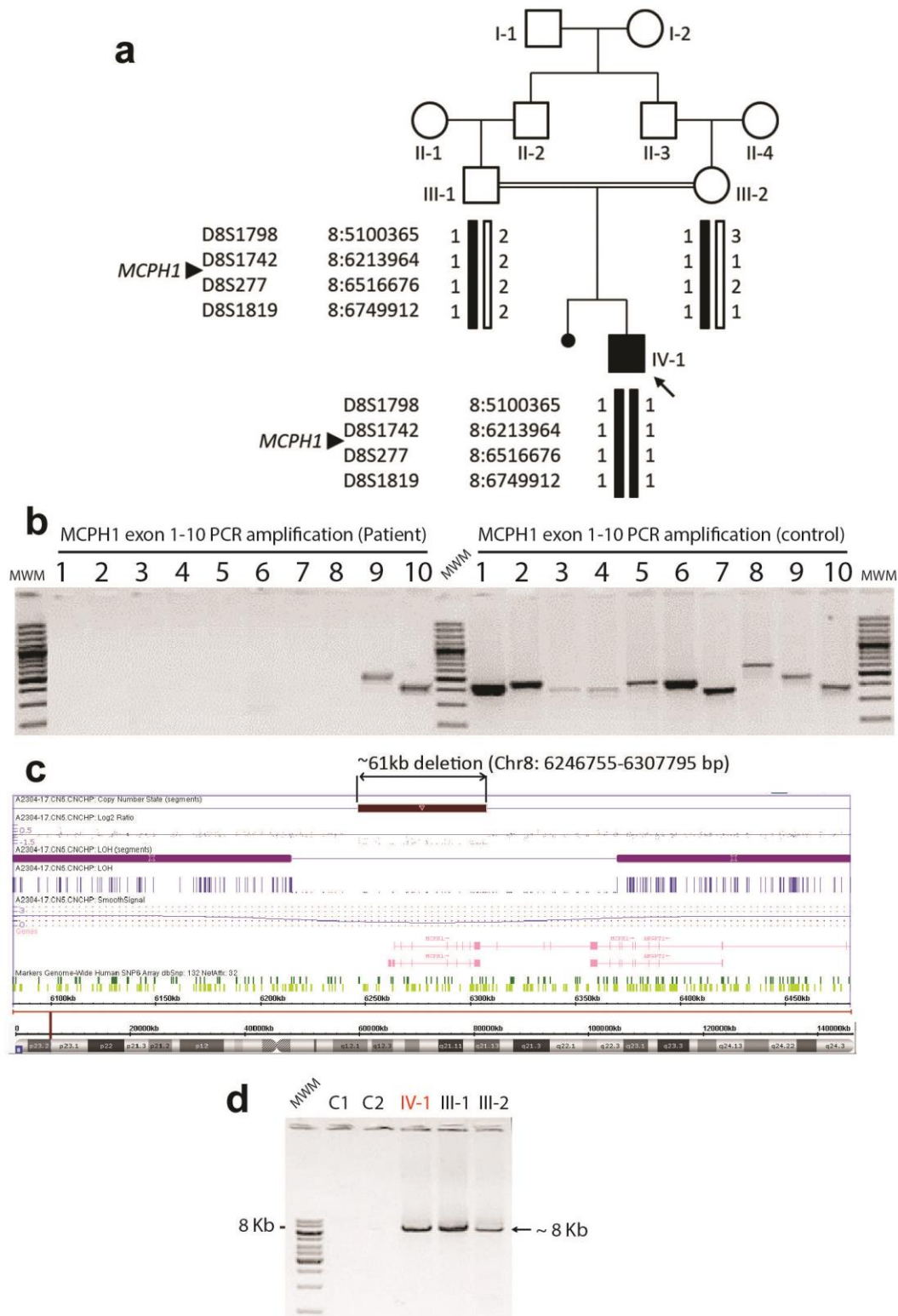


**Figure S1.** Genetic testing results. **(a)** Pedigree with sequence-tagged markers for the MCPH1 locus. **(b)** PCR amplification of MCPH1 exons 1-10 in proband (left) and healthy control (right). **(c)** SNP array dialog window showing a deletion of about 61kb. **(d)** Long-range PCR amplification of two controls, the proband and both parents with genomic primers flanking the deletion.



**Figure S2.** MLPA analysis of loci associated with primary macrocephaly in proband and parents. Ratios for probes MCPH1-2, MCPH1-4, MCPH1-8 (GRCh37 positions 6254211, 6276358, 6290246 on chromosome 8) indicate a biallelic deletion of *MCPH1* exons 1-8 in the proband and a heterozygous deletion in both parents.

