Supplementary data :

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	Non responder	Partial responder	Responder
Year of paper publication			
< 2015	12 (86%)	1 (7%)	1 (7%)
2015-2020	27 (70%)	3 (7%)	9 (23%)
2020-2025	21 (60%)	1 (3%)	13 (37%)
Continent			
Europe	18 (60%)	2 (7%)	10 (33%)
Asia	24 (75%)	1 (3%)	7 (22%)
North America	14 (74%)	0	5 (26%)
Souht America	0	0	1
Australia	3 (60%)	2 (40%)	0
Africa	1	0	0
Type of pathology reported			
Early-onset epilepsy	43 (67%)	1 (1%)	20 (32%)
Late-onset epilepsy	9 (64%)	3 (22%)	2 (14%)
Both	8 (80%)	1 (10%)	1 (10%)
Team with expertise in the team (> 1 publication on KCNT1)			
Yes	24 (65%)	3 (8%)	10 (27%)
No	36 (70%)	2 (4%)	13 (26%)

Supplementary Table 1: **Comparison of demographics of responders vs. non-responders**

A

Paper reference (ex: Nabbout et al. 2024)
Patient identification in your paper (ex: patient 1) - *please use the same anonymisation that you used for the article*
Age at last visit (year)
Death since the publication (Y/N)
If yes, age at death (years)
If yes, cause of death
Head circumference (OFC) at last visit (SD)
Active epilepsy at last follow-up (> 1 seizure/year) (Y/N)
If yes, add frequency
Seizure types at last follow-up (generalized/focal/both/unknown)
ASM at last follow-up? (Y/N)
If yes, please detail them (ex: sodium valproate, lamotrigine, Cannabidiol…)
KD at last follow-up? (Y/N)
Epilepsy surgery since the publication ? (Y/N)
If yes, age at surgery
Type of surgery (resection/VNS…)
Feeding disorders (Y/N)
G tube feeding (Y/N)
If other feeding issues, describe them
Vascular malformations? (Y/N/unexplored)
If yes, revelation mode (screening, incidental finding, symptomatic etc)
If any other cardiovascular information, describe them
Precocious puberty ? (Y/N/too young)
New brain MRI since publication? (Y/N)
Brain MRI results (normal/abnormal: main findings)
Possibility of sending brain MRI images to us? (Y/N: CD, web link, etc)
Neurodeveloppement at last Follow-Up? (normal/slight, moderate, severe delay)
Mode of neurodevelopment evaluation (clinically, scales…)
Orthopedic disorders ? (Y/N)
If yes, detail orthopedic disorders (hip spasticity/scoliosis…)
Surgery done for orthopedic disorders or scheduled ? (Y/N)
If yes, detail which surgery
Autism spectrum disorder ? (Y/N)

If yes, which critere/scale of diagnosis (ADOS, DSM….)
Behavioral disorders (Y/N)
Psychiatric therapies (Y/N)
If yes, detail them
Sleep disorders (Y/N)
Melatonin (Y/N)

Any other information ? Feel free to add it there

B

Paper reference (ex: Nabbout et al. 2024)
Patient identification in your paper (ex: patient 1) - *please use the same anonymisation that you used for the article*
Age at last visit (year)
Active epilepsy at last follow-up (> 1 seizure/year) (Y/N)
If yes, add frequency
Seizure types at last follow-up (generalized/focal/both/unknown)
ASM at last follow-up? (Y/N)
If yes, please detail them (ex: sodium valproate, lamotrigine, Cannabidiol…)
Neurodeveloppement at last follow-up? (normal/slight, moderate, severe delay)
Mode of neurodevelopment evaluation (clinically, scales…)
Autism spectrum disorder ? (Y/N)
If yes, which critere/scale of diagnosis (ADOS, DSM….)
Other psychiatric disorder at last FU (Y/N)
If yes, precise type (depression/anxiety/behavior disorders…)
Psychiatric therapies (Y/N)
If yes, detail them

Supplementary Table 2: Questionnaire sent to clinicians

A- For EIMFS and non-EIMFS DEE patients

B- For SHE and other patients

ADOS: Autism Diagnostic Observation Schedule, ASM: AntiSeizure Medications, DEE: Developmental and Epileptic Encephalopathy, DSM: Diagnostic and Statistical Manual of Mental Disorders, EIMFS: Epilepsy of Infancy with Migrating Focal Seizures, KD: Ketogenic Diet, MRI: Magnetic Resonance Imgaing, OFC: Occipital-Frontal Circumference, SD: Standard Deviation, SHE: Sleep-related Hypermotor Epilepsy, VNS = Vagus Nerve Stimulation

