

SUPPLEMENTAL FIG. S1. Microsatellite genotyping of marker D15S128 within a stably inherited interstitial 15q duplication. Maternal genotypes (201/205/207 bp) confirm the presence of a 15q duplication. Paternal alleles are 201/205 bp. Genotypes present (201/201/207 bp) in the affected child are consistent with maternal transmission of the duplication allele (201/207 bp). In the affected child, the presence of two alleles at bin size 201 bp is reflected in the peak height ratio of allele 201 bp to allele 207 bp (2.85) compared to the peak height ratio of allele 201 bp to allele 207 bp in the mother (1.97). The difference in these peak height ratios indicates duplication of the 201 bp allele. The data shown here is representative of and consistent with additional genotyping (155CA1, 155CA2, D15S1002) data collected this family, demonstrating maternal transmission of an interstitial 15 q duplication. Additional unlabeled peaks represent stutter artifacts commonly seen in fragment length analysis.