Supplementary Table 2: Clinical features of all patients

Subject	Sex (M/F)	Age Last Seen	Age of Onset	Seizures types	Epilepsy	Treatment Resistant	ID/GDD/Autism/ other	Family History	MRI	Other
001P	M	18 mos	7.5 mos	Focal motor	Dravet	Yes	Mild speech/ language delay	negative	9 mos : Normal	
002R	M	3 yr	3.2 mos	Focal motor	Dravet-like	Yes	GDD	Mat=migraines and epilepsy	9 mos and 3yr 5 mos: slightly thin CC 3 yr 6 mos: left Hemisphere swelling; right atrophy 3 yr 10 mos: bilateral atrophy (Left>Right)	Mild hemiparesis
003R	F	6 yr	Neonate and 30 mos	Focal motor to bilateral TC	EE	Yes	ADHD	Mat: febrile seizures	Day 3: normal 6 yr: prominent perivascular spaces	Perinatal: hypoglycemia & respiratory distress
004R	F	3 yr	21 mos	Focal motor	CSWS	Yes	Regression; language disorder	Mat: epilepsy Pat: epilepsy	2 &3 yr: normal	Born at 34 weeks
005R	F	5 yr	12 mos	Epileptic Spasms	West syndrome	Yes	GDD/ID	Negative	1 & 9 yr: normal	Microcephaly, CVI, hypotonia, hyperkinetic movement disorder
006R	M	4 yr	36 mos	Generalized	LGS	Yes	GDD/ID	Mat: epilepsy	3 yr: prominant extra-axial spaces	Macrocephaly, ataxia
007R	M	4 yr	36 mos	Generalized	MAE	Yes	Mild speech/ language delay	Mat: seizure Pat: school difficulties	3 yr: normal	
010P	F	3 yr	23 mos	Focal motor	Unclassified	Yes	GDD	Pat: epilepsy	2 yr: normal	In utero exposure; hypotonia
011R	F	12 yr	24 mos	Focal motor	Unclassified	Yes	Anxiety/ depression/OCD	Sibs: FS GPs: migraine	8 & 12 yr: normal	migraine
012R	M	19 yr	3 mos	Epileptic spasms Generalized	West to LGS	Yes	GDD/ID	Sibs: ADHD	4 mos; 2 & 3 yr: normal	CVI, spasticity, hyperkinetic movement disorder
015P	M	20 mos	7 mos	Epileptic spasms	West syndrome	Yes	GDD	Negative	7 mos: normal	Hypotonic
016R	F	22 yr	36 mos	Focal and generalized	EE	Yes	GDD/ID/Autism/ anxiety	Pat: epilepsy	8 & 14 yr: normal	Hypotonic, ataxia
018P	F	6 mos	2 mos	Focal epileptic spasms, tonic	Ohtahara, West	Yes	GDD	Negative	2 mos: normal	Facial dysmorphism, axial hypotonia, CVI, strabismus
019R	**F**	**12 yr**	**30 mos**	**Generalized**	**Unclassified**	**Yes**	**GDD/ID/Autism/ADHD/OCD**	**Adopted**	**2 & 10 yr: normal**	**VPA exposure in utero** **Facial dysmorphism, strabismus, bicuspic AV, scoliosis**
023R	M	9 yr	12 mos	Focal and Generalized	EE	Yes	GDD/ID	Consanguinity: 2nd degree	11 mos, 2 & 5 yr: left occipital lobe focal white matter signal abnormality	Hypotonic, ataxic

