The American Journal of Human Genetics, Volume 90

Supplemental Data

Strikingly Different Clinicopathological Phenotypes

Determined by Progranulin-Mutation Level

Katherine R. Smith, John Damiano, Silvana Franceschetti, Stirling Carpenter, Laura Canafoglia, Michela Morbin, Giacomina Rossi, Davide Pareyson, Sara E. Mole, John F. Staropoli, Katherine B. Sims, Jada Lewis, Wen-Lang Lin, Dennis W. Dickson, Hans-Henrik Dahl, Melanie Bahlo, and Samuel F. Berkovic

Table S1. Alignment Statistics for Three Exomes from an Italian Family

Sample	Father		Mother		Son	
	N	%	N	%	N	%
Read Sequences	73,568,070	100.00	67,593,008	100.00	56,978,536	100.00
Aligned	69,181,736	94.04	63,380,692	93.77	53,589,746	94.05
Unique Alignment	62,945,609	85.56	57,801,021	85.51	48,655,068	85.39
post PCR duplicate removal	54,616,711	74.24	49,880,709	73.80	41,618,887	73.04
to chr7 linkage interval	44,326	0.06	40,488	0.06	33,517	0.06
to chr17 linkage interval	237,458	0.32	208,634	0.31	179,021	0.31

Table S2. Coverage of Targeted Bases for Three Exomes from an Italian Family

Sample Coverage	Father	Mother	Son
Min.	0	0	0
1st quartile	14	13	11
Median	40	37	31
Mean	47.88	44.36	36.84
3rd quartile	72	67	55
Max.	3396	5308	2389
% bases covered ≥5	86.2	85.5	84.7
% bases covered ≥10	79.9	79.0	77.0

Coverage statistics consider the 62,085,295 bases targeted the by TruSeq platform. Coverage statistics refer to uniquely aligning reads post PCR duplicate removal. Bases were required to have base quality≥13 and come from a read with mapping quality≥13 in order to contribute to coverage

Table S3. Rare Functional Candidate Variants Detected in Linkage Peaks

Gene	Description	UCSC knownGene ID	cDNA Change	Protein Change	dbSNP132	Frequency	PolyPhen-2 Prediction
GRN	granulin precursor	uc002igp.1	c.813_816del	p.Thr272Serfs*10	rs63749877	-	-
DBF4B	DBF4 homolog B isoform 2	uc002ihf.2	c.1711T>A	p.Ser571Thr	rs117190451	0.006	Unknown
ADAM11	ADAM metallopeptidase domain 11 preproprotein	uc002ihh.2	c.569G>A	p.Arg190Gln	rs116926739	0.006	Benign

Variant frequency information is from the May 2011 release of the 1000 Genomes data; - indicates that the variant was not present. For all variants listed, the two parents are heterozygous while the affected son is homozygous.