

Supplementary Table 1. Description of the genetic and genetic-structural etiology in children with infantile epileptic spasms syndrome (N=75)

Chromosomal disorder (N=23)	No.	Classification
Down syndrome	8	G
Miller-Dieker syndrome	3	G-S
15q duplication syndrome	3	G
2q21.2q22.3 microduplication	1	G
2p25 microduplication	1	G
4p16.1 microdeletion	1	G
6q14.3 duplication	1	G
9q34.3 deletion	1	G
Trisomy 9 mosaicism	1	G-S
13q14.11 microduplication	1	G
15p11.2p13 microdeletion	1	G-S
16q23 microdeletion	1	G
Nuclear gene disorder (N=52)	No.	Classification
Tuberous sclerosis complex	35	G-S
Neurofibromatosis type 1	2	G
<i>CASK</i> c.173-1G>A	1	G-S
<i>CDKL5</i> c.1675C>T, p.Arg559* <i>CDKL5</i> (no variant information)	2	G
<i>CHD7</i> c.7891C>T, p.R2631*	1	G-S
<i>DYNC1H1</i> c.874C>T, p.Arg292Trp	1	G-S
<i>HDAC8</i> c.145G>A, p.Ala49Thr	1	G-S
<i>IKBKG</i> exons 4-10 heterozygous deletion (incontinentia pigmenti)	1	G-S
<i>KMT2E</i> c.3784A>G, p.Arg1262Gly	1	G
<i>PDHA1</i> c.1256_1259dupATCA, p.Trp421fs	1	G
<i>PRRT2</i> c.649dupC, p.Arg217Profs*8	1	G
<i>RAC1</i> c.61A>G, p.Ile21Val	1	G
<i>SCN2A</i> c.2932 T>C, p.F978L	1	G
<i>SCN8A</i> c.3995T>C, p.L1332P	1	G
<i>SIX3</i> c.770G>C, p.Arg257Pro	1	G-S
<i>SLC16A2</i> 2876bp hemizygous deletion (Allan-Herndon-Dudley syndrome)	1	G

G, genetic; G-S, genetic-structural.