Patient's reference	Variant c. / p. / inheritance	Epilepsy syndrome	Sex	Age at publication (y)	Sz onset (m)	Sz frequency at publication	Sz types (global) at publication	MRI at publication	Cognition at publication	Age at last visit (y)	Death since the publication (age (y) ; cause)	Active epilepsy at LFU	Seizure frequency at LFU	Seizure types at LFU	ASM at LFU ?	KD at LFU?	Epilepsy surgery since the publication ? (age (y) and type)	New brain MRI since publication ? (results)	Neurodeveloppement at LFU?
Bonardi et al. 2021[1] (10)	c.862G>A / p.Gly288Ser / De novo	EIMFS	M	1,4	6	Multiple daily	HemiC, TAS, eTCS, SE	WMA, frontal gliosis, thin CC and brainstem, fronto- temporo-insular CA, cerebellar atrophy, enlarged ventricles	Severe NDD	7	No	Yes	Yearly	GS, FS	Yes (LTG, VPA, PHB)	No	No	No	Severe to profound ID
Bonardi et al. 2021(11)	c.862G>A / p.Gly288Ser / NA	EIMFS	M	0,8	4	NA	HemiC, eTCS, SE	Normal (age NA)	Severe NDD	6	No	Yes	Yearly	GS, FS	Yes (VPA, STP, CLB)	No	No	Yes (3 yo: CA, subdural hygromas)	Severe to profound ID
Bonardi et al. 2021(20°), Fitzgerald et al. 2019[2] (16)	c.1429G>A / p.Ala477Thr / De novo	EIMFS	F	14	0,5	Daily	FS, Subclinical, eTCS, Spasms	Global CA, thin CC	Severe ID	18	Yes (18 ; respiratory infection)	Yes	Weekly	FM, spasms	Yes (Epidyolex, QUIN(weaning off-ultimately came off successfully without worsening seizures), VPA, CZP, BRV, CLB)	Yes, 4:1 ratio	No	No	Severe to profound ID
Bonardi et al. 2021(31)	c.2849G>A / p.Arg950Gln / De novo	EIMFS	M	18,5	5	Weekly	FS, TCS, SE	Bilateral temporal WMA	ID	24	No	Yes	Multiple daily	TS, rare TC	Yes (VPA, PHB, LTG, CBD (not commercial))	No	No	No	Severe to profound ID
Bonardi et al. 2021(33°), Fitzgerald et al. 2019 (6)	c.2881C>A / p.Arg961Ser / De novo	EIMFS	M	2	2	Yearly	HemiC, FIA, Subclinical, TCS, Myo	Normal (age NA)	Severe NDD	11	No	Yes	Yearly	FM (clonic)	Yes (LEV)	No	No	No	Severe to profound ID
Cherian et al. 2021[3] (patient 5) IV-5	c.2882G>A / p.Arg961His / inherited	EIMFS	F	7	3	Weekly	FM (3 m), FNMIA+Au (6 m), FM (4y), Gmyo, unknown onset TC Sz, MFS	5 yo: normal	Severe ID	11	No	Yes	Multiple daily	FM (tonic or clonic)	Yes (BRV, PHE, TPM)	Yes	No	No	Severe to profound ID
Dilena et al. 2018[4] (patient 1)	c.2849G>A / p.Arg950Gln / De novo	EIMFS	M	1	0,06	Multiple daily	Au	1st dol: normal	Severe NDD	7,5	No	Yes	Multiple daily	F migrating (tonic, versive, clonic, autonomic); FTB(TC)	Yes (LEV)	No	No	No	Severe to profound ID
Dilena et al. 2018 (patient 2)	c.2677G>A / p.Glu893Lys / De novo	EIMFS	M	2	0,03	Multiple daily	FM+Au, Spasms	NA	Severe NDD	6	Yes (6 ; severe pulmonary hemorrhage)	Yes	Multiple daily	FS, GS	Yes (QUIN, FBM, CBD, CLB, DZP)	No	No	No	Severe to profound ID
Ferretti et al. 2022[5] (patient 1)	c.337G>A / p.Val113Met / De novo	EIMFS	M	5,75	9	Daily	FM, Spasms	6 mo: thin CC	Severe NDD	7	Yes (7 ; respiratory infection)	Yes	Multiple daily	GS	Yes (TPM, LCS, QUIN, CLZ)	No	No	No	Severe to profound ID
Ferretti et al. 2022 (patient 3)	c.862G>A / p.Gly288Ser / De novo	EIMFS	M	2,5	1,3	NA	FM	2mo : normal	NDD	5	No	Yes	Multiple daily	GS	Yes (CBZ, PHE, QUIN)	No	No	No	Moderate NDD
Ikeda et al. 2021[6] (patient 2), Ohba et al. 2015[7] (patient 2)	c.808C>G / p.Gln270Glu / De novo	EIMFS	F	8	0,006	NA	FM	28 do: thin CC ; 7 mo: diffuse CA ; 35 mo: delayed myelination	Severe ID	14	No	Yes	Multiple daily	FS	Yes (PB, LEV, LTG, KBr)	No	No	No	Severe to profound ID

Kawasaki et al. 2017[8] (patient 3), Ohba et al. 2015 (patient 7)	c.1421G>A / p.Arg474Cys / De novo	EIMFS	M	8	0,5	Multiple daily	FM, Au	7 mo: normal	Severe ID	6,5	No	Yes	Multiple daily	FS, TS, automatic seizure	Yes (PHE)	No	No	No	Severe to profound ID
Kohli et al. 2020[9]	c.1420C>T / p. Arg474Cys / NA	EIMFS	F	0,83	NA	Multiple daily	FM	2 mo: delayed myelination, thin CC	Severe NDD	5	No	Yes	Multiple daily	FS, GS	Yes (PHE, VGV, LEV)	Yes	No	No	Severe NDD
Kravetz et al. 2021[10]	c.2795T>C / p.Phe932Ser / De novo	EIMFS	F	4	3	NA	FM	3 yo: subtle signs of CA, hypoplastic CC, WMA - more prominent in the left hemisphere.	NA	15	No	Yes	Monthly	FTB(TC)	Yes (TPM)	NA	NA	NA	Severe to profound ID
Kuchenbuch et al. 2019[11] (patient 1), Barcia et al. 2012[12] (patient 2), Kim et al. 2014[13] (1)	c.1283G>A / p.Arg428Gln / De novo	EIMFS	M	16,2	2	Weekly	FS, GS	5yo: Myelination delay, thin CC, CA	ID	23	No	Yes	Nighlty	GS (tonic)	No (Rivotril when needed)	No	No	No (CT scan: normal cerebral arteries; minimal bilateral symmetrical cerebellar atrophy; minimal frontal atrophy)	Severe to profound ID
Kuchenbuch et al. 2019 (patient 10)	c.1283G>A / p.Arg428Gln / De novo	EIMFS	F	7	NA	NA	NA	NA	Severe ID	12,4	Yes (13,4 ; respiratory infection)	Yes	Multiple daily	GS	No (Rivotril when needed)	Yes	No	No	Severe to profound ID
Kuchenbuch et al. 2019 (patient 11)	c.1420C>T / p.Arg474Cys / De novo	EIMFS	F	3,8	NA	NA	NA	NA	Severe NDD	6	No	Yes	Multiple daily	FM (tonic)	Yes (LTG, Rivotril when needed)	Yes	No	No	Severe to profound ID
Kuchenbuch et al. 2019 (patient 14)	c.1421G>A / p.Arg474His / De novo	EIMFS	M	2,1	0,03	NA	NA	NA	Severe NDD	9,4	No	Yes	Multiple daily	FM	Yes (CBD)	No	No	No	Severe to profound ID
Kuchenbuch et al. 2019 (patient 16)	c.2800G>A / p.Ala934Thr / De novo	EIMFS	M	0,9	NA	NA	NA	Early MRI: thinCC	Severe NDD	2,75	No	Yes	Multiple daily	FS	Yes (CBD)	No	No	No	Severe NDD
Kuchenbuch et al. 2019 (patient 17), Merdariu et al. 2012[14]	c.2688G>A / p.Met896Ile / De novo	EIMFS	F	8	2	Weekly	FIA, FC, auto	6 mo: mild enlargement of pericerebral spaces	Mild ID	15,25	No	Yes	Multiple weekly	Noctural, FS	Yes (LEV, Diacomit, Rivotril, Epidyolex)	No	No	No	ID
Numis et al. 2018[15] (patient 2), Poisson et al. 2020[16] (patient 2), Cornet et al. 2021[17] (patient 15)	c.776C>A / p.Ala259Asp / De novo	EIMFS	M	NA	0,03	Multiple daily	FM, Au	5 mo: severe diffuse thin CC, mild WMA ; MRS: normal	Profound ID	12	No	Yes	Multiple daily	Fmigrating, FIA, Auto	Yes (Cenobamate, Fycompa, RUF, GPT)	No	No	No	Severe to profound ID
Ohba et al. 2015 (patient 3)	c.1225C>T / p.Pro409Ser / De novo	EIMFS	F	5,3	2	NA	FS	2 mo: Mildly thin CC and CA ; 4 mo: subdural hematoma	Severe NDD	16	No	Yes	Multiple daily	NA	Yes (PHE, VPA, LEV, TPM, PER)	No	No	No	Severe to profound ID
Ohba et al. 2015 (patient 4)	c.862G>A / p.Gly288Ser / De novo	EIMFS	M	1,83	4	NA	FM	4mo: subdural hygroma ; 7 mo: diffuse CA	Severe NDD	1	Yes (1 ; respiratory infection)	Yes	Multiple daily	TS	Yes (LEV, KBr, PB, VPA)	Yes	Yes (1 ; VNS, no effect)	No	Severe NDD
Ohba et al. 2015 (patient 5)	c.1421G>A / p.Arg474His / De novo	EIMFS	M	2,25	1	NA	FM	2 mo: normal ; 1 yo: CA	Severe NDD	13	No	Yes	Multiple daily	TS	Yes (PB, LCM, BRV, RUF, KBr, PER, FFA)	No	No	Yes (8 yo: diffuse CA)	Severe to profound ID
Ohba et al. 2015 (patient 6)	c.2800G>A / p.Ala934Thr / De novo	EIMFS	M	6,16	0,13	NA	FM	2 mo: normal ; 2 yo: normal	Profound ID	17	No	Yes	Multiple weekly	NA	Yes (LEV, PHE, LTG)	No	No	No	Severe to profound ID
Ohba et al. 2015 (patient 9)	c.2771C>T / p.Pro924Leu / Inherited from	EIMFS	F	1,16	1,5	NA	InM	2 mo: normal	Severe NDD	12	No	Yes	Multiple yearly	FS	Yes (NZP, acetazolamide, NaBr)	No	No	No	Severe to profound ID

