Supplementary Data

CSF1R mutations link POLD and HDLS as a single disease entity

Alexandra M. Nicholson PhD[1], Matt C. Baker BSc[1], NiCole A. Finch MS[1], Nicola J. Rutherford BSc[1], Christian Wider MD[1,2], Neill R. Graff-Radford MD[3], Peter T. Nelson MD, PhD[4], H. Brent Clark MD PhD[5], Zbigniew K. Wszolek MD[2], Dennis W. Dickson MD[1], David S. Knopman MD[6] and Rosa Rademakers PhD[1]*

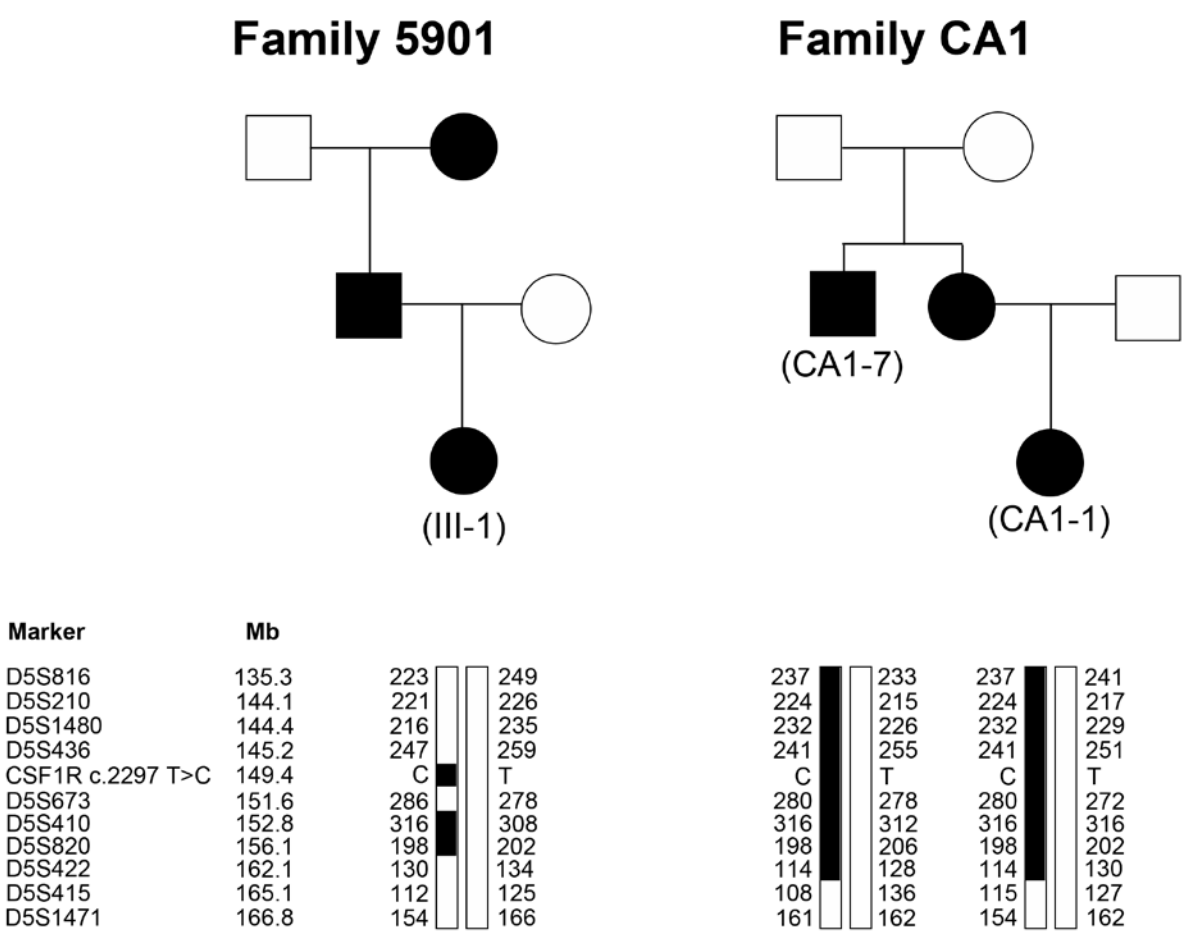

Figure e-1. CSF1R p.M766T haplotype sharing study.

We analyzed ten STR markers from within a 31.5Mb region of chromosome 5 that flanks *CSF1R* in one patient from POLD family 5901 and two patients from the previously reported HDLS family CA1.[1] For each marker, the two alleles with their lengths in base pairs are shown. Alleles from the disease haplotype (defined by sharing between affected family members in family CA1) are highlighted in black. Control frequencies of the 316bp allele of D5S410 and 198bp allele of D5S820 are 36.8% and 30.2% respectively suggesting that these alleles are likely shared between families 5901 and CA1 by chance.

e-References

1. Rademakers R, Baker M, Nicholson AM, et al. Mutations in the colony stimulating factor 1 receptor (CSF1R) gene cause hereditary diffuse leukoencephalopathy with spheroids. Nat Genet 2011;44:200-205.