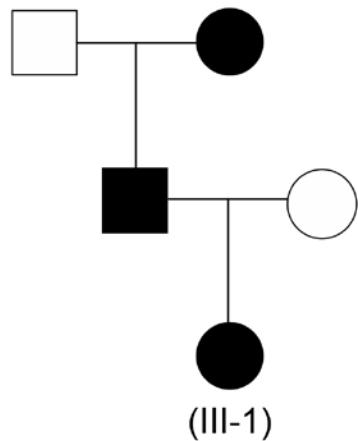


# ***Supplementary Data***

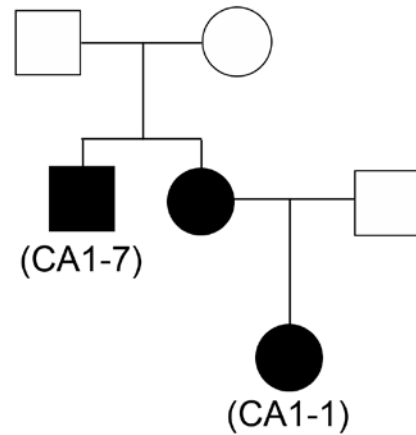
## ***CSF1R mutations link POLD and HDLS as a single disease entity***

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## Family 5901



## Family CA1



Marker	Mb	223	249	237	233	237	241
D5S816	135.3	221	226	224	215	224	217
D5S210	144.1	216	235	232	226	232	229
D5S1480	144.4	247	259	241	255	241	251
D5S436	145.2	C	T	C	T	C	T
CSF1R c.2297 T>C	149.4	286	278	280	278	280	272
D5S673	151.6	316	308	316	312	316	316
D5S410	152.8	198	202	198	206	198	202
D5S820	156.1	130	134	114	128	114	130
D5S422	162.1	112	125	108	136	115	127
D5S415	165.1	154	166	161	162	154	162
D5S1471	166.8						

### 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.<sup>1</sup> 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.