	the mother who carried a somatic mosaic mutation																	
Trivisano et al. 2025[18] (patient 1)	c.1193G>A / p.Arg398Gln / De novo	EIMFS	M	2,5	NA	Multiple daily	InM	NA	Severe NDD	3	No	Yes	Multiple daily	GS	Yes (TPM, CBD, CLB)	No	No	No	Moderate NDD
Trivisano et al. 2025 (patient 3)	c.1066C>T / p.Arg356Trp / De novo	EIMFS	F	15	NA	Multiple daily	NA	NA	Severe ID	15	No	Yes	Multiple daily	GS	Yes (PHE, LRZ, CLB, RUF, LCS)	No	No	No	Severe to profound ID
Yoshitomi et al. 2019[19] (patient 1)	c.1283G>A / p.Arg428Gln / De novo	EIMFS	M	1,7	1	Multiple daily	FM	NA	Severe NDD	9	No	Yes	Multiple daily	FS (tonic)	Yes (LCM, KBr, QUIN)	Yes	No	Yes (Abnormal. Non-specific diffuse CA, moderate. Delayed myelination.)	Severe to profound ID
Yoshitomi et al. 2019 (patient 2)	c.2800G>A / p.Ala934Thr / De novo	EIMFS	F	NA	2	Multiple daily	FM	NA	Severe NDD	10	No	Yes	Multiple daily	FS (tonic)	No (QUIN)	No	No	Yes (Abnormal. Non-specific diffuse CA, moderate. Delayed myelination.)	Severe to profound ID
Yoshitomi et al. 2019 (patient 3)	c.862G>A / p.Gly288Ser / De novo	EIMFS	M	NA	2	Multiple daily	FM, Au	NA	Severe NDD	4	No	Yes	Multiple daily	FS (tonic)	No (QUIN)	No	Yes (3,75 ; total callosotomy)	Yes (Abnormal. Non-specific diffuse CA,mild. Delayed myelination.)	Severe to profound ID
Bonardi et al. 2021(40)	c.2686A>G / p.Met896Val / De novo	DEE	F	24	6	Yearly	Spasms, Myo	7 mo: CA	Severe ID	31	No	Yes	Variable: monthly to daily	FS, GS	Yes (VPA, LTG, CNZ, LEV)	NA	No	No	Severe to profound ID
Bonardi et al. 2021(48)	c.940A>G / p.Thr314Ala / De novo	DEE	F	31,5	15	Multiple daily	HMS, FIA, eTCS, TCS, TS, A-abs, SE, FLS	7 yo: normal ; 19 yo: normal ; 6y: PET: normal	Severe ID	37	No	Yes	Monthly	FIA, noctural HMS, GTC	Yes (VPA, PHE, STP, Thylloflazepate)	No	No (in 2018 VNS implantation)	No	Severe to profound ID
Chong et al. 2016[20], Ohba et al. 2015 (patient 11)	c.1283G>A / p.Arg428Gln / De novo	DEE	M	6	1,5	Multiple daily	NA	1 mo: normal ; 2,5 yo: hypoplastic CC, delayed myelination, left lateral ventricular dilatation	Profound ID	15	No	Yes	Multiple daily	FS, GS	Yes (LEV, LTG, VPA)	No	No (VNS at age 2)	No	Severe to profound ID
Datta et al. 2019[21] (patient 2)	c.1420C>G / p.Arg474Gly / De novo	DEE	M	NA	0,75	Multiple daily	FM	2,5 yo: normal	NDD	9,5	No	Yes	Multiple weekly	FM, FIA	Yes (VPA, PHB, LAC, LEV)	No	No	Yes (Asymmetric prominece of sulcal spaces L hemisphere, susupicious for left frontal, temporal and Parieto occipital cortical dysplasia.)	Moderate ID
Filareto et al. 2025[22] (case 6)	c.785G>A / p.Arg262Gln / De novo	DEE	M	NA	36	NA	NA	NA	Severe NDD	8	No	Yes	Multiple daily	FS, myoclonic seizure and focal with fall	Yes (oxCBZ, VPA, Cenobamate, CNZ)	NA	NA	NA	Severe to profound ID
Fukuoka et al. 2017[23]	c.1955G>T / p.Gly652Val / De novo	DEE	M	2,6	5	Multiple daily	Spasms	5 mo: normal	NDD	12	No	Yes	Monthly	GS	Yes (QUIN, VPA)	No	Yes (4 ; total callosotomy)	Yes (Disconnected CC)	Severe to profound ID
Ikeda et al. 2021[6] (patient 1), Ohba et al. 2015 (patient 1)	c.1420C>T / p.Arg474Cys / De novo	DEE	M	12	0	NA	FS	2 mo: thin CC ; 10 mo: delayed myelination and CA	Severe ID	15	Yes (15 ; severe pulmonary hemorrhage)	Yes	Multiple daily	FM (tonic)	Yes (PB, LCM, KBr)	No	No	No	Severe to profound ID

