

Supplementary Table 2 **Causative variants detected in the 71 patients.**

Patient code	Sex	Gene	Variant	Mode of inheritance	Variant segregation	Variant Classification	ID	ASD	Epilepsy	Phenotype correlation
9916	M	<i>AKT3</i>	NM_005465.7:c.503A>G p.Lys168Arg	AD	Mat	P	x	-	-	Yes
8513	F	<i>ANKRD11</i>	NM_013275.6:c.2408-2412del p.Lys803ArgfsTer5	AD	<i>de novo</i>	P	x	-	-	Yes
10231	F	<i>ANKRD11</i>	NM_013275.6:c.1903_1907del p.Lys635GlnfsTer26	AD	Mat	P	x	-	x	Yes
10462	M	<i>ANKRD11</i>	NM_013275.6:c.4384dupA p.Arg1462LysfsTer92	AD	<i>de novo</i>	P	x	-	x	Yes
88-19	F	<i>ARID1B</i>	NM_017519.3:c.5003_5004insC p.Gln1668HisfsTer38	AD	<i>de novo</i>	P	-	-	x	Yes
212-20	M	<i>ARID1B</i>	NM_017519.3:c.4258_4259insC p.Gly1420AlafsTer109	AD	<i>de novo</i>	P	-	-	x	Yes
482-19	M	<i>ATPIA2</i>	NM_000702.4:c.1091C>T p.Thr364Met	AD	Mat	P	-	-	x	Yes
78-20	F	<i>ATPIA2</i>	NM_000702.4:c.2750C>T p.Thr917Met	AD	<i>de novo</i>	P	-	-	x	Yes
514-21	F	<i>ATPIA2</i>	NM_000702.4:c.904del p.Val302SerfsTer25	AD	nd	P	x	-	-	Yes

<b>10727</b>	M	<b>CACNA1A</b>	NM_000068.4: c.1067T>C p.Leu356Pro	AD	<i>de novo</i>	P	x	-	-	Yes
<b>927-19</b>	F	<b>CACNA1A</b>	NM_000068.4: c.5005C>T p.Arg1669Ter	AD	<i>de novo</i>	P	x	-	x	Yes
<b>8804</b>	M	<b>CACNA1A</b>	NM_000068.4: c.3958G>A p.Asp1320Asn	AD	<i>de novo</i>	P	x	-	x	Yes
<b>928-19</b>	F	<b>CASK</b>	NM_003688.3:c.252_255del p.Asp84GlufsTer20	X-linked	<i>de novo</i>	P	x	-	x	Yes
<b>10635</b>	F	<b>CASK</b>	NM_003688.3:c.1090A>G p.Ser364Gly	X-linked	nd	LP	x	-	-	Yes
<b>10851</b>	M	<b>CDKL5</b>	NM_003159.2:c.2798-1G>A	X-linked	Mat	P	-	x	-	Yes
<b>513-20</b>	F	<b>CDKL5</b>	NM_003159.2:c.484G>T p.Glu162Ter	X-linked	<i>de novo</i>	P	x	x	x	Yes
<b>8325</b>	F	<b>CHD7</b>	NM_017780.4:c.5665+1G>C	AD	nd	P	x	-	-	Yes
<b>10458</b>	M	<b>CREBBP</b>	NM_004380.3: c.3832G>A p.Glu1278Lys	AD	<i>de novo</i>	P	x	-	-	Yes
<b>948-19</b>	M	<b>CTNNB1</b>	NM_001904.4:c.936_936+1insTG p.Leu313CysfsTer13	AD	<i>de novo</i>	P	x	-	-	Yes
<b>8511</b>	M	<b>CTNND2</b>	NM_001332.4: c.313C>T p.Gln105Ter	AD	Mat	P	-	-	-	Partial

