

Supplementary data

Fig. S1. Pedigree and co-segregation analyses of the novel frameshift mutation in the *BCL11A* gene. Sequencing chromatograms confirmed that the new frameshift deletion (c.271delG) occurred *de-novo* in the patient (II.2.). The unaffected family members, including mother (I.1), father (I.2), and older sister (II.1) did not show any sequence alteration at this nucleotide position.

Table S1. Summary of WES data after variant calling and annotation.

ID	Reads	% Aln	Depth	Variants	Hom.	Het.	SNP	Indel
P	114.90 M	99.5%	83.94	57'553	21'371	31'566	52'964	4'608
M-P	109.28 M	99.5%	79.42	58'174	21'589	31'606	53'227	4'592
F-P	121.47	99.5%	72.68	57'809	20'852	32'747	53'633	4'555

Abbreviations: number of aligned reads in percent (% Aln), average depth of coverage (depth), homozygous variants (hom.), heterozygous variants (het.), single nucleotide polymorphism (SNP), inserts and/or deletions (Indel), the patient (P), the healthy mother (M-P), and the healthy father (F-P).

Table S2. The *BCL11A* sequence variants detected in the investigated patient.

Variant effect	gDNA start	gDNA stop	cDNA	AA change	LF	AIF	ExAC	GTN P	GTN M-P	GTN F-P
synonymous	60,687,959	60,687,959	c.2088T>C	p.(=)	41	7'311	0.70	1/1	0/1	1/1
intronic	60,768,978	60,768,978	c.385+4128C>T	p.(=)	35	1'532	0.65	0/1	1/1	-
frameshift	60,773,219	60,773,220	c.271delG	p.E91Rfs*2	1	0	nf	0/1	-	-

The variants were annotated based on the isoform L of the *BCL11A* gene (NM_018014.3). Abbreviations: nucleotide positions on the human chromosome 2 (gDNA start, gDNA stop), amino acid exchange (AA), statistical data from local database (LF), statistical data from external database (AF), total allele frequencies in percent according to the Exome Aggregation Consortium (ExAC), numeric genotypes (GTN), homozygous variant (1/1), heterozygous variant (0/1), lack of alterations in the reference sequence (-), not found (nf), the patient (P), the healthy mother (M-P), and the healthy father (F-P).

Table S3. Panel of genes associated with epilepsies and epileptic syndromes.

No.	Gene	OMIM No.	Phenotype
1	ABAT	137150	Ep-s
2	ABCC8	600509	Ep-s
3	ABCD1	300371	Ep-s
4	ACY1	104620	Ep-s
5	ADAR	146920	Ep-s
6	ADK	102750	Ep-s
7	ADSL	608222	Ep-s
8	AFG3L2	604581	Ep-s
9	AIFM1	300169	Ep-r
10	AIMP1	603605	Ep-r
11	ALDH4A1	606811	Ep-s
12	ALDH5A1	610045	Ep-s
13	ALDH7A1	107323	Ep-s
14	ALG13	300776	Ep-s
15	ALG3	608750	Ep-s
16	AMACR	604489	Ep-s
17	AMT	238310	Ep-s
18	AP3B2	602166	Ep-s
19	AP4B1	607245	Ep-s
20	AP4E1	607244	Ep-s
21	AP4M1	602296	Ep-s
22	AP4S1	607243	Ep-s
23	ARG1	608313	Ep-s
24	ARHGEF9	300429	Ep-s
25	ARSA	607574	Ep-s
26	ARV1	611647	Ep-s
27	ARX	300382	Ep-s
28	ASAH1	613468	Ep-s
29	ASNS	108370	Ep-s
30	ASPA	608034	Ep-r
31	ATIC	601731	Ep-s
32	ATP13A2	610513	Ep-s
33	ATP1A2	182340	Ep-s

34	ATP1A3	182350	Ep-s
35	ATP6AP2	300556	Ep-s
36	ATRX	300032	Ep-s
37	AUH	600529	Ep-r
38	BCKDHA	608348	Ep-s
39	BCKDHB	248611	Ep-s
40	BCKDK	614901	Ep-s
41	BCS1L	603647	Ep-s
42	BRAT1	614506	Ep-s
43	BTB	609019	Ep-s
44	CACNA1A	601011	Ep-s
45	CACNA1D	114206	Ep-s
46	CACNA1H	607904	Ep-s
47	CACNB4	601949	Ep-s
48	CAD	114010	Ep-s
49	CARS2	612800	Ep-s
50	CASK	300172	Ep-s
51	CASR	601199	Ep-s
52	CDKL5	300203	Ep-s
53	CERS1	606919	Ep-s
54	CHD2	602119	Ep-s
55	CHRNA2	118502	Ep-s
56	CHRNA4	118504	Ep-s
57	CHRNA7	118511	Ep-s
58	CHRNA2	118507	Ep-s
59	CLCN2	600570	Ep-s
60	CLCN4	302910	Ep-s
61	CLN3	607042	Ep-s
62	CLN5	608102	Ep-s
63	CLN6	606725	Ep-s
64	CLN8	607837	Ep-s
65	CNNM2	607803	Ep-s
66	CNTNAP2	604569	Ep-s
67	COL4A1	120130	Ep-s
68	COQ4	612898	Ep-s