Krygier et al. 2024[24] (72 F)	c.1193G>A / p.Arg398Gln / NA	DEE	F	5	6	NA	FM, FIA	Normal (age NA)	NDD	7	No	No			Yes (PRIMIDONUM)	Yes	No	No	Moderate ID
Krygier et al. 2024 (87M)	c.862G>A / p.Gly288Ser / NA	DEE	M	5	1	NA	FM	Normal	NDD	7	No	Yes	Weekly	FS, GS	Yes (PRIMIDONUM)	Yes	No	No	Severe to profound ID
Shchubelka et al. 2024[25] (16)	c.1309C>T / p.Leu437Phe / NA	DEE	NA	NA	NA	NA	NA	NA	NA	3	No	Yes	Multiple monthly	FS, GS	Yes (VPA)	No	No	No	Moderate NDD
Shchubelka et al. 2024 (17)	c.784C>T / p.Arg262Trp / NA	DEE	NA	NA	NA	NA	NA	NA	NA	10	No	Yes	Multiple yearly	GS	Yes (VPA, LTG, CBD)	No	No	No	Severe to profound ID
Tsang et al. 2018[26] (patient 7)	c.1038C>A (VUS) / p.Phe346Leu / inherited (paternal mosaicism)	DEE	M	0,75	3	NA	NA	< 9 mo: CA and delayed myelination	NDD	9	No	Yes	Monthly	FS, GS	Yes (CBZ, LEV)	No	No	No	Severe to profound ID
Vanderver et al. 2014[27]	c.2794T>A / p.Phe932Ile / De novo (variant LoF et pas GoF)	DEE	M	10	1	NA	Gmyo	1 mo: severe delayed myelination ; 7 yo: progression of myelination but at levels that are still severely reduced.	Severe ID	19	No	Yes	Weekly	FS	Yes (TPM, LTG)	No	No	No	Severe to profound ID
Yamamoto et al. 2023[28] - patient 2	c.1421G>A / p.Arg474His / De novo	DEE	F	4	1	Daily	FM, Spasms	NA	Severe NDD	5,9	Yes (7,9 ; uncertain (sudden death))	Yes	Multiple daily	FS, GS	Yes (LTG, clorazepate)	No	No	No	Severe NDD
Yoshitomi et al. 2019[19] (patient 4)	c.1420C>T / p.Arg474Cys / de novo	DEE	M	NA	1	Multiple daily	FM	NA	Severe NDD	15	Yes (15 ; severe pulmonary hemorrhage)	Yes	Multiple daily	FS (tonic)	Yes (PB, LCM, KBr)	No	No	Yes (Abnormal. Non-specific diffuse CA,mild. Delayed myelination.)	Severe to profound ID
Derry et al. 2008[29] (B II-5), Heron et al. 2012[30] (A), Mullen et al. 2017[31] (2)	c.2782C>T / p.Arg928Cys / inherited	SHE	M	54	180	Multiple nighlty	FLS, auto, GTC	Normal (age NA)	Mild ID	60	NA	Yes	Multiple yearly	Sleep-related and awake hypermotor seizures with choking, gasping for breath, dystonia, oral automatisms	Yes (VPA, TPM, CNZ)	NA	NA	NA	Borderline ID
Derry et al. 2008 (B III-2), Heron et al. 2012 (A), Mullen et al. 2017 (5)	c.2782C>T / p.Arg928Cys / inherited	SHE	F	21	36	Multiple nighlty	FLS, auto	Normal (age NA)	Mild ID	39	NA	Yes	1/2-3 night	Sleep-related hypermotor seizures; GTC	Yes (CNZ, CBZ, TPM, VPA, PHE)	NA	NA	NA	Moderate ID
Derry et al. 2008 (B III-6), Heron et al. 2012 (A), Mullen et al. 2017 (3)	c.2782C>T / p.Arg928Cys / inherited	SHE	F	6	33	Multiple nighlty	FLS, auto	33 mo: normal	Mild to moderate ID	26	NA	Yes	Multiple weekly	Hypermotor seizures; tonic seizures; GTC	Yes (CNZ, CBZ, TPM, VPA, PHE)	NA	NA	NA	Severe to profound ID
Derry et al. 2008 (B III-7), Heron et al. 2012 (A), Mullen et al. 2017 (4)	c.2782C>T / p.Arg928Cys / inherited	SHE	M	5	12	Multiple nighlty	FLS, auto	NA	Normal	25	NA	Yes	Weekly	Sleep-related and awake hypermotor seizures; FTB(TC)	Yes (TPM, CBZ, CNZ, LEV)	NA	NA	NA	Mild ID
Derry et al. 2008 (family B II-4), Heron et al. 2012 (family A),	c.2782C>T / p.Arg928Cys / inherited	SHE	M	43	24	Multiple nighlty	FLS, auto	Normal (age NA)	Mild ID	49	NA	Yes	Nightly	Sleep-related hypermotor seizures	Yes (VPA, TPM, CBZ)	NA	NA	NA	Mild ID

