Supplementary Online Content

Harms M, Benitez BA, Cairns N, et al; for the NIA-LOAD/NCRAD Family Study Consortium. *C9orf72* hexanucleotide repeat expansions in clinical Alzheimer disease. *JAMA Neurol*. Published online April 15, 2013. doi:10.1001/2013.jamaneurol.537.

This supplementary material has been provided by the authors to give readers additional information about their work.

eTable 1: Familial patients with AD with abnormal *C9ORF72* repeat expansions share the at-risk haplotype found in patients with FTD and ALS

Patient genotypes for SNPs defining the known repeat expansion risk haplotype were extracted from Illumina SNP genotyping chips. Haplotypes were phased using MACH and the HapMap CEU population as reference. Yellow highlighting shows the allele matching the risk haplotype and demonstrates it is present in its entirety in all 5 index cases with abnormal repeat expansions.

	Risk Allele	Family 1		Family 2		Family 3		Family 4		Family 5	
rs1444533	T	T	C	T	T	T	T	T	T	T	C
rs1822723	C	C	C	C	C	C	T	C	C	C	C
rs4879515	T	T	C	T	C	T	C	T	T	T	C
rs895023	A	A	A	A	A	A	A	A	A	A	A
rs868856	A	A	G	A	G	A	G	A	A	A	G
rs7046653	A	A	G	A	G	A	G	A	A	A	G
rs2440622	T	T	T	T	T	T	T	T	T	T	T
rs1977661	C	C	C	C	C	C	C	C	C	C	C
rs903603	G	G	A	G	A	G	G	G	G	G	A
rs10812610	C	C	A	C	A	C	C	C	C	C	A
rs2814707	T	T	C	T	C	T	C	T	T	T	C
rs3849942	T	T	C	T	C	T	C	T	T	T	C
rs12349820	T	T	T	T	T	T	T	T	T	T	T
rs10122902	G	G	G	G	G	G	G	G	G	G	G
rs10757665	T	T	T	T	T	T	T	T	T	T	T
rs1565948	G	G	G	G	G	G	A	G	G	G	G
rs774359	C	C	T	C	T	C	T	C	C	C	T
rs2282241	C	C	A	C	A	C	A	C	C	C	A
rs1948522	C	C	C	C	C	C	T	C	C	C	C
rs1982915	G	G	G	G	A	G	G	G	G	G	A
rs7868845	T/C	C	C	T	C	C	C	T	T	T	C
rs2453556	G	G	A	A	A	G	A	G	G	G	G
rs702231	A	A	C	G	A	A	C	A	A	A	C
rs696826	G	G	G	G	G	G	G	G	G	G	G
rs2477518	T	T	C	T	T	T	C	T	T	T	T

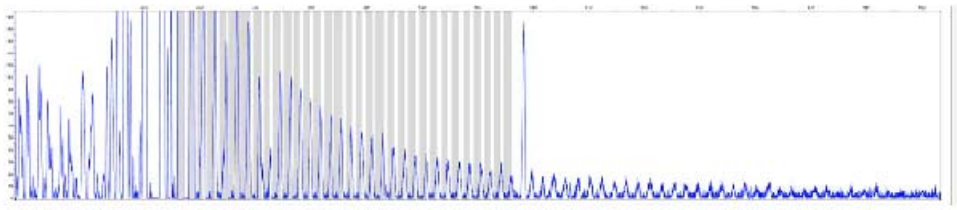
eTable 2: **At-risk haplotype segregation within AD families carrying abnormal *C9ORF72* repeat expansions**