69	COX15	603646	Ep-s
70	COX6B1	124089	Ep-s
71	COX8A	123870	Ep-s
72	CPA6	609562	Ep-s
73	CPT1A	600528	Ep-s
74	CPT2	600650	Ep-s
75	CRH	122560	Ep-s
76	CSF1R	164770	Ep-s
77	CSTB	601145	Ep-s
78	CTC1	613129	Ep-s
79	CTSD	116840	Ep-s
80	CTSF	603539	Ep-s
81	CUL4B	300304	Ep-s
82	CYP27A1	606530	Ep-s
83	D2HGDH	609186	Ep-s
84	DARS2	610956	Ep-s
85	DBT	248610	Ep-s
86	DCX	300121	Ep-s
87	DDC	107930	Ep-s
88	DDX3X	300160	Ep-s
89	DENND5A	617278	Ep-s
90	DEPDC5	614191	Ep-s
91	DHFR	126060	Ep-s
92	DLD	238331	Ep-s
93	DNAJC5	611203	Ep-s
94	DNM1	602377	Ep-s
95	DNM1L	603850	Ep-s
96	DOCK7	615730	Ep-s
97	DPYD	612779	Ep-s
98	DPYS	613326	Ep-s
99	DYRK1A	600855	Ep-s
100	EARS2	612799	Ep-s
101	ECHS1	602292	Ep-s
102	ECM1	602201	Ep-s
103	EEF1A2	602959	Ep-s

104	EFHC1	608815	Ep-s
105	EIF2B1	606686	Ep-s
106	EIF2B2	606454	Ep-s
107	EIF2B3	606273	Ep-r
108	EIF2B4	606687	Ep-r
109	EIF2B5	603945	Ep-r
110	EPM2A	607566	Ep-s
111	ETFA	608053	Ep-s
112	ETFB	130410	Ep-s
113	ETFDH	231675	Ep-s
114	ETHE1	608451	Ep-s
115	FA2H	611026	Ep-s
116	FAM126A	610531	Ep-s
117	FAR1	616107	Ep-s
118	FARS2	611592	Ep-s
119	FGF12	601513	Ep-s
120	FH	136850	Ep-s
121	FLNA	300017	Ep-s
122	FOLR1	136430	Ep-s
123	FOXG1	164874	Ep-s
124	FOXRED1	613622	Ep-s
125	FRRS1L	604574	Ep-s
126	GABRA1	137160	Ep-s
127	GABRB1	137190	Ep-s
128	GABRB2	600232	Ep-s
129	GABRB3	137192	Ep-s
130	GABRD	137163	Ep-s
131	GABRG2	137164	Ep-s
132	GALC	606890	Ep-s
133	GAMT	601240	Ep-s
134	GCDH	608801	Ep-s
135	GCH1	600225	Ep-s
136	GCK	138079	Ep-s
137	GCSH	238330	Ep-s
138	GFAP	137780	Ep-s

139	GFM1	606639	Ep-s
140	GJC2	608803	Ep-s
141	GLB1	611458	Ep-s
142	GLDC	238300	Ep-s
143	GLRA1	138491	Ep-s
144	GLRB	138492	Ep-s
145	GLUD1	138130	Ep-s
146	GLUL	138290	Ep-s
147	GNAO1	139311	Ep-s
148	GNB1	139380	Ep-s
149	GNE	603824	Ep-s
150	GOSR2	604027	Ep-s
151	GPHN	603930	Ep-s
152	GRIA3	305915	Ep-s
153	GRIK2	138244	Ep-s
154	GRIN1	138249	Ep-s
155	GRIN2A	138253	Ep-s
156	GRIN2B	138252	Ep-s
157	GRIN2D	602717	Ep-s
158	GRN	607485	Ep-s
159	GTPBP3	608536	Ep-s
160	HACE1	610876	Ep-s
161	HADH	601609	Ep-s
162	HCN1	602780	Ep-s
163	HDAC4	605314	Ep-s
164	HECW2	617245	Ep-s
165	HEPACAM	611642	Ep-s
166	HIBCH	610690	Ep-s
167	HLCS	609018	Ep-s
168	HNRNPU	602869	Ep-s
169	HPD	609695	Ep-s
170	HSD17B10	300256	Ep-s
171	HSPD1	118190	Ep-s
172	HTRA1	602194	Ep-s
173	HTT	182138	Ep-s

