

## SUPPLEMENTARY DATA

**Table e1:** Example of patient diary.

		Type of symptoms	Duration of symptoms (min)	Severity of symptoms
<b>Motor events</b>	Convulsion*			
	Seizure*			
	Stiffness			
	Abnormal movement	X	5	2
	Tremor			
	Speech disorder			
	Abnormal eye movements			
	Partial/total weakness of a limb			
<b>Non-motor events</b>	Fatigue			
	Disorientation			
	Headaches			
	Sleep disorder			
	Mood switch			
	Other			
<b>Severity code</b>				
<b>1</b>	No change in daily activity; no need for medication or medical intervention			
<b>2</b>	Partial change in daily activity while medication or medical intervention is not mandatory			
<b>3</b>	Change in daily activity with medication or medical intervention needed			

\* No patient reported convulsions/seizures during the course of this study.

**SUPPLEMENTARY DATA**

**Table e2:** Main characteristics of patients with GLUT1 deficiency and detailed response to treatment.

		<b>Patients</b>						
		<b>P1</b>	<b>P2</b>	<b>P3</b>	<b>P4</b>	<b>P5</b>	<b>P6</b>	
<b>Gender</b>		F	F	M	M	M	M	
<b>Age</b>		23	20	7	14	16	47	
<b>Age of onset</b>		1.5	1.5	2	3.5	2.5	14	
<b>CSF/plasma glucose</b>		0.50	0.40	0.44	-	0.42	-	
<b>SLC2A1 mutation</b>		c.880T>C	c.425T>A	c.940G>A	c.34C>T	c.34C>T	c.34C>T	
		p. Ser294Pro	p. Met142Lys	p. Gly314Ser	p. Leu12Phe	p. Leu12Phe	p. Leu12Phe	
<b>Main permanent manifestations</b>		Cerebellar ataxia; attention deficit	Mental retardation; cerebellar ataxia	Learning disability; mild ataxia; pyramidal syndrome	Segmental dystonia; pyramidal syndrome	Generalized dystonia; pyramidal syndrome	Pyramidal syndrome	
<b>Main paroxysmal manifestations</b>		Dystonia; headache	Dyskinesia	Dyskinesia; ocular movement; headache	Fatigue; headache	Dyskinesia; fatigue; headache	Dystonia; fatigue; headache	
<b>Baseline</b>	<b>Motor</b>	Hyperkinetic movement disorders	3	14	4	0	12	54
		Other <sup>a</sup>	3	2	6	1	0	0

<sup>a</sup> speech disorder, partial/total weakness of a limb, abnormal eye movement; <sup>b</sup> disorientation, headache, mood switch.

**SUPPLEMENTARY DATA**

**Table e2:** continued

			<b>Patients</b>					
			<b>P1</b>	<b>P2</b>	<b>P3</b>	<b>P4</b>	<b>P5</b>	<b>P6</b>
<b>Baseline</b>	<b>Non motor</b>	Fatigue	1	1	2	6	16	2
		Headache	1	1	0	7	3	2
		Other <sup>b</sup>	2	2	1	12	0	27
	<i>Total</i>		<i>10</i>	<i>20</i>	<i>13</i>	<i>26</i>	<i>31</i>	<i>85</i>
<b>Treatment</b>	<b>Motor</b>	Hyperkinetic movement disorders	2	7	0	0	1	0
		Other <sup>a</sup>	0	0	3	0	0	0
		<i>Total</i>		<i>2</i>	<i>7</i>	<i>3</i>	<i>0</i>	<i>1</i>
	<b>Non motor</b>	Fatigue	0	0	0	0	0	0
		Headache	0	0	2	0	0	0
		Other <sup>b</sup>	2	0	0	0	0	0
<i>Total</i>		<i>2</i>	<i>0</i>	<i>2</i>	<i>0</i>	<i>0</i>	<i>0</i>	
<b>Withdrawal</b>	<b>Motor</b>	Hyperkinetic movement disorders	9	9	1	1	15	39
		Other <sup>a</sup>	0	0	0	0	0	1
		<i>Total</i>		<i>9</i>	<i>9</i>	<i>1</i>	<i>1</i>	<i>15</i>
	<b>Non motor</b>	Fatigue	0	1	3	4	16	8
		Headache	1	0	1	15	5	7
		Other <sup>b</sup>	0	1	0	0	0	8
<i>Total</i>		<i>1</i>	<i>1</i>	<i>4</i>	<i>19</i>	<i>21</i>	<i>15</i>	
<i>Total</i>		<i>10</i>	<i>11</i>	<i>5</i>	<i>20</i>	<i>36</i>	<i>63</i>	

<sup>a</sup> speech disorder, partial/total weakness of a limb, abnormal eye movement; <sup>b</sup> disorientation, headache, mood switch.