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Supplemental Data

Characterization of a 8q21.11 Microdeletion Syndrome Associated with Intellectual Disability and a Recognizable Phenotype

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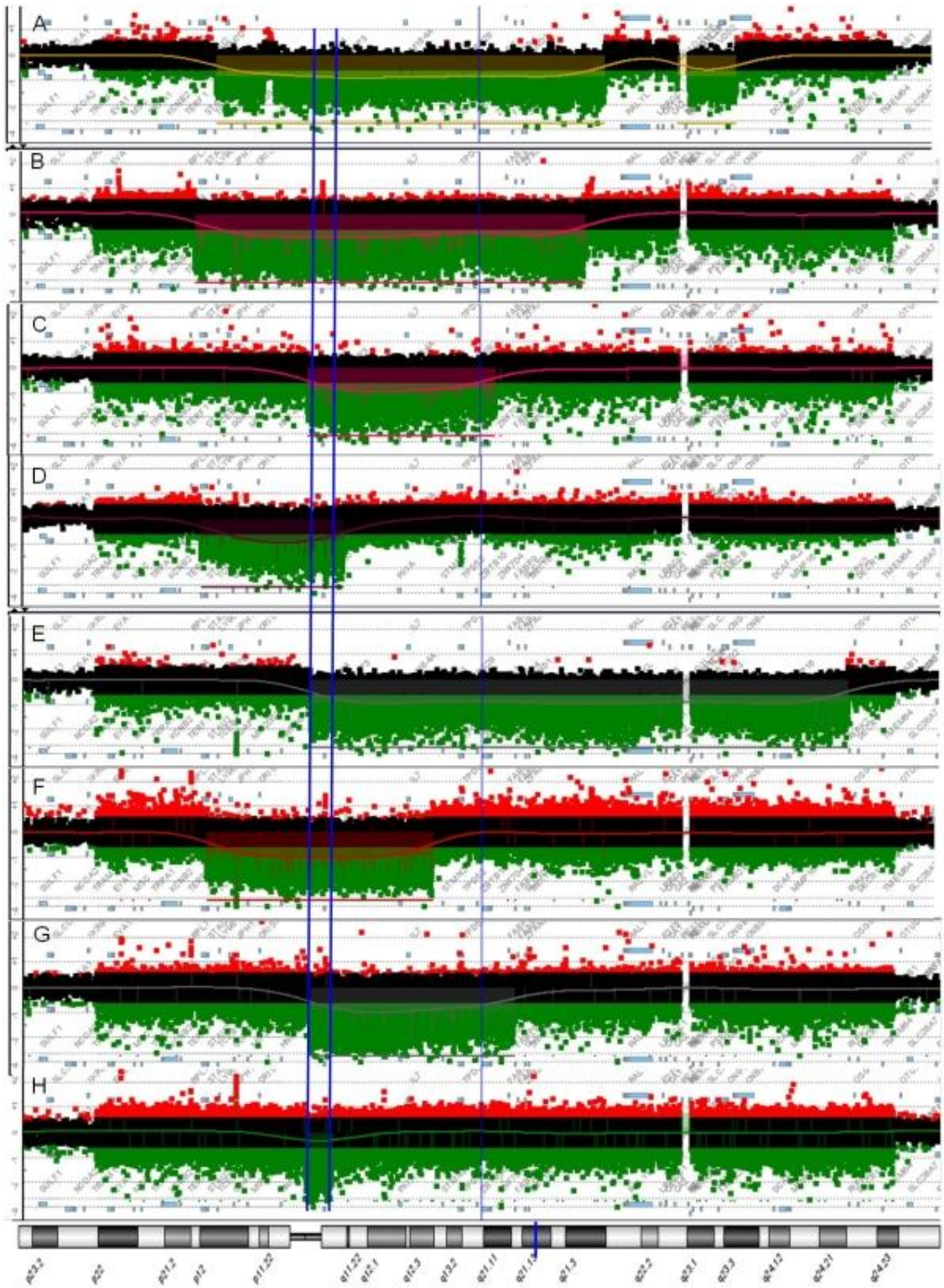

Figure S1. Array CGH profiles of all individuals with 8q21 microdeletions using high-density oligonucleotide arrays with mean density one probe per 140 bp showing a 20 Mb region of 8q13.3-q21.3, hg 19 chromosome 8:72000000-92000000. The array plots of individuals 1 to 8 show overlapping deletions in all eight individuals and define a minimal deleted region for this syndrome in hg 19 chromosome 8:77226464-77766239.

