

Supplementary Table 1 Summary for targeted genomic sequencing data of five cases and one control

sample	Family 1			Family 2	Family 3	Family 4
	III-15*	II -4	III-14	IV-2	II -2	III-1
Bases (Mb)	2874.44	2922.26	2987.40	2635.44	2953.09	3390.09
Map bases (Mb)	2665.19	2725.40	2773.49	2450.51	2752.09	3144.94
Map bases rate (%)	92.7	93.3	92.8	93.0	93.2	92.8
Average read length (bp)	85.58	86.17	85.96	85.71	86.08	85.8
Average sequencing depth	184.67	187.97	191.45	168.72	189.87	216.92
Covered length (Mb)	13.18	13.18	13.18	13.18	13.18	13.17
Coverage at least 20× (%)	98.9	99.0	99.0	98.9	98.9	99.0

* Healthy control did not carry the risk haplotype we previously identified.

Supplementary Table 2 Identification of the causal gene by targeted genomic sequencing

Filter	Family 1		Family 2	Family 3	Family 4
	II -4	III-14	IV -2	II -2	III-1
NS/SS/I	160	157	155	149	158
Not in dbSNP129	30	30	38	31	35
Not in dbSNP129, nor in 1000 Genomes	24	22	26	20	27
Not in dbSNP129, 1000 Genomes, nor in one control	15	21	15	17	22

NS, nonsynonymous variant; SS, splice-site acceptor or donor variants; I, coding insertions or deletions.

Variants were excluded by dbSNP129, 1000 Genomes, one healthy control (from family 1 did not carry the risk haplotype we previously identified) or all. With loss-of-function filter, two genes were remaining in more than one family.

Supplementary Table 3 Clinical characteristics of sp affected individuals

No.	Gender	Age	Age at onset	Trigger	Duration of attacks (s)	Involuntary movement	Response to anticonvulsants	Complication	Family history
1	F	18	11	SM	60	C	No treatment	None	None
2	F	22	12	SM	<10	D	CBZ+	None	None
3	M	21	15	SM	30	D	CBZ+	None	None
4	M	23	14	SM	2 -3	D	CBZ+	None	None
5	M	20	10	SM	10	D	No treatment	None	None
6	M	22	10	SM	10	C	No treatment	None	None
7	M	29	18	SM	7-8	C/D	CBZ+	None	None
8	F	22	13	SM	30	D	CBZ+	None	None
9	M	23	16	SM	15	C/D	VPA+	None	None
10	M	22	13	SM	3-4	C/D	Haloperidol+	None	None

11	M	24	7	SM	10	D	No treatment	None	None
12	F	21	10	SM	30	D	No treatment	None	None
13	M	26	15	SM	2-3	D	CBZ+	Epilepsy	None
14	M	35	9	SM	10-60	D	CBZ+	Epilepsy	None
15	F	19	11	SM	15	D	No treatment	Cerebral dysgenesis	None
16	M	32	15	S/SM	10	D	TPM+	None	None
17	M	20	5	SM	10-60	D	No treatment	IC	None
18	M	24	13	SM	5	D	CBZ+	None	None
19	M	23	12	SM	30	C	CBZ+	None	Brother
20	M	20	12	S/SM	30	D	CBZ+	Migraine, epilepsy	None
21	M	28	7	S/SM	60	C	CBZ+	None	None
22	M	15	8	SM	15	D	No treatment	None	None
23	F	18	12	SM	20	D	CBZ+	None	None
24	M	16	7	S/SM	15	D	No treatment	None	None
25	M	26	13	SM	10	C	No treatment	None	Maternal uncle
26	M	23	10	SM	20	D	No treatment	None	None
27	M	23	13	SM	10-30	D	CBZ+	None	None
28	M	32	10	SM	60	C/D	CBZ+	None	None
29	M	24	7	SM	3-5	C	CBZ+	None	None

F, female; M, male; S, startle; SM, sudden movement; C, choreoathetotic; D, dystonic; CBZ, carbamazepine; VPA, sodium valproate; TPM, topiramate; IC, infantile convulsion.