	Risk Allele	Family 1		Family 2						Family 3						Family 4						Family 5					
Status		Index Case		Index Case		Case		Control		Index Case		Case		Case*		Index Case		Case		Case		Index Case		Case		Control	
Age/AAO		73		68		65		72		60		60		65		60		70		60		67		70		72	
rs1444533	T	T	C	T	T	T	T	T	T	T	T	T	T	T	C	T	T	T	T	T	T	T	C	T	T	T	T
rs1822723	C	C	C	C	C	C	T	T	T	C	T	C	T	T	C	C	C	C	C	C	C	C	C	C	C	C	C
rs4879515	T	T	C	T	C	T	C	C	C	T	C	T	C	C	C	T	T	T	T	T	T	T	C	T	T	T	T
rs895023	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A
rs868856	A	A	G	A	G	A	G	G	G	A	G	A	G	G	G	A	A	A	G	A	A	A	G	A	A	A	A
rs7046653	A	A	G	A	G	A	G	G	G	A	G	A	G	G	G	A	A	A	G	A	A	A	G	A	A	A	A
rs2440622	T	T	T	T	T	T	T	T	T	T	T	T	T	T	T	T	T	T	T	T	T	T	T	T	T	T	T
rs1977661	C	C	C	C	C	C	C	C	C	C	C	C	C	C	C	C	C	C	C	C	C	C	C	C	C	C	C
rs903603	G	G	A	G	A	G	A	A	A	G	G	G	G	A	A	G	G	G	G	G	G	G	A	G	G	G	G
rs10812610	C	C	A	C	A	C	A	A	A	C	C	C	C	A	A	C	C	C	C	C	C	C	A	C	C	C	C
rs2814707	T	T	C	T	C	T	C	C	C	T	C	T	C	C	C	T	T	T	C	T	T	T	C	T	T	T	T
rs3849942	T	T	C	T	C	T	C	C	C	T	C	T	C	C	C	T	T	T	C	T	T	T	C	T	T	T	T
rs12349820	T	T	T	T	T	T	T	C	T	T	T	T	T	C	T	T	T	T	T	T	T	T	T	T	T	T	T
rs10122902	G	G	G	G	G	G	G	G	G	G	G	G	G	G	G	G	G	G	A	G	G	G	G	G	G	G	G
rs10757665	T	T	T	T	T	T	T	C	T	T	T	T	T	C	T	T	T	T	T	T	T	T	T	T	T	T	T
rs1565948	G	G	G	G	G	G	G	A	G	G	A	G	A	A	G	G	G	G	A	G	G	G	G	G	G	G	G
rs774359	C	C	T	C	T	C	T	T	T	C	T	C	T	T	T	C	C	C	T	C	C	C	T	C	C	C	C
rs2282241	C	C	A	C	A	C	A	C	A	C	A	C	A	C	A	C	C	C	A	C	C	C	A	C	C	C	C
rs1948522	C	C	C	C	C	C	C	C	C	C	T	C	T	C	C	C	C	C	T	C	C	C	C	C	C	C	C
rs1982915	G	G	G	G	A	G	A	A	A	G	G	G	G	A	A	G	G	G	G	G	G	G	A	G	G	G	G
rs7868845	T/C	C	C	T	C	T	C	C	C	C	C	C	C	C	C	T	T	T	T	T	T	T	C	T	C	C	C
rs2453556	G	G	A	G	A	G	A	A	A	G	A	G	A	A	A	G	G	G	G	G	G	G	G	G	G	G	G
rs702231	A	A	C	A	A	A	A	A	A	A	C	A	C	A	A	A	A	A	A	A	A	A	C	A	A	A	A
rs696826	G	G	G	G	G	G	G	G	G	G	G	G	G	G	G	G	G	G	G	G	G	G	G	G	G	G	G
rs2477518	T	T	C	T	T	T	T	T	T	T	C	T	C	T	T	T	T	T	T	T	T	T	T	T	C	C	C

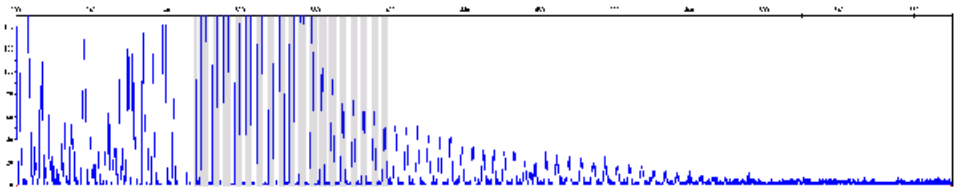
Patient genotypes for SNPs defining the known expansion risk haplotype were extracted from Illumina SNP genotyping chips. Haplotypes were phased using MACH and the HapMap CEU population as reference Yellow highlighting shows the allele matching the haplotype, Non-matching genotypes are highlighted in red.
*This individual showed only 7 repeats on repeat-primed PCR

eFigure: Example repeat-primed PCR results

Carrier of the expanded repeat (>1000 repeats by Southern blot)



Carrier of an abnormal repeat expansion (35-100 repeats by Southern blot)



Negative for the repeat expansion (normal repeats by Southern blot)