174	HUWE1	300697	Ep-s
175	IBA57	615316	Ep-s
176	IDH2	147650	Ep-s
177	INSR	147670	Ep-s
178	IQSEC2	300522	Ep-s
179	ITPA	147520	Ep-s
180	IVD	607036	Ep-s
181	JRK	603210	Ep-s
182	KANSL1	612452	Ep-s
183	KCNA1	176260	Ep-s
184	KCNA2	176262	Ep-s
185	KCNB1	600397	Ep-s
186	KCNC1	176258	Ep-s
187	KCNH1	603305	Ep-s
188	KCNJ10	602208	Ep-s
189	KCNJ11	600937	Ep-s
190	KCNMA1	600150	Ep-s
191	KCNQ1	607542	Ep-s
192	KCNQ2	602235	Ep-s
193	KCNQ3	602232	Ep-s
194	KCNT1	608167	Ep-s
195	KCTD7	611725	Ep-s
196	KDM5C	314690	Ep-s
197	KIF1A	601255	Ep-s
198	KIF5C	604593	Ep-s
199	L2HGDH	609584	Ep-s
200	LGI1	604619	Ep-s
201	LIAS	607031	Ep-s
202	LMNB1	150340	Ep-s
203	LMNB2	150341	Ep-s
204	LRPPRC	607544	Ep-s
205	LYRM7	615831	Ep-m
206	MAGI2	606382	Ep-s
207	MAPK10	602897	Ep-s
208	MARS2	609728	Ep-s

209	MBD5	611472	Ep-s
210	MBOAT7	606048	Ep-s
211	MCCC1	609010	Ep-s
212	MDH2	154100	Ep-s
213	MECP2	300005	Ep-s
214	MED12	300188	Ep-s
215	MEF2C	600662	Ep-s
216	MFSD8	611124	Ep-s
217	MLC1	605908	Ep-s
218	MLYCD	606761	Ep-s
219	MMACHC	609831	Ep-s
220	MOCS1	603707	Ep-s
221	MOCS2	603708	Ep-s
222	MRPL44	61184	Ep-s
223	MTFMT	611766	Ep-m
224	MTHFR	236250	Ep-s
225	MTOR	601231	Ep-s
226	NACC1	610672	Ep-s
227	NARS2	612803	Ep-s
228	NDUFA1	300078	Ep-s
229	NDUFAF5	612360	Ep-m
230	NDUFAF6	612392	Ep-m
231	NDUFS2	602985	Ep-m
232	NDUFS4	602694	Ep-m
233	NDUFS7	601825	Ep-m
234	NDUFS8	602141	Ep-m
235	NDUFV1	161015	Ep-s
236	NECAP1	611623	Ep-s
237	NEU1	608272	Ep-s
238	NHLRC1	608072	Ep-s
239	NOTCH3	600276	Ep-s
240	NPRL2	607072	Ep-s
241	NPRL3	600928	Ep-s
242	NR2F1	132890	Ep-s
243	NRXN1	600565	Ep-s

244	NUBPL	613621	Ep-m
245	OFD1	300170	Ep-s
246	OPHN1	300127	Ep-s
247	PACS2	610423	Ep-s
248	PC	608786	Ep-s
249	PCBD1	126090	Ep-s
250	PCCA	232000	Ep-s
251	PCCB	232050	Ep-s
252	PCDH19	300460	Ep-s
253	PDHA1	300502	Ep-s
254	PDHX	608769	Ep-s
255	PDSS2	610564	Ep-s
256	PET100	614770	Ep-m
257	PGAP1	611655	Ep-s
258	PGAP2	615187	Ep-s
259	PGAP3	611801	Ep-s
260	PGK1	311800	Ep-s
261	PHF6	300414	Ep-s
262	PHGDH	606879	Ep-s
263	PIGA	311770	Ep-s
264	PIGG	616918	Ep-s
265	PIGL	605947	Ep-s
266	PIGM	610273	Ep-s
267	PIGN	606097	Ep-s
268	PIGO	614730	Ep-s
269	PIGT	610272	Ep-s
270	PIGV	610274	Ep-s
271	PIGW	610275	Ep-s
272	PIGY	610662	Ep-s
273	PIK3R2	603157	Ep-s
274	PLCB1	607120	Ep-s
275	PLP1	300401	Ep-s
276	PLPBP	604436	Ep-s
277	PNKP	605610	Ep-s
278	PNPO	603287	Ep-s