<b>9481</b>	F	<b>DDX3X</b>	NM_001356.5:c.1171-1G>T	X-linked	<i>de novo</i>	P	x	-	-	Yes
<b>8836</b>	M	<b>DHCR7</b>	NM_001360.3:c.964-1G>C	AR	Mat	P	x	-	-	Yes
			NM_001360.3:c.523G>A p.Asp175Asn	AR	Pat	P				
<b>765-20</b>	M	<b>EHMT1</b>	NM_024757.5:c.523C>G p.Thr1133Arg	AD	<i>de novo</i>	P	x	x	-	Yes
<b>345-19</b>	F	<b>FOXG1</b>	NM_005249.5: c.694A>G p.Asn232Asp	AD	<i>de novo</i>	P	x	-	x	Yes
<b>213-20</b>	M	<b>FOXPI</b>	NM_032682.6:c.570-1G>C	AD	<i>de novo</i>	P	x	-	x	Yes
<b>749-20</b>	F	<b>FOXPI</b>	NM_032682.6:c.1240dup p.Leu414ProfsTer47	AD	<i>de novo</i>	P	x	-	-	Yes
<b>574-19</b>	F	<b>FOXPI</b>	NM_032682.6:c.777_780del p.Lys259AsnfsTer6	AD	<i>de novo</i>	P	x	x	x	Yes
<b>684-19</b>	F	<b>GABRA1</b>	NM_000806.5:c.641G>A p.Arg214His	AD	<i>de novo</i>	P	-	-	x	Yes
<b>9226</b>	F	<b>GATAD2B</b>	NM_020699.4:c.332G>A p.Arg111Gln	AD	<i>de novo</i>	P	x	-	-	Yes
<b>490-19</b>	F	<b>GDI1</b>	NM_001493.3:c.1036C>T p.Gln346Ter	X-linked	Pat	P	x	-	-	Partial

<b>2-20</b>	M	<b>GNAS</b>	NM_080425.3:c.303_304insT p.Met102TyrfsTer6	AD	nd	P	x	-	-	Yes
<b>446-20</b>	F	<b>GNAS</b>	NM_080425.3:c.214G>A p.Gly716Ser	AD	nd	LP	-	x	-	Yes
<b>796-19</b>	F	<b>GNAS</b>	NM_080425.3:c.2936G>A p.Arg979Gln	AD	Mat	P	x	-	-	Yes
<b>328-19</b>	M	<b>GRIN2B</b>	NM_000834.5: c.1385T>C p.Ile462Thr	AD	<i>de novo</i>	P	x	-	x	Yes
<b>422-20</b>	F	<b>GRIN2B</b>	NM_000834.5:c.286G>A p.Gly96Arg	AD	<i>de novo</i>	P	x	-	-	Yes
<b>947-19</b>	M	<b>KANSLI</b>	NM_001193466.2:c.1653-2A>G	AD	nd	P	-	-	-	Yes
<b>275-19</b>	M	<b>KANSLI</b>	NM_001193466.2:c.2021A>G p.Asp674Gly	AD	Mat	P	-	x	-	Partial
<b>8579</b>	M	<b>KCNQ2</b>	NM_172107.4:c.1090G>C p.Glu364Gln	AD	Pat	LP	x	-	x	Yes
<b>525-21</b>	M	<b>KCNQ2</b>	NM_172107.4:c.641G>A p.Arg214Gln	AD	<i>de novo</i>	P	-	-	x	Yes
<b>10617</b>	M	<b>KDM5B</b>	NM_006618.5:c.3133C>T p.Arg1045Ter	AD	Pat	P	x	-	-	Yes
<b>1002-19</b>	M	<b>KDM6A</b>	NM_021140.3:c.1952C>A p.Ser651Ter	X-linked	Mat	P	x	-	-	Yes

<b>32-19</b>	F	<b>MECP2</b>	NM_004992.4:c.397C>T p.Arg133Cys	X-linked	<i>de novo</i>	P	x	-	x	Yes
<b>257-19</b>	F	<b>MED13L</b>	NM_015335.4:c.4963delG p.Glu1655LysfsTer5	AD	<i>de novo</i>	P	x	-	-	Yes
<b>7790</b>	F	<b>NAA10</b>	NM_003491.4:c.384T>A p.Phe128Leu	X-linked	<i>de novo</i>	P	x	-	x	Yes
<b>254-19</b>	F	<b>NALCN</b>	NM_052867.4:c.2294G>C p.Arg765Thr	AD	Pat	LP	-	-	x	Yes
<b>139-20</b>	F	<b>NEXMIF</b>	NM_001008537.3:c.2552C>A p.Ser851Ter	X-linked	<i>de novo</i>	P	x	-	x	Yes
<b>9322</b>	M	<b>NLGN1</b>	NM_014932.5:c.1156G>T p.Gly386Ter	AD	Pat	P	x	-	-	Partial
<b>494-19</b>	F	<b>PAK3</b>	NM_001128168.3: c.500C>G p.Thr167Arg	X-linked	Mat	LP	-	-	x	Yes
<b>781-20</b>	F	<b>PHIP</b>	NM_017934.7:c.1096G>T p.Asp366Tyr	AD	<i>de novo</i>	P	x	-	-	Yes
<b>85-19</b>	F	<b>PRRT2</b>	NM_145239.3:c.649dupC p.Arg217ProfsTer8	AD	Mat	P	-	-	x	Yes
<b>9574</b>	M	<b>RAI1</b>	NM_030665.4:c.2000T>C p.Leu667Pro	AD	<i>de novo</i>	LP	x	-	-	Yes
<b>10488</b>	F	<b>SCN1A</b>	NM_001165963.4:c.2266A>T p.Lys756Ter	AD	nd	P	-	-	x	Yes

