

Supplementary Table 2. Clinical features of individuals with homozygous variant c.1287+5G>A in TRAPPC11.

Abbreviations: M = male; F = female; y= years; IU= International Units; FM-IA= Focal motor with impaired awareness seizure; FNM-IA= Focal non-motor with impaired awareness seizure; FBTC= Focal onset to bilateral tonic-clonic seizure; GM= Generalized onset myoclonic seizures; GT= Generalized onset tonic seizures; BTC= Bilateral tonic-clonic seizures; SW= Spike-waves; NA/NP= Not available/Not performed.

Ind	Fam	Sex	Age last exam	Intellectual disability	CK (IU)	Muscle weakness	Muscle biopsy	Movement disorder	Seizure onset	Infection-related seizures	Seizure types	Status epilepticus	Epilepsy course (Brodie class)	Current antiseizure medication	EEG	Brain MRI
1	1	F	13-16y	Yes (severe)	300-600	Yes	Not performed	No	6 m	Yes	BTC + GT	No	A	LEV	Normal background. Multifocal spikes, sharp, slow and SW, mainly left centro-parieto-temporal.	Normal
2	1	F	9-12y	Yes (severe)	300-1100	Yes	Not performed	Yes	6 m	Yes	FM-IA + FNM-IA + FBTC	Yes (several status with fever)	C	LEV	Normal background. Focal sharp waves in right fronto-temporal.	Generalized decrease in cortico-subcortical volume.
3	2	M	17-20y	Yes (moderate)	500-1400	Yes	Not performed	Yes	4 m	Yes	BTC + FBTC + FM-IA	Yes (febrile status epilepticus)	C	LEV+ OXC	Normal background. Multifocal SW, mainly left parieto-temporal.	Normal
4	3	M	5-8y	Yes (moderate)	500-800	Yes	Unspecific myopathic pattern	Yes				No epilepsy			Normal (2x)	Normal
5	3	M	5-8y	Yes (moderate-severe)	300-500	Yes	Unspecific myopathic pattern	Yes	14 m	Yes	FBTC + BTC	Yes (Febrile status epilepticus)	B	LTG + VPA + CLB	Slow background. Multifocal SWs, mainly left parieto-occipital and right parieto-temporal.	Generalized decrease in cortico-subcortical volume.

6	4	F	13-16y	Yes (moderate-severe)	450	Yes	Not performed	Yes								No epilepsy	Generalized decrease in cortico-subcortical volume.
7	4	M	9-12y	Yes (mild)	NA/NP	Yes	Not performed	Yes								No epilepsy	NA/NP
8	5	F	13-16y	Yes (moderate)	800-3000	Yes	Unspecific myopathic pattern	Yes	6 m	Yes	BTC	Yes (Febrile status epilepticus)	A	LEV	Normal background. Multifocal SWs, mainly frontal. No photosensitivity.	Generalized decrease in cortico-subcortical volume.	
9	6	F	9-12y	Yes (severe)	350-750	Yes	Not performed	Yes	15 m	No	BTC	No	C	LEV	Normal background. Multifocal discharges.	Periventricular leukomalacia (history of prematurity)	
10	6	M	21-24y	Yes (severe)	3500	No	Not performed	Yes	7y	NA	BTC	No	B	No	Slow background. Some EEG with multifocal activity: spikes in the right centrotemporal region and left centrotemporal region.	Normal	
11	6	M	5-8y	Yes (moderate)	700-1000	No	Unspecific myopathic pattern	Yes								No epilepsy	Normal
12	7	F	5-8y	Yes (moderate)	1700-4500	Yes	Performed but not revised	Yes								No epilepsy	Normal
13	8	F	9-12y	Yes (severe)	820-1500	Yes	Unspecific myopathic pattern	Yes	4m	Yes	GM + FM-IA	No	C	VPA + OXC	Focal discharges (left centro-temporal)	Generalized decrease in cortico-subcortical volume.	
14	9	F	17-20y	Yes (moderate)	600-5000	No	Unspecific myopathic pattern	No								No epilepsy	Normal
15	10	M	5-8y	Yes (moderate)	1950	Yes	Not performed	Yes								No epilepsy	Bilateral parieto-occipital corticosubcortical volume decrease with

16	10	M	13-16y	Yes (severe)	2970	Yes	Not performed	Yes				No epilepsy			Slow background activity	prominence of cerebellar folia and thin corpus callosum. NA/NP
17	11	M	21-24y	Yes (severe)	1660	No	Unspecific myopathic pattern	Yes	13 m	Yes	BTC	Yes	A	No	Slow background activity. Epileptic abnormalities not detected	Normal
18	12	F	9-12y	Yes (moderate-severe)	260-780	No	Not performed	Yes	12m	Yes	BTC	Yes	A	No	Normal background activity. Epileptic abnormalities not detected	Normal
19	13	M	5-8y	Yes (moderate-severe)	370	No	Not performed	Yes					No epilepsy			Normal
20	14	F	13-16y	Yes (severe)	2450	Yes	Not performed	No					No epilepsy			Normal
21	15	M	2-4y	≤ 4 years old	1035	No	Not performed	Yes	14m	Yes	BTC	No	A	LEV	Slow background activity. Multifocal discharges.	Generalized decrease in cortico-subcortical volume.
22	16	M	13-16y	Yes (grade unknown)	1200-2400	Yes	Not performed	Yes					No epilepsy			Megacisterna magna
23	17	M	2-4y	≤ 4 years old	550	Yes	Not performed	No					No epilepsy			Not performed
24	17	F	9-12y	Yes (grade unknown)	330-1720	Yes	Not performed	No					No epilepsy			Not performed
25	18	F	17-20y	Yes (moderate-severe)	370	Yes	Not performed	Yes	8y	Yes	FM-IA + BTC	No	A	LEV	Slow background activity. Epileptic abnormalities not detected	Normal