Mullen et al. 2017 (1)																		
Héron et al. 2012 (case D)	c.2688G>A / p.Met896Ile / De novo	SHE	M	NA	108	NA	NA	NA	Normal	49	NA	Yes	Monthly	Sleep-related and awake focal impaired awareness seizures; tonic-clonic seizures reported once	Yes (VPA, LTG, TPM, CNZ)	NA	NA	NA	Borderline ID (NPA)
Hildebrand et al. 2016[32] (9092), Mullen et al. 2017 (6)	c.2849G>A / p.Arg950Gln / De novo	SHE	M	28	36	NA	Focal dyscognitive (13y), Aura (3y)	Normal (age NA)	Mild ID	38	NA	Yes	Multiple daily	Hypermotor seizures with and without impaired awareness; FTB(TC); focal motor status epilepticus	Yes (CBZ, CNZ, Midazolam)	NA	NA	NA	Mild ID
Licchetta et al. 2020[33]	c.2800G>A / p.Ala934Thr / De novo	SHE	F	NA	108	NA	NA	NA	ID	29	NA	No			Yes (CBZ, LCS)	NA	NA	NA	Mild ID
Cataldi et al. 2019[34] (cas index)	c.2896G>A / p.Ala966Thr / inherited	SHE + EIMFS	M	NA	24	NA	FLS (noctural), migrating focal (diurnal)	24 mo: multiple very small cerebral cavernomas with no sign of previous bleedings.	Mild ID	NA	NA	Yes	NA	NA	NA	NA	NA	NA	Mild ID
Heron et al. 2012 (patient III2-familyB)	c.2386T>C / p.Tyr796His / inherited	SHE/ FCD Ib	M	35	36	NA	FLS, nocturnal wandering	Focal cortical dysplasia Ib	Learning difficulties	31	NA	Yes	Yearly	seizures during wakefulness with staring	Yes (CBZ, VPA, PHE, DZP)	NA	NA	NA	ID
Cherian et al. 2021 (patien t 1) II-5	c.2882G>A / p.Arg961His / NA	Focal epilepsy	F	64	168	NA	TLS (staring, feeling of terror, stightness).	NA	Normal	64	NA	Yes	Monthly	FNM, GS once	No	NA	NA	NA	Normal
Cherian et al. 2021 (patien t 2) III-11	c.2882G>A / p.Arg961His / inherited	Focal epilepsy	F	40	120	Multiple daily (FNM + Au) - Multiple yearly (FGTC)	FNM + Au (presumed insular/opercular onset), FGTC	19 yo: normal ; 24 yo: normal ; 37 yo: normal	Normal	41	NA	Yes	Monthly	FNM; FBTCS; Questionable auditory aura (from wakefulness)	Yes (CBZ)	NA	NA	NA	Normal
Cherian et al. 2021 (patien t 4) III-14	c.2882G>A / p.Arg961His / inherited	Focal epilepsy	F	30	216	Multiple nighlty	FNM + Au (presumed insular/opercular onset)	24 yo: normal	Cognitive variability	31	NA	Yes	Monthly	FNM; FBTCS	Yes (PHE, BRV, TPM)	NA	NA	NA	Normal
Mosca et al. 2024[35], Filareto et al. 2025[22] (case 5)	c.2809A>G / p.Ser937Gly / De novo	Focal epilepsy	F	20	36	None	TLS (staring, feeling of terror, swallowing, abdomen tightness).	3 yo: normal ; 18 yo: normal	Normal	21	fNo	Yes	Yearly	FS	Yes (oxCBZ, CLZ)	NA	NA	No	Mild ID

Supplementary Table 3: Individual neurological features of patients with clinical updates.

Abbreviations :A-Abs: Atypical Absences, ASM: AntiSeizure Medications, Au: Autonomic, BRV: Brivacetam, CA: Cerebral Atrophy, CBD: Cannabidiol, CBZ: Carbamazepine, CC: Corpus Callosum, CLB: Clobazam, CLZ: Clorazepam, CNZ: Clonazepam, CT-Scan: Computed Tomography scan, DEE: Developmental Epileptic Encephalopathy, DOI: day of life, DZP: Diazepam, EIMFS: Epilepsy of Infancy with Migrating Focal Seizure, eTCS: evolving to Tonic Clonic Seizure, FBM: Felbamate, FIA: Focal Impaired Awarenesse, FLS: frontal lobe seizures, F: Focal, FTB(TC): Focal To Bilateral (Tonic-Clonic), FNM: Focal Non Motor, FNMIA: Focal Non Motor Impaired Awareness, FM: Focal Motor, FS: Focal Seizures, FU: Follow-Up, GMyo: Generalized Myoclonia, GS: Generalized

Seizures, GPT: Gabapentine,HemiC: hemiclonic, ID: Intellectual Disability, KBr: Potassium Bromide, KD: Ketogenic Diet, LAC: Lacosamide, LEV: Levetiracetam, LFU: Last Follow-Up, LCM: Lacosamide, LRZ: Lorazepate, LTG: Lamotrigine, MFS: Migrating Focal Seizure, M/F: Male/Female, mo: month old, MRI: Magnetic Resonance Imaging, MRS: Magnetic Resonance Spectroscopy, Myo: myoclonus, NA: Non Available, NaBr: Sodium Bromide; NDD: Neurodevlopmental Delay, NZP: Nitrazepam, oxCBZ: Oxcarbazepinde, PER: Perampanel, PHB: Phenobarbital,PHE: Phenytoine, QUIN: quinidine, RUF: Rufinamide, SE: Statuts Epilepticus, SHE: Sleep-related Hypermotor Epilepsy, STP: Stiripentol, Sz: Seizure, TCS: Tonic Clonic Seizure, TLS: Temporal Lobe Seizure, TPM: Topiramate, TS: Tonic Seizures, VGV: Vigabatrin, VNS: Vagus Nerve Stimulation, VPA: Valproate, WMA: White Matter Abnormalities, yo: year old

Patient's reference	Variant c. / p. / inheritance	Epilepsy syndrome	Sex	Age at last visit (y)	Death since the publication (age (y) ; cause)	Feeding disorders	Vascular malformations (detail / revelation mode)	Precocious puberty ?	Orthopedic features	Behavioral disorders	Autism spectrum disorder (diagnosis scale)	Other psychiatric disorder associated	Psychiatric therapies	Sleep disorders
Bonardi et al. 2021(10)	c.862G>A / p.Gly288Ser / De novo	EIMFS	M	7	No	Yes (GT)	NA	No	No	No	No	NA	No	No
Bonardi et al. 2021(11)	c.862G>A / p.Gly288Ser / NA	EIMFS	M	6	No	Yes (GT)	NA	No	Yes (Hip spasticity)	No	No	NA	No	No
Bonardi et al. 2021(20°), Fitzgerald et al. 2019 (16)	c.1429G>A / p.Ala477Thr / De novo	EIMFS	F	18	Yes (18 ; respiratory infection)	Yes (GT)	NA	No	Yes (Spastic quad, scoliosis, osteopenia, neuromuscular hip dysplasia / Right hip femoral head and neck resection with valgus osteotomy, spinal fusion)	No	No (too impaired to assess)	NA	NA	No
Bonardi et al. 2021(31)	c.2849G>A / p.Arg950Gln / De novo	EIMFS	M	24	No	Yes (eats blended food)	No	No	No	Yes	No	NA	No	No
Bonardi et al. 2021(33°), Fitzgerald et al. 2019 (6)	c.2881C>A / p.Arg961Ser / De novo	EIMFS	M	11	No	No	NA	No	Yes (Spastic quad, neuromuscular hip dysplasia / Bilateral hip surgery)	No	No (too impaired to assess)	NA		No
Cherian et al. 2021 (patient 5) IV-5	c.2882G>A / p.Arg961His / inherited	EIMFS	F	11	No	Yes (GT)	No	Yes	Yes (Scoliosis, Superolaterally subluxed right femoral head that reduces in frog-leg view, generalized osteopenia)	No	No (too delayed to say)	NA	No	Yes (insomnia but likely due to frequent seizures)
Dilena et al. 2018 (patient 1)	c.2849G>A / p.Arg950Gln / De novo	EIMFS	M	7,5	No	Yes (GT)	No	No	No	No	No	NA		Yes (Melatonin)
Dilena et al. 2018 (patient 2)	c.2677G>A / p.Glu893Lys / De novo	EIMFS	M	6	Yes (6 ; severe pulmonary hemorrhage)	Yes (GT, GERD, dysphagia)	Yes (Ectasia of the bronchial arteries and the main pulmonary artery / Symptomatic (recurrent respiratory infections))	Too young	Yes (Bilateral cervico-diaphyseal valgus with incomplete coverage of the femoral head (>right))	No	No	NA	No	Yes (Melatonin)