<b>10457</b>	F	<b>SCN1A</b>	NM_001165963.4: c.695-6_695-3del	AD	<i>de novo</i>	P	x	-	x	Yes
<b>11-19</b>	F	<b>SCN1A</b>	NM_001165963.4: c.4793A>T p.Tyr1598Phe	AD	Mat	LP	-	x	-	Partial
<b>88-20</b>	F	<b>SCN1A</b>	NM_006920.6:c.5104G>T p.Asp1702Tyr	AD	<i>de novo</i>	P	-	x	-	Partial
<b>10463</b>	F	<b>SCN8A</b>	NM_014191: c.4594A>T p.Ile1532Phe	AD	<i>de novo</i>	P	x	-	x	Yes
<b>81-19</b>	M	<b>SETD1B</b>	NM_001353345.2:c.5291_5292insA p.Asn1765GlnfsTer9	AD	<i>de novo</i>	P	x	-	x	Yes
<b>9137</b>	M	<b>SHANK2</b>	NM_133266.5:c.769C>T p.Arg257Ter	AD	<i>de novo</i>	P	x	-	-	Yes
<b>9320</b>	F	<b>SLC2A1</b>	NM_006516.4:c.388G>A p.Gly130Ser	AD	Mat	P	-	-	x	Yes
<b>483-19</b>	M	<b>SLC2A1</b>	NM_006516.4:c.988C>T p.Arg330Ter	AD	<i>de novo</i>	P	x	-	x	Yes
<b>8697</b>	M	<b>SLC2A1</b>	NM_006516.4:c.85_89del p.Asn29TrpfsTer58	AD	nd	P	-	-	x	Yes
<b>10360</b>	F	<b>SLC2A1</b>	NM_006516.4: c.967G>A p.Val323Met	AD	Mat	LP	-	-	x	Yes
<b>12-21</b>	F	<b>SLC2A1</b>	NM_006516.4:c.109C>T p.Gln37Ter	AD	nd	P	-	-	x	Yes

<b>1-19</b>	F	<b>SLC9A6</b>	NM_001042537.1:c.264C>G p.Ile88Met	X-linked	<i>de novo</i>	LP	x	x	-	Yes
<b>84-19</b>	F	<b>SMARCC2</b>	NM_003075.5:c.2984dupC p.Ala996CysfsTer16	AD	nd	P	x	-	x	Yes
<b>896-19</b>	F	<b>SYNGAP1</b>	NM_006772.3:c.509G>A p.Arg170Gln	AD	<i>de novo</i>	P	-	x	x	Yes
<b>559-19</b>	M	<b>TPO</b>	NM_000547.5:c.1472G>A p.Arg491His	AR	Mat/Pat	P	x	-	-	Yes
<b>298-19</b>	M	<b>TBX1</b>	NM_080646.1:c.1010-2A>G	AD	nd	P				Yes
		<b>UBE3A</b>	NM_000462.5: c.2576_2579del p.Lys859ArgfsTer4	AD	nd	P	x	-	-	Yes
<b>10456</b>	M	<b>VPS13B</b>	NM_017890.4:c.9406-1G>T	AR	Pat	P				Yes
			del ex4-7	AR	Mat	P	x	-	-	Yes
<b>211-20</b>	M	<b>ZMYM2</b>	NM_197968.4:c.1765C>T p.Arg589Ter	AD	Pat	P	x	x	-	Yes
<b>8634</b>	F	<b>ZMYM2</b>	NM_197968.4:c.474dup p.Ser159GlnfsTer6	AD	<i>de novo</i>	P	x	-	-	Yes

**Abbreviations:** AD, autosomal dominant; AR, autosomal recessive; ASD, autism spectrum disorder; F, female; ID, intellectual disability; LP, likely pathogenic; M, male; Mat, maternal; nd, not determined; P, pathogenic; Pat, paternal.