

Segmental duplications

IMR338M

69/06

inv dup(15)

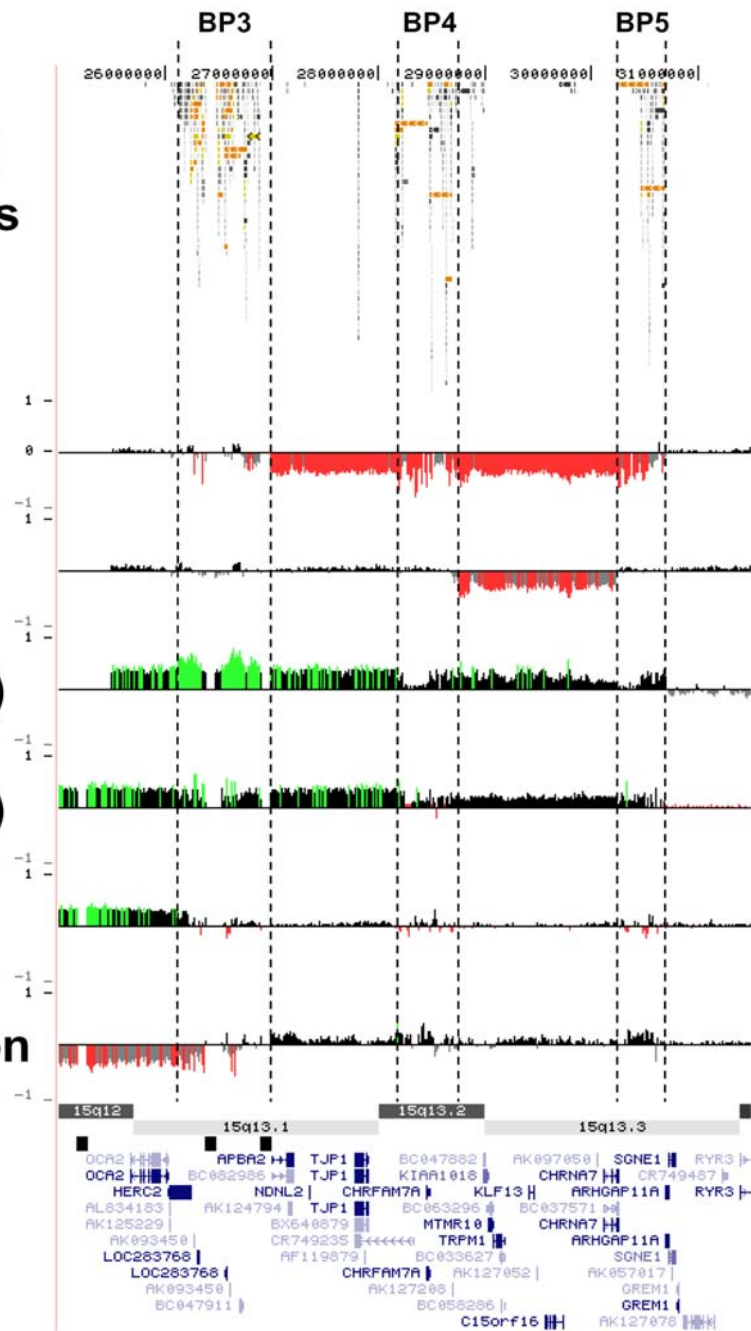
inv dup(15)

mar(15)

PWS deletion

Gaps

Genes



Supplementary Figure 6. Breakpoints of multiple different rearrangements of proximal 15q map to large blocks of segmental duplications at BP3, BP4, and BP5. Image shows the deletions identified in IMR338 (BP3-BP5) and 69/06 (BP4-BP5). Results obtained in two unrelated patients with inv dup(15) chromosomes show both are composed of two copies of the region 15cen-BP4 and a single copy of region BP4-BP5. This was subsequently confirmed by FISH (data not shown). We also tested one patient carrying a marker chromosome 15 (four copies of the region 15cen-BP3), and one patient with a class II Prader-Willi deletion (one copy of the region BP2-BP3), providing further evidence of recurrent chromosomal breakpoints in 15q. Image shows a 6.5 Mb region of 15q12-15q13.3 (chr15:25,000,001-31,500,000). For each individual, deviations of probe log₂ ratios from zero are depicted by grey/black bars, with those exceeding a threshold of 1.5 standard deviations from the mean probe ratio colored green and red to represent relative gains and losses, respectively. Tracks above the plot indicate segmental duplications (grey/yellow/orange bars representing duplications with 90-98%/98-99%/99-100% sequence identity, respectively).