

Table E4: SNP genotyping results in patients negative for mutation in *DNAI1* for homozygosity exclusion

Patients #6158, #8139, #8144, #8148, #8194 and #8472 (shaded cells in column 1) where no heterozygosity could be found were retained for full length sequencing of *DNAI1* coding regions. Shaded boxes in SNP columns represent homozygous genotypes.

PCR reactions were undertaken using 0.04 U of Taq polymerase enzyme (Pharmacia Biotech) in its supplied buffer (15 mM MgCl₂) with 0.2 mM dNTP (Pharmacia Biotech) and 10 µmol of their respective forward and reverse primers. PCR was performed using 1 cycle of 95 °C for 5 min followed by 40 cycles of 95°C for 30 s, 55°C for 30 s, and 72°C for 30 s. Following a final incubation at 72 °C for 5 min the products were cooled and maintained at 4 °C. PCR products were checked for size and yield on a 2% agarose gel. Amplicons were purified and minisequenced using 30 pmol of sequencing primer per reaction under standard conditions according to the manufacturer's instructions (Pyrosequencing, Biotage, Uppsala, Sweden) in a Pyrosequencing PSQ HS 96 System. Single-stranded DNA-binding protein was added to the primed DNA template prior to the reaction to reduce mispriming and increase signal intensity. Data were captured using PSQ HS 96 SNP software (Pyrosequencing, Biotage AB, Uppsala, Sweden).

Patient ID	SNPs								
	rs4879792	rs10738928	rs3793472	rs4878575	rs3750395	rs17353380	rs9657620	rs12001113	rs11999046
6135	C/C	G/G	T/C	G/G			G/G	A/A	
6142			T/C	G/T				G/A	
6168	G/C	G/G	T/T	G/T			G/A	G/A	
6184	G/G	G/G	T/T	G/G			G/A	G/A	
6191	G/C	G/G	T/C	G/T			G/A	A/A	
3038	C/C		T/C	G/G			G/A	A/A	
8422			T/C	G/T				G/A	
8466			T/C	G/T				G/A	
8472	G/G	G/G	T/T	G/G	T/T	G/G	G/G	A/A	C/C
8473	G/G	G/C	T/T	G/T			G/A	G/A	
8476	G/C	G/G	T/T	G/G			G/G	A/A	
8492			T/C	G/G				G/A	
6419	G/G	G/C	T/T	G/T			G/A	G/G	
5342	G/C	C/C	T/C	G/G				A/A	
6121	G/C	G/G	T/T	G/G		G/G	G/G	G/A	
6132	G/G	G/G	T/T	G/T			G/A	G/G	
6138	G/C	G/C	T/C	G/T			G/A	A/A	
6151	G/C	G/G	T/T	G/G			G/G	G/A	
6158	G/G	G/G	T/T	G/G	T/T	G/G	G/G	A/A	C/C
6162	G/C	G/C	T/C	G/T			G/A	A/A	
6048	G/C	G/C	T/T	G/T			G/A	A/A	
6101	G/C	G/C	T/T	G/T			A/A	G/A	
6170			T/C	G/T				G/A	
8507	G/C	G/C	T/C	G/T			G/A	A/A	
6195			T/C	G/T				G/A	
6197	G/C	G/C	T/T	G/G			G/A	G/A	
6476			T/C	G/G				A/A	
6266	G/C	G/G	T/C	G/T			G/A	G/G	
6270	C/C	G/G	T/C	G/G			G/G	A/A	
6276			T/C	G/T				G/A	
6144	C/C	G/G	T/C	G/G			G/G	A/A	
8197			T/C	G/G				G/A	
8104			T/C	G/T				G/A	
8105	G/G	G/C	T/T	G/T			G/A	G/A	
8130	C/C	G/G	T/C	G/G			G/G	A/A	
8131	G/G	G/C	T/T				A/A	G/A	
8132	G/C	G/G	T/T	G/G			G/G	G/A	
8133	G/C	G/G	T/C	G/G			G/G	A/A	
8137	G/C		T/C	G/T				G/A	
8138	G/G	G/G	T/C	G/G			G/G	A/A	
8139	G/G	G/G	T/T	G/G	T/T	G/G	G/G	G/G	C/C
8140	G/C	G/G	T/C	G/G			G/G	A/A	
8144	G/G	G/G	T/T	T/T	C/C	G/G	A/A	G/G	G/G
8145	C/C	G/G	T/C	G/G			G/G	A/A	
8146	G/C	G/C	T/T	G/T			G/A	G/A	
8147	G/C	G/G	T/C	G/G			G/G	A/A	
8148	C/C	G/G	C/C	G/G	C/C	G/G	G/G	A/A	C/C
8149	G/G	G/C	T/T	G/T			G/A	G/G	
8150	C/C	G/G	T/C	G/G			G/G	A/A	
3148	G/C	G/G	T/T	G/G			G/G	G/A	
5063	G/C	G/G	T/C	G/G			G/G	A/A	
8172	G/C	G/C	T/T	G/T			G/A	A/A	
8173	G/C	G/G	T/C	G/T			G/A	A/A	
8174	G/G	G/C	T/T	G/T			G/A	G/G	
8175			T/C	G/G				G/A	
8176			T/C	G/G				G/A	
8178	G/C	G/C	T/T	G/T			G/A	A/A	
8182			T/T	G/T				G/A	
8183	G/C	G/C	T/C	G/T			G/A	A/A	
8194	C/C	G/G	T/T	G/G	C/C	G/G	G/G	A/A	C/C
8224	G/C	G/G	T/T	G/G			G/G	A/A	
8225			T/T	G/G				G/A	
8226	G/G	G/G	T/C	G/G		G/G	G/G	G/G	G/G
8568	G/C	G/C	T/T	G/T			G/A	A/A	
8228	G/G	G/C	G/T	G/T			G/A	G/A	
8271	G/G	C/C	T/T	T/T			A/A	G/A	