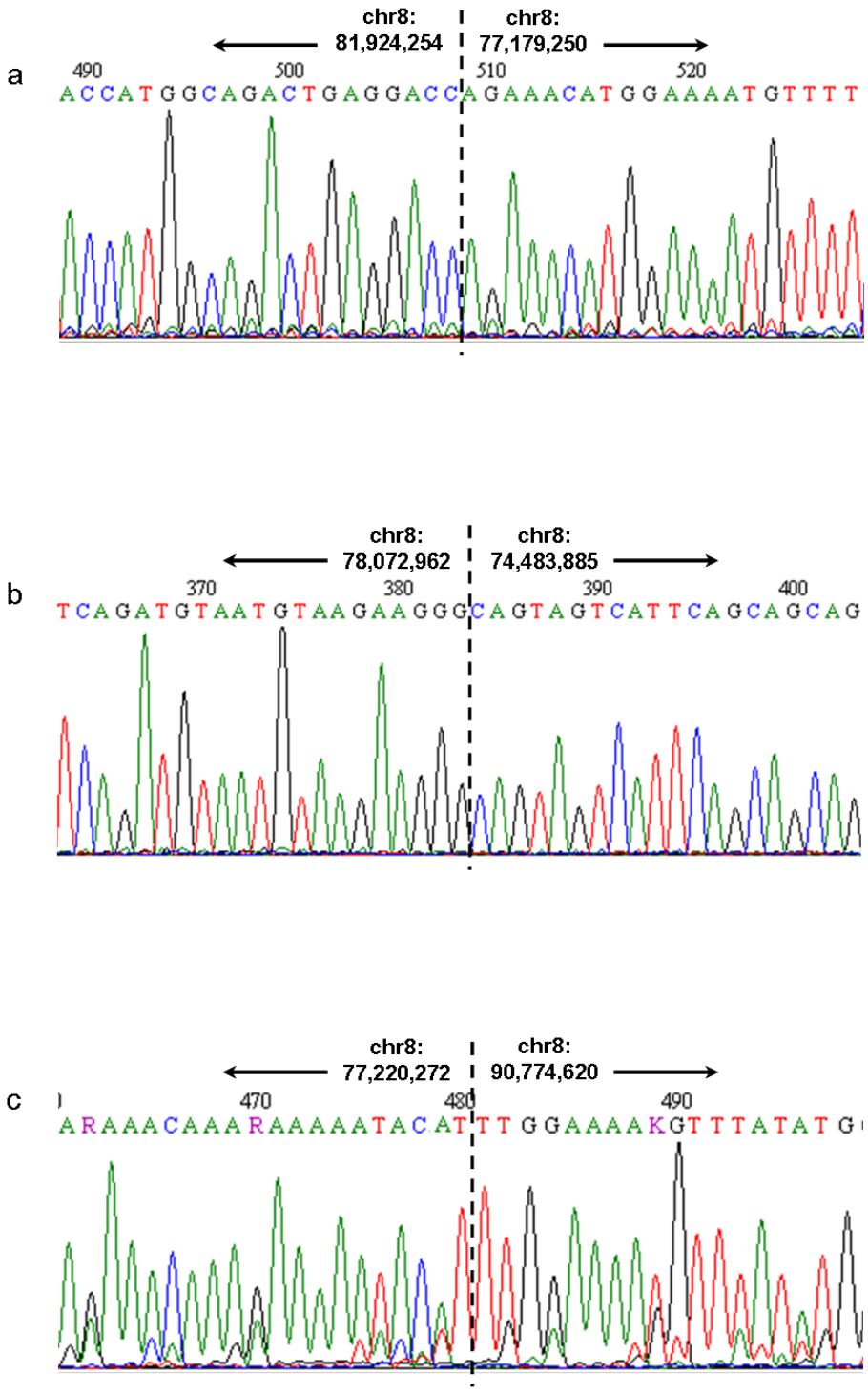

Figure S2. Breakpoint sequences of individuals 3 (a), 4 (b) and 5 (c). Panels a and b show reverse strands, while panel c shows forward strand. Breakpoints are indicated as dashed lines, with hg 19 coordinates of flanking nucleotides. Breakpoint junction sequencing analysis provided base pair resolution and showed the deletions spanned 4,745,004 bp in 8q21 in individual 3, hg 19 chromosome 8: 77179250-81924254; 3,589,077 bp in individual 4, hg 19 chromosome 8:74483885-78072962; and 13,554,348 bp in individual 5, hg 19 chromosome 8:77220272-90774620.