Supplementary Figure 2. Parental origin and inversion analysis of the 17q21.31 deletion in the family of IMR103.

(A) PCR of a microsatellite (chr17:44344325-44344661, hg10), located within the 17q deletion region in the family of IMR103, shows the deletion occurred on the paternally derived chromosome. (B) Use of a diagnostic SNP marker to assay a common 900 kb inversion of 17q21.3 in the family of IMR103. The T>C SNP rs64965 is in linkage disequilibrium with the inversion (Stefansson et al. 2005), and alters the restriction pattern for Tsp509I. PCR and subsequent digestion with Tsp509I therefore allows the inversion state to be genotyped, showing that the father is a heterozygous carrier of the inversion, in which the recurrent deletion occurs. Similar results were gained from analysis of a second diagnostic SNP rs186547 (Stefansson et al. 2005). (C) PCR of a 238 bp deletion within the MAFT gene specific to the H2 inversion haplotype in the family of 338H5 (Stefansson et al. 2005). The mother is heterozygous carrier of the inversion, and the deletion occurred on the maternally derived chromosome. Although the inversion is present in ~20% of northern Europeans, these data are consistent with the notion that inversion of this region may be a predisposing factor to subsequent microdeletion, as has been observed for other recurrent genomic disorders (Gimelli et al. 2003, Osborne et al. 2001, Vissers et al. 2005). PCR primers are listed in Supplementary Table 6.