024R	F	11 yr	60 mos	Focal	LKS	Yes	GDD/ID/ regression/ADHD	Adopted	8 yr: mild PVL	Born at 34 weeks In utero exposure
025R	M	19 yr	4 mos	Focal	Unclassified	Yes	Mild LD, DCD, anxiety	Cousin: FS	5 mos, 8 & 13 yr: normal	
026R	F	5 yr	40 mos	Generalized Epileptic spasms	EE	Yes	GDD/ID	Mat: epilepsy Parent: migraine	4 yr: normal	
027R	F	2 yr	24 mos	Focal Generalized	Febrile Seizures plus	Yes	normal	GP: epilepsy, FS Sibs: ID	1.5 yr: normal	
030R	M	7 yr	41 mos	Generalized Absence	Childhood absence epilepsy	Yes	OCD, ADHD	Pat: epilepsy	4 yr: normal	
031R	F	3 yr	22 mos	Generalized Myoclonic absence	Unclassified	No	normal	Negative	2.5 yr: normal	
033P	F	16 mos	9 mos	Focal, EPC, myoclonic	EE	Yes	GDD, regression	Pat: FS	9 mos: normal	Liver failure, hypotonic Passed away 16 months
036P	M	1 yr	6 mos	Generalized myoclonic	Unclassified	Yes	GDD	Negative	5 mos: thin CC, arachnoid cyst, basilar kyphosis	Polyhydramnios, postnatal hypoglycemia, hypotonic, cleft palate
037P	F	6 yr	60 mos	Focal	Unclassified	No	GDD, ID, Autism	GP: epilepsy	7 yr: normal	
038R	F	7 yr	13 mos	Focal FS	MTLE with HS	Yes	GDD, ID	Negative	2 yr: Right MTLS 3yr: same + atrophy right hemisphere 4yr: surgical changes with adjacent signal abnormalities 5yr: post 2nd surgical changes & early left HS	Microcephaly, atrial septal defect 2 epilepsy surgeries for HS and FCD III
039R	**M**	**10 yr**	**Day 1** **60 mos**	**Neonatal seizures** **Focal**	**EE**	**Yes**	**GDD, ID, Autism**	**Negative**	**Day 3 &10: focal diffusion restriction left occipital lobe** **2 yr: normal**	**In utero seizures?** **Hypotonic, ataxic**
040R	F	3 yr	Day 3	Neonatal seizures Focal	EE	Yes	GDD, ID	Negative	Day 5: normal 4 yr: white matter signal abnormality and volume loss in cerebral hemispheres	Spasticity, CVI
043R	F	2 yr	3 mos	Focal Epileptic spasms	West syndrome	No	GDD, ID	Negative	3 mos: lissencephaly posterior > anterior	Microcephaly CVI Hypotonic
044R	M	5 yr	1.4 mos	Focal	EE	Yes	GDD, ID, regression	Paternal: epilepsy	7 weeks: normal 2 yr: atrophy; thin CC; abnormal myelination 4 yr: progressive atrophy; abnormal signal BG, thalami	CVI, axial hypotonia, hyperkinetic movement disorder, feeding difficulties
047P	F	4 yr	44 mos	Generalized FS	Unclassified	Yes	Speech/language delay	Father, 2 sibs and Pat cousin: FS	Not done	