279	POLG	174763	Ep-s
280	POLR3A	614258	Ep-r
281	POLR3B	614366	Ep-s
282	PPT1	600722	Ep-s
283	PRDM8	616639	Ep-s
284	PRICKLE1	608500	Ep-s
285	PRIMA1	613851	Ep-s
286	PRODH	606810	Ep-s
287	PRRT2	614386	Ep-s
288	PSAP	176801	Ep-s
289	PSAT1	610936	Ep-s
290	PSPH	172480	Ep-s
291	PTS	612719	Ep-s
292	PURA	600473	Ep-s
293	PYCR2	616406	Ep-s
294	QDPR	612676	Ep-s
295	RAB39B	300774	Ep-s
296	RELN	600514	Ep-s
297	RMND1	614917	Ep-s
298	RNASEH2A	606034	Ep-s
299	RNASEH2B	610326	Ep-s
300	RNASEH2C	610330	Ep-s
301	RNASET2	612944	Ep-s
302	RNF216	609948	Ep-s
303	ROGDI	614574	Ep-s
304	RORB	601972	Ep-s
305	SAMHD1	606754	Ep-s
306	SCARB2	602257	Ep-s
307	SCN1A	182389	Ep-s
308	SCN1B	600235	Ep-s
309	SCN2A	182390	Ep-s
310	SCN8A	600702	Ep-s
311	SCN9A	603415	Ep-s
312	SCO1	603644	Ep-m
313	SDHA	600857	Ep-s

314	SDHAF1	612848	Ep-m
315	SERAC1	614725	Ep-s
316	SERPINI1	602445	Ep-s
317	SIK1	605705	Ep-s
318	SLC12A5	606726	Ep-s
319	SLC13A5	608305	Ep-s
320	SLC16A1	600682	Ep-s
321	SLC19A3	606152	Ep-s
322	SLC1A2	600300	Ep-s
323	SLC1A3	600111	Ep-s
324	SLC25A1	190315	Ep-s
325	SLC25A12	603667	Ep-s
326	SLC25A15	603861	Ep-s
327	SLC25A22	609302	Ep-s
328	SLC2A1	138140	Ep-s
329	SLC35A2	314375	Ep-s
330	SLC35A3	605632	Ep-s
331	SLC39A8	608732	Ep-s
332	SLC46A1	611672	Ep-s
333	SLC6A1	137165	Ep-s
334	SLC6A5	604159	Ep-s
335	SLC6A8	300036	Ep-s
336	SLC6A9	601019	Ep-s
337	SLC9A6	300231	Ep-s
338	SMARCA2	600014	Ep-s
339	SMC1A	300040	Ep-s
340	SMS	300105	Ep-s
341	SNAP25	600322	Ep-s
342	SNIP1	608241	Ep-s
343	SNORD118	616663	Ep-r
344	SOX10	602229	Ep-s
345	SPATA5	613940	Ep-s
346	SPTAN1	182810	Ep-s
347	SRPX2	300642	Ep-s
348	ST3GAL3	606494	Ep-s

349	ST3GAL5	604402	Ep-s
350	STX1B	601485	Ep-s
351	STXBP1	602926	Ep-s
352	SUMF1	607939	Ep-s
353	SUOX	606887	Ep-s
354	SURF1	185620	Ep-s
355	SYN1	313440	Ep-s
356	SYNGAP1	603384	Ep-s
357	SYNJ1	604297	Ep-s
358	SYP	313475	Ep-s
359	SZT2	615463	Ep-s
360	TAF1	313650	Ep-s
361	TBC1D24	613577	Ep-s
362	TBCD	604649	Ep-s
363	TBCE	604934	Ep-s
364	TBCK	616899	Ep-s
365	TBL1XR1	608628	Ep-s
366	TCF4	602272	Ep-s
367	TPP1	607998	Ep-s
368	TREX1	606609	Ep-s
369	TSC1	605284	Ep-s
370	TSC2	191092	Ep-s
371	TTC19	613814	Ep-m
372	TUBB3	602661	Ep-s
373	TUBB4A	602662	Ep-s
374	TWINK	606075	Ep-s
375	UBA5	610552	Ep-s
376	UBE2A	312180	Ep-s
377	UBE3A	601623	Ep-s
378	UNC80	612636	Ep-s
379	VAR2	612802	Ep-s
380	VPS13A	605978	Ep-s
381	WDR26	617424	Ep-s
382	WDR45	300526	Ep-s
383	WWOX	605131	Ep-s

384	YY1	600013	Ep-s
385	ZDHHC9	300646	Ep-s
386	ZEB2	605802	Ep-s
387	ZFYVE26	612012	Ep-s

Abbreviations: genes associated with epilepsy, epileptic syndromes and seizures (Ep-s), genes related to epilepsy (Ep-r), genes related to mitochondrial diseases and epilepsy (Ep-m).