Ferretti et al. 2022 (patient 1)	c.337G>A / p.Val113Met / De novo	EIMFS	M	7	Yes (7 ; respiratory infection)	Yes (GT)	No	No	Yes (Mild kyphoscoliosis, bilateral valgus-pronated foot)	No	No	NA	No	No
Ferretti et al. 2022 (patient 3)	c.862G>A / p.Gly288Ser / De novo	EIMFS	M	5	No	Yes (delay in feeding and swallowing skills)	No	Too young	No	No	No	NA	No	No
Ikeda et al. 2021 (patient 2), Ohba et al. 2015 (patient 2)	c.808C>G / p.Gln270Glu / De novo	EIMFS	F	14	No	Yes (GT)	Yes (SPCA / Symptomatic (pulmonary hemorrhage))	No	Yes (Hip dislocation, limb joint contracture)	No	No	NA	No	No
Kawasaki et al. 2017 (patient 3), Ohba et al. 2015 (patient 7)	c.1421G>A / p.Arg474Cys / De novo	EIMFS	M	6,5	No	Yes (GT)	Yes (Systemic to pulmonary collateral arteries / Incidental finding)	No	No	NA	No	NA		Yes
Kohli et al. 2020	c.1420C>T / p. Arg474Cys / NA	EIMFS	F	5	No	Yes (GT, GERD, dysphagia, feeding intolerance)	Yes (SPCA)	Too young	Yes (Scoliosis, hips spasticity, feet inversion)	No	No	NA	No	Yes
Kravetz et al. 2021	c.2795T>C / p.Phe932Ser / De novo	EIMFS	F	15	No	NA	NA	NA	NA	No	No	NA	No	NA
Kuchenbuch et al. 2019 (patient 1), Barcia et al. 2012 (patient 2), Kim et al. 2014 (1) - CONSOLIDATION	c.1283G>A / p.Arg428Gln / De novo	EIMFS	M	23	No	Yes (eats blended food)	NA	No	Yes (Hip dislocation (2), Scoliosis / Sinal arthrodesis (scoliosis))	No	NA	NA	No	NA
Kuchenbuch et al. 2019 (patient 10) - CONSOLIDATION	c.1283G>A / p.Arg428Gln / De novo	EIMFS	F	12,4	Yes (13,4 ; respiratory infection)	Yes (GT, constipation)	NA	No	Yes (Hip dislocation, Severe scoliosis / Spinal arthrodesis, hip dislocation (2))	No	NA	NA	No	Yes (Melatonin)
Kuchenbuch et al. 2019 (patient 11) - STORMY	c.1420C>T / p.Arg474Cys / De novo	EIMFS	F	6	No	Yes (GT)	NA	Too young	Yes (Hip dislocation (1), Scoliosis)	No	NA	NA	No	No
Kuchenbuch et al. 2019 (patient 14) - STORMY	c.1421G>A / p.Arg474His / De novo	EIMFS	M	9,4	No	Yes (constipation)	NA	Too young	Yes (Hip dislocation (1), Scoliosis)	No	NA	NA	No	No
Kuchenbuch et al. 2019 (patient 16) - STORMY	c.2800G>A / p.Ala934Thr / De novo	EIMFS	M	2,75	No	Yes (painful dyspepsia)	NA	Too young	Yes (Hip dislocation without pain (1))	No	No	NA	No	No
Kuchenbuch et al. 2019 (patient 17), Merdariu et al. 2012 - CONSOLIDATION after Sz free period	c.2688G>A / p.Met896Ile / De novo	EIMFS	F	15,25	No	NA	NA	No	NA	No	NA	NA	No	No
Numis et al. 2018 (patient 2), Poisson et al. 2020 (patient	c.776C>A / p.Ala259Asp / De novo	EIMFS	M	12	No	Yes (GT, GERD, constipation)	No	No	Yes (Hip dysplasia (2), scoliosis, spasticity with contractures and clonus, mild bilateral	Yes	No (too impaired to assess)	Agitation started 10/2023, managed with	Yes (Gabapentin, clonidine)	Yes (no regular cycling)