050R	M	3 yr	14 mos	Focal	Unclassified	Yes	GDD, ID	Negative	1 yr: complex asymmetric brain malformation 2 yr: same plus right HS	Strabismus, asymmetric pupils , hemiparesis, hemihypertrophy
056R	M	5 yr	32 mos	Generalized Absence	Unclassified	Yes	GDD	Pat: epilepsy	5 yr: normal	macrocephaly
057R	F	19 mos	2 mos	Focal	Unclassified	Yes	GDD	Consanguinity: 1st degree	2 mos: abnormal CC; abnormal signal BG 6 mos: atrophy; abnormal white matter and BG signal	Microcephaly, hypotonic
059P	M	3 yr	2 yr	Focal	Unclassified	Yes	GDD, Autism	Negative	18 mos: possible atrophy 2 yr: delayed myelination	Microcephaly
049R	F	4 yr	13 mos	Generalized focal FS	Febrile seizures plus	Yes	Speech/language delay	Pat: 2nd cousin seizure	2 yr: normal	
062R	M	10 yr	48 mos	Generalized, focal	EE	yes	normal	Negative	5 yr: normal	
063R	M	3 yr	24 mos	Generalized absence myoclonic	Unclassified	Yes	GDD	Negative	2 yr: normal	
061R	**F**	**14 yr**	**4 mos**	**Generalized, focal FS**	**Dravet-like**	**Yes**	**GDD, ID**	**Brother: Dravet-like died Mother, GP, Mat cousin: FS**	**1 yr: normal**	**Microcephaly Oromotor difficulties Ataxic/crouch gait**
064R	F	28 yr	7 mos	Generalized, focal	LGS	Yes	GDD, ID	Negative	5 yr: normal 13 yr: normal	
065R	F	3 yr	3.5 mos	Epileptic spasms	West syndrome	Yes	GDD	Negative	4 mos: delayed myelination 2 yr: Hypomyelination and atrophy	Premature (34 weeks); twin Dysmorphic, limb asymmetry Microcephaly, hypotonic, spasticity
067R	M	16 yr	18 mos	Generalized, focal, epileptic spasms, FS	LGS	Yes	GDD, ID, ASD	Paternal side: migraine and epilepsy Maternal: migraine	1 yr; 2 yr-normal 6 yr: heterotopic grey 7yr: incomplete myelination and heterotopic grey	
069P	M	5 yr	52 mos	Generalized Tonic, tonic-clonic	Unclassified	No	GDD, ID	Brother similarly affected	3 yr: normal	Spastic CP, dystonia, CVI, microcephaly, G-tube fed
071R	F	7 yr	46 mos	Focal, FS	Unclassified	Yes	GDD, Autism	Negative	2 yr: delayed myelination small thoracic hydromyelia 4 yr: normal head hydromyelia smaller 8 yr: normal head; minimal hydromelia	Hypotonia, microcephaly, hand stereotypies, intermittent hyperventilation, minor anomalies, scoliosis
074R	M	5 yr	44 mos	Generalized myoclonic, tonic	Unclassified	Yes	Normal	Negative	4 yr: prominent subarachnoid spaces	macrocephaly
077R	F	10 yr	2 mos	Epileptic spasms, focal, tonic-clonic	West syndrome, LGS	Yes	GDD, ID	Negative	2, 4, 10 mos: normal 1 year: possible atrophy 4 yr: normal except post epilepsy surgery changes	Progressive microcephaly CVI, spastic CP, hyperkinetic movement disorder, cyclic vomiting

087R	F	2 yr	10 mos	Generalized, focal, tonic-clonic, myoclonic	Unclassified	Yes	GDD, ID	Negative	9 mos: normal 2 yr: diffuse cerebral edema	Episode of status epilepticus associated with significant brain edema with worsening
104P	M	6 mos	2 weeks	Focal	Self-limited familial neonatal epilepsy	No	Normal	Neonatal seizure: mother, Mat uncle, grandmother, & first cousin	Not done	GERD
120P	**F**	**7 mos**	**2 mos**	**Epileptic spasms**	**West syndrome**	**Yes**	**GDD**	**Negative**	**2 mos: swelling frontal and parietal gyri, diffusely abnormal white matter signal**	**Microcephaly, hypotonia, hyperkinetic movement disorder** **Passed away 16 months**
125P	F	5 mos	1 week	Focal	Unclassified	Yes	borderline to normal development	Paternal uncle FS; paternal cousin epilepsy	Day 5: abnormal white matter signal in both cerebral hemispheres, restricted diffusion in basal ganglia and thalami, swelling of pons, mild lactate peak Day 13: bilateral white matter signal abnormality in cerebral hemispheres; abnormal signal in basal ganglia and thalami	Low Apgar scores, sensorineural hearing loss, G-tube fed
106R	F	17 yr	10 mos	Focal	EE	Yes	GDD, ID	Maternal aunt: epilepsy and died in infancy	2 yr: normal 4 yr: nonspecific abnormal white matter signal 8 yr: normal	Cerebral palsy, exotropia, scoliosis

ADHD=attention deficit hyperactivity disorder; AV=atrial valve; BG=basal ganglia; CC=corpus callosum; CP=cerebral palsy; CSWS=epileptic encephalopathy continuous spike-and-wave during sleep; CVI=cortical visual impairment; DCD=developmental coordination disorder; EE=unspecified epileptic encephalopathy; EPC=epilepsia partialis continua; F=female; FCD=focal cortical dysplasia; FS=febrile seizures; GDD=global developmental delay; GP=grandparent; ID=intellectual disability; LD=learning disorder; LGS=Lennox-Gastaut syndrome; LKS=Landau-Kleffner syndrome; M=male; MAE=myoclonic atonic epilepsy; Mat=maternal; mos=months; MTLE with HS=mesial temporal lobe epilepsy with hippocampal sclerosis; OCD=obsessive compulsive disorder; P=prospective; Pat=paternal; TC=tonic-clonic; R=retrospective; yr=year;