

Supplementary Fig. S3. Distribution of SNPs throughout the *CERKL* genomic region. Along the 160165 bp encompassing the complete *CERKL* gene, 1088 SNPs have been reported. Only 69 of those are located within exons and 8 correspond to pathogenic reported mutations (highlighted in green). Exons with more SNPs than mean are highlighted in red, whereas those with no SNPs –or less SNPs than mean– are shown in blue.

	hg19 End	hg19 Start	hg19 Size	hg19 N mut	hg19 N SNF	hg19 SNPs/Kb		
5'UTR_NeuroD1/5'UTR_CERKL2	182545156	182545381	226	0	3	13,274		
NeuroD1	182542517	182543587	1071	0	5	4,6685		
3'UTR_NeuroD1	182541194	182542516	1323	0	16	12,094		
5'UTR_CERKL	182521734	182521851	118	0	1	8,4746		
Exon1	182521496	182521733	238	1	4	16,807		
Exon1a	182521703	182521733	31	0	0	0		
Exon1b	182521496	182521562	67	1	2	29,851		
ExonVega1	182514687	182514804	118	0	2	16,949		
ExonVega2	182514307	182514463	157	0	0	0		
Exon2a	182481982	182482148	167	0	2	11,976		
Exon2	182468564	182468806	243	2	4	16,461		
Exon3a	182438480	182438529	50	0	0	0		
Exon3	182438480	182438611	132	0	0	0		
Exon4	182430785	182430848	64	0	0	0		
Exon4b	182430160	182430237	78	0	1	12,821		
Exon5	182423293	182423435	143	2	2	13,986		
Exon6	182414361	182414435	75	0	0	0		
Exon7	182413407	182413584	178	0	0	0		
Exon8	182413272	182413331	60	1	0	0		
Exon8_intron8_exon9	182412549	182413331	783	1	20	25,543	All intronic SNPs	
Exon9	182412549	182412574	26	0	0	0		
Exon10	182412345	182412453	109	0	0	0		
Exon11	182409427	182409523	97	0	1	10,309		
Exon12	182403819	182403991	173	0	2	11,561		
Exon13	182402911	182402971	61	0	0	0		
3'UTR_CERKL	182402471	182402910	440	0	4	9,0909		
PH domain			480		8	16,667		
NLS-1 domain			36		0	0		
NLS-2 domain			36		2	55,556		
NES-1 domain			30		0	0		
NES-2 domain			36		1	27,778		
DAGK domain			459		2	4,3573		
CERK domain			216		1	4,6296		
					14			
Genomic region analyzed	160165							
SNPs analyzed	1088							
SNPs within exons	69							
SNP density (mean)	6,79299472							
Pathogenic mutations	8							
Exons with mutations								
Exons with more SNPs than mean								
Exons with less SNPs than mean								
SNPs within UTRs	24	11,3906028						
SNPs within exons	45	10,9196797						