Supporting Online Materials

Human adenylate kinase 2 deficiency causes a profound haematopoietic defect associated with sensorineural deafness

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Supplementary Table 1: Candidate region corresponding to the 2Mb genomic interval on human chromosome 1p32-1p34 and containing the adenylate kinase 2 gene.

start	stop	Symbol	location	description
32146380	32176575	PTP4A2	1p35	protein tyrosine phosphatase type IVA, member 2
32252078	32282059	KHDRBS1	1p32	KH domain containing, RNA binding, signal transduction associated
32311116	32341052	TMEM39B	1p35.1	transmembrane protein 39B
32346231	32414756	KPNA6	1p35.1-p34.3	karyopherin alpha 6 (importin alpha 7)
32417932	32436473	TXLNA	1p35.1	taxilin alpha
32438789	32443578	CCDC28B	1p35.1	coiled-coil domain containing 28B
32443708	32446980	LOC100128669	1p35.1	hypothetical protein LOC100128669
32443859	32446875	IQCC	1p36.11-p34.2	IQ motif containing C
32447282	32454384	DCDC2B	1p35.1	doublecortin domain containing 2B
32454385	32460513	C1orf91	1p36.11-p34.2	chromosome 1 open reading frame 91
32460558	32469792	EIF3I	1p34.1	eukaryotic translation initiation factor 3, subunit I
32469846	32479779	LOC339483	1p35.1	hypothetical LOC339483
32485405	32487048	C1orf90	1p35.1	chromosome 1 open reading frame 90
32489427	32524353	LCK	1p34.3	lymphocyte-specific protein tyrosine kinase
32530295	32571811	HDAC1	1p34	histone deacetylase 1
32572027	32574410	MARCKSL1	1p35.1	MARCKS-like 1
32597776	32600431	LOC100128071	1p35.1	similar to hCG41624
32600449	32602513	TSSK3	1p35-p34	testis-specific serine kinase 3
32603359	32632614	BSDC1	1p35.1	BSD domain containing 1
32640303	32642909	LOC343338	1p35.1	similar to Glyceraldehyde-3-phosphate dehydrogenase (GAPDH)
32661399	32662353	LOC100131615	1p35.1	hypothetical LOC100131615
32662452	32665767	LOC100133117	1p35.1	hypothetical LOC100133117
32694834	32726047	RP1-27O5.1	1p35.1	zinc finger and BTB domain containing 8-like
32777359	32844129	ZBTB8	1p35.1	zinc finger and BTB domain containing 8
32859892	32888772	ZBTB8OS	1p35.1	zinc finger and BTB domain containing 8 opposite strand
32889411	32918841	RBBP4	1p35.1	retinoblastoma binding protein 4
32918548	32933460	SYNC1	1p34.3-p33	syncoilin, intermediate filament 1
32980099	33013158	KIAA1522	1p35.1	KIAA1522
33013427	33056220	YARS	1p35.1	tyrosyl-tRNA synthetase
33055763	33097063	S100PBP	1p35.1	S100P binding protein
33100464	33108934	FNDC5	1p35.1	fibronectin type III domain containing 5
33124685	33132834	HPCA	1p35-p34.2	hippocalcin
33125700	33132004	LOC100129200	1p35.1	hypothetical protein LOC100129200
33132783	33139540	TMEM54	1p35-p34	transmembrane protein 54
33174634	33202301	RNF19B	1p35.1	ring finger protein 19B
33218133	33218831	LOC127545	1p35.1	hCG1641703
33246173	**33275079**	**AK2**	**1p34**	**adenylate kinase 2**
33319301	33358582	ADC	1p33-p34.3	arginine decarboxylase
33383591	33419854	TRIM62	1p35.1	tripartite motif-containing 62
33494761	33538907	ZNF362	1p35.1	zinc finger protein 362
33544954	33559286	A3GALT2	1p35.1	alpha 1,3-galactosyltransferase 2 (isoglobotriaosylceramide synthase)
33561811	33613781	PHC2	1p34.3	polyhomeotic homolog 2 (Drosophila)
33704437	33707163	LOC100131451	1p35.1	hypothetical LOC100131451
33710819	33734582	ZSCAN20	1p34.3	zinc finger and SCAN domain containing 20
33750191	33756426	LOC100132171	1p35.1	hypothetical LOC100132171
33752196	34404030	CSMD2	1p35.1-p34.3	CUB and Sushi multiple domains 2
34098663	34102979	HMGB4	1p35.1	high-mobility group box 4
34415858	34457319	C1orf94	1p34.3	chromosome 1 open reading frame 94

Supplementary Table 2: Primers sequences used for RT-PCR analysis and shRNA constructs

RT-PCR primers

AK2	FWD	5'-AACTGGTGGCAGTGAGAGAC-3'
	REV	5'-TGCTGAATTCAATCACAGAA-3'
28S	FWD	5'-ATAAGCGCCTCTCGGTTTTG-3'
	REV	5'-GCTCCTTGATGTGTTCTGGT-3'

shRNA primers used for cloning into pLVTHM

shAK2: containing target sequences (in bold) from AK2 cDNA at position 232-250.

FWD

5'CGCGTCCCC**ATGGTAGTGGAGCTCATTG**TTCAAGAGA**CAATGAGCTCCACTACCAT**TTTTTGGAAAT3'

REV

5'CGATTTCCAAAAA**ATGGTAGTGGAGCTCATTG**TCTCTTGAA**CAATGAGCTCCACTACCAT**GGGGA3'

ShScramble : containing a scramble version of the AK2 target sequence.

FWD

5'CGCGTCCCC**GACTCGGGTAGTTTATGGA**TTCAAGAGA**TCCATAAACTACCCGAGTC**TTTTTGGAAAT3'

REV

5'CGATTTCCAAAAA**GACTCGGGTAGTTTATGGA**TCTCTTGAA**TCCATAAACTACCCGAGTC**GGGGA3'