapping precision at the distal end due to the CNP that exists between H2 coordinates 1103477-1197879. So even though the distal breakpoint signal ends abruptly by the distal duplication block, due to the CNP of that region the distal breakpoint could still map closer to the internal H2-specific duplicon (i.e. a reduction in copy would not necessarily be expected to show a detectable difference when compared to an H1/H1 haplotype reference).