2), Cornet et al. 2021 (patient 15)									equinus / Hip abductor surgery 4/2018)			gabapentin and clonidine		
Ohba et al. 2015 (patient 3)	c.1225C>T / p.Pro409Ser / De novo	EIMFS	F	16	No	Yes (GT)	NA	No	Yes (Right femoral fracture, Scoliosis)	No	No	NA	No	Yes
Ohba et al. 2015 (patient 4)	c.862G>A / p.Gly288Ser / De novo	EIMFS	M	1	Yes (1 ; respiratory infection)	Yes (GT)	No	Too young	No	No	No	NA	No	No
Ohba et al. 2015 (patient 5)	c.1421G>A / p.Arg474His / De novo	EIMFS	M	13	No	Yes (GT)	Yes (Aorto-pulmonary collateral artery / Symptomatic)	No	No	No	No	NA		Yes (Melatonin)
Ohba et al. 2015 (patient 6)	c.2800G>A / p.Ala934Thr / De novo	EIMFS	M	17	No	Yes (GT)	NA	No	Yes (Hip dislocation (1), Right supracondylar femoral fracture, Scoliosis)	No	No	NA	No	No
Ohba et al. 2015 (patient 9)	c.2771C>T / p.Pro924Leu / Inherited from the mother who carried a somatic mosaic mutation	EIMFS	F	12	No	Yes (eats blended food)	NA	No	Yes (Hip dislocation (1), scoliosis / Left hip arthroplast was performed at age 9)	No	No	NA	No	Yes (Melatonin)
Trivisano et al. 2025 (patient 1)	c.1193G>A / p.Arg398Gln / De novo	EIMFS	M	3	No	Yes (GT)	No	Too young	No	No	No	NA	No	Yes (Melatonin)
Trivisano et al. 2025 (patient 3)	c.1066C>T / p.Arg356Trp / De novo	EIMFS	F	15	No	Yes (GT)	Yes (Angiodysplasia of the bronchial arteries and anomalous pulmonary venous return, abdominal vascular anomalies (celiac tripod ectasia, superior mesenteric arteryectasia, splenic arteryectasia), treated with embolizations / Symptomatic (hemoptysis/hemoptysis episodes))	No	Yes (Right hip dislocation, feet in talipes valgus, knees in valgus, scoliosis / Tenotomy of the adductors and psoas bilaterally; stretching of the medial flexors bilaterally	No	No	NA	No	Yes (lorazepam)
Yoshitomi et al. 2019 (patient 1)	c.1283G>A / p.Arg428Gln / De novo	EIMFS	M	9	No	Yes (GT, vomiting)	No	No	Yes (Contracture of bil ankle joints. Mild scoliosis. Acetabular dysplasia.)	No	No (too serverly delayed to diagnose ASD)	NA	No	No
Yoshitomi et al. 2019 (patient 2)	c.2800G>A / p.Ala934Thr / De novo	EIMFS	F	10	No	Yes (GT, vomiting, GERD)	No	No	No	No	No (too serverly delayed to diagnose ASD)	NA	No	No
Yoshitomi et al. 2019 (patient 3)	c.862G>A / p.Gly288Ser / De novo	EIMFS	M	4	No	Yes (NA)	NA	No	No	No	No (too serverly delayed to diagnose ASD)	NA	No	No
Bonardi et al. 2021(40)	c.2686A>G / p.Met896Val / De novo	DEE	F	31	No	Yes (eats blended food)	NA	No	Yes (Feet deformity / hip spasticity)	Yes	NA	NA	Yes (Risperidone)	Yes

Bonardi et al. 2021(48)	c.940A>G / p.Thr314Ala / De novo	DEE	F	37	No	Yes (mild dysphagia, eats blended food)	No	No	Yes (Scoliosis)	No	Not diagnosed	NA		No
Chong et al. 2016, Ohba et al. 2015 (patient 11)	c.1283G>A / p.Arg428Gln / De novo	DEE	M	15	No	Yes (GT)	NA	No	Yes (Hip dislocation, Scoliosis)	No	No	NA	No	No
Datta et al. 2019 (patient 2)	c.1420C>G / p.Arg474Gly / De novo	DEE	M	9,5	No	No	No	No	Yes (Right foot everted when walking)	NA	NA	NA	NA	No
Filareto et al. 2025 (case 6)	c.785G>A / p.Arg262Gln / De novo	DEE	M	8	No	NA	NA	NA	NA	Yes	No	Severe behaviour disorder characterized by hyperactivity, irritability and several stereotypes	Yes (Xenazina at low dosage for oral dyskinesias)	NA
Fukuoka et al. 2017	c.1955G>T / p.Gly652Val / De novo	DEE	M	12	No	Yes (GT)	No	Too young	Yes (Scoliosis)	No	No	NA	No	Yes
Ikeda et al. 2021 (patient 1), Ohba et al. 2015 (patient 1)	c.1420C>T / p.Arg474Cys / De novo	DEE	M	15	Yes (15 ; severe pulmonary hemorrhage)	Yes (GT)	Yes (SPCA / Symptomatic (pulmonary hemorrhage))	No	Yes (Scoliosis)	No	No	NA	No	No
Krygier et al. 2024 (72 F)	c.1193G>A / p.Arg398Gln / NA	DEE	F	7	No	No	No	No	No	Yes	No	NA	No	No
Krygier et al. 2024 (87M)	c.862G>A / p.Gly288Ser / NA	DEE	M	7	No	No	No	No	No	Yes	No	NA	No	No
Shchubelka et al. 2024 (16)	c.1309C>T / p.Leu437Phe / NA	DEE	NA	3	No	Yes (GT, food selectivity)	NA	Too young	Yes (Scoliosis, connective tissue disorder clinically)	Yes	Yes (ADOS)	NA	Yes (Risperidone)	Yes (Melatonin)
Shchubelka et al. 2024 (17)	c.784C>T / p.Arg262Trp / NA	DEE	NA	10	No	NA	NA	Too young	NA	Intellectual disability	No	NA	No	No
Tsang et al. 2018 (patient 7)	c.1038C>A (VUS) / p.Phe346Leu / inherited (paternal mosaicism)	DEE	M	9	No	Yes (GT, dysphagia)	No	No	Yes (Hip dislocation (2), Scoliosis)	No	No	NA	No	No
Vanderver et al. 2014	c.2794T>A / p.Phe932Ile / De novo (variant LoF et pas GoF)	DEE	M	19	No	Yes (GT, constipation)	No	No	Yes (Neuromuscular scoliosis (right thoracolumbar), bilateral knee clunking or crepitus, right ACL abnormality, osteopenia)	No	No	NA	No	Yes (Melatonin)
Yamamoto et al. 2023 - patient 2	c.1421G>A / p.Arg474His / De novo	DEE	F	5,9	Yes (7,9 ; uncertain (sudden death))	Yes (eats blended food)	Yes (SPCA / Screening)	No	No	No	No	NA	No	No
Yoshitomi et al. 2019 (patient 4)	c.1420C>T / p.Arg474Cys / de novo	DEE	M	15	Yes (15 ; severe pulmonary hemorrhage)	Yes (GT)	Yes (SPCA / Symptomatic (nasal bleeding, hemoptysis))	No	Yes (Acetabular dysplasia. Severe scoliosis)	No	No (too serverly delayed to diagnose ASD)	NA	No	No

Derry et al. 2008 (B II-5), Heron et al. 2012 (A), Mullen et al. 2017 (2)	c.2782C>T / p.Arg928Cys / inherited	SHE	M	60	NA	NA	NA	NA	NA	NA	No	Mood disorder	Yes (Doxepin)	NA
Derry et al. 2008 (B III-2), Heron et al. 2012 (A), Mullen et al. 2017 (5)	c.2782C>T / p.Arg928Cys / inherited	SHE	F	39	NA	NA	NA	NA	NA	Yes	No	Rapid cyclic unipolar depression	Yes (Duloxetine)	NA
Derry et al. 2008 (B III-6), Heron et al. 2012 (A), Mullen et al. 2017 (3)	c.2782C>T / p.Arg928Cys / inherited	SHE	F	26	NA	NA	NA	NA	NA	NA	No	None	No	NA
Derry et al. 2008 (B III-7), Heron et al. 2012 (A), Mullen et al. 2017 (4)	c.2782C>T / p.Arg928Cys / inherited	SHE	M	25	NA	NA	NA	NA	NA	No	No	None	No	NA
Derry et al. 2008 (family B II-4), Heron et al. 2012 (family A), Mullen et al. 2017 (1)	c.2782C>T / p.Arg928Cys / inherited	SHE	M	49	NA	NA	NA	NA	NA	NA	No	Anxiety	No	NA
Héron et al. 2012 (case D)	c.2688G>A / p.Met896Ile / De novo	SHE	M	49	Yes (49 : glioblastoma)	NA	NA	NA	NA	NA	No	NA	Yes (Paroxetine, olanzapine, valium)	NA
Hildebran d et al. 2016 (9092), Mullen et al. 2017 (6)	c.2849G>A / p.Arg950Gln / De novo	SHE	M	38	NA	NA	NA	NA	NA	NA	Yes (Level 2 ASD using the ADOS at an autism clinic (30 y))	Recurrent psychosis (last at 38 y), anxiety, obsessive compulsive disorder	Yes (Quetiapine, diazepam)	NA
Licchetta et al. 2020	c.2800G>A / p.Ala934Thr / De novo	SHE	F	29	NA	NA	NA	NA	NA	NA	No	None	No	No
Cataldi et al. 2019 (cas index)	c.2896G>A / p.Ala966Thr / inherited	SHE + EIMFS	M	NA	NA	NA	NA	NA	NA	NA	No	None	No	NA
Heron et al. 2012 (patient III2-familyB)	c.2386T>C / p.Tyr796His / inherited	SHE/ FCD Ib	M	31	NA	NA	NA	NA	NA	NA	No	Schizophrenia, depression	Yes (Quetiapine, Clotiapine, Carbolithium, Diazepam)	NA
Cherian et al. 2021 (patien t 1) II-5	c.2882G>A / p.Arg961His / NA	Focal epilepsy	F	64	NA	NA	NA	NA	NA	NA	No	None	No	NA
Cherian et al. 2021 (patien t 2) III-11	c.2882G>A / p.Arg961His / inherited	Focal epilepsy	F	41	NA	NA	NA	NA	NA	NA	No	None	No	NA
Cherian et al. 2021 (patien t 4) III-14	c.2882G>A / p.Arg961His / inherited	Focal epilepsy	F	31	NA	NA	NA	NA	NA	NA	No	None	No	NA
Mosca et al. 2024, Filareto et al. 2025 (case 5)	c.2809A>G / p.Ser937Gly / De novo	Focal epilepsy	F	21	No	NA	NA	NA	NA	Yes	No	Depression and anxiety, improved after	Yes (Fluoxetine)	NA

	treatment with Fluoxetine

Supplementary Table 4 : Individual extra-neurological features of patients with clinical updates.

ACL: Anterior Cruciate Ligament, ADOS: Autism Diagnostic Observation Schedule, ASD: Autism Spectrum Disorder, DEE: Developmental and Epileptic Encephalopathy, EIMFS: Epilepsy of Infancy with Migrating Focal Seizures, FCD: Focal Cortical Dysplasia, GERD: Gastro-oesophageal reflux, GT: Gastrostomy, M/F: Male/Female, NA: Non Available, SHE: Sleep-related Hypermotor Epilepsy, SPCA: Systemic to pulmonary collateral arteries, y: yes

		EIMFS	DEE	SHE	Others	
Nter	p.Arg85Ser		1			
	p.Val113Met	1				
	p.Arg133His				1	
	p.Gly243Ser	1				
S5	p.His257Asp	1				GOF (Hinckley 2023[36])
S5	p.Ala259Asp	1				GOF (Numis 2018[15])
S5	p.Arg262Gln	1	1			GOF (Hinckley 2023)
S5	p.Arg262Trp		1			GOF (Hinckley 2023)
S5	**p.Met267Thr**	**3**				
S5	**p.Gln270Glu**	**3**				GOF (Hinckley 2023)
S5	p.Val271Phe	2				GOF (Hinckley 2023, McTague 2018[37])
pore	p.Leu274Ile	2				GOF (Hinckley 2023, McTague 2018)
pore	**p.Gly288Ser**	**24**	**7**	**2**	**2**	GOF (Hinckley, Rizzo)
S6	p.Thr314Ala		1			
S6	p.Ala338Gly	1				
S6	p.Val340Met				1	LOF (Hinkley)
	p.Phe346Leu	2	1			GOF (Hinckley 2023, McTague 2018)
RCK1	**p.Arg356Trp**	**4**				GOF (Trivisano 2025[18])
RCK1	p.Cys377Ser	1	1			
RCK1	**p.Arg398GIn**	**5 (6*)**	**5**	**6 (13*)**	**1**	GOF (Hinckley 2023, Milligan 2014[38], Trivisano 2025)
RCK1	p.Arg398Leu	1				
RCK1	p.Pro409Ser	1	1			GOF (Hinkley 2023, Barcia 2012[12])
RCK1	**p.Arg428GIn**	**21**	**1**			GOF (Hinkley 2023, Mikati 2015[39], Milligan 2014)
RCK1	p.Ser435Cys	1				
RCK1	p.Leu437Phe	1	2			
RCK1	p.Asn449Ser			1		
RCK1	**p.Arg474Cys**	**10**	**5**			LOF (Hinckley)
RCK1	p.Arg474Gly		1			

RCK1	**p.Arg474His**	**18**	**7**	**1**		GOF (Hinckley 2023, Trivisano 2025)
RCK1	p.Arg474Lys	1				
RCK1	p.Trp476Arg	2				
RCK1	p.Ala477Thr	2				GOF (Hinckley 2023)
RCK1	p.Asp480Asn	2				
RCK1	p.Phe502Val	1				GOF (Hinckley 2023, McTague 2018)
RCK1	**p.Met516Val**	**4**				GOF (Hinckley 2023, Rizzo 2016[40])
RCK1	p.Arg538Cys		1			
RCK1	p.Gln550del	1				GOF (Numis 2018[15])
	p.Lys629Asn	1				GOF (Hinckley 2023, Mikati 2015)
	p.Lys629Glu	2	1			GOF (Hinckley 2023)
	p.Gly652Val		1			LOF (Hinckley 2023)
	p.Asp718Val			1 (3*)		
	p.Ile760Met	**3**				GOF (Hinckley 2023)
	p.Ile760Phe	1				
	p.Leu781Val		1			
RCK2	p.Tyr796His			2 (5*)		LOF (Hinckley 2023)/ GOF (Milligan 2014, Mikati 2015)
NADBD	**p.Glu893Lys**	**3**				
NADBD	p.Glu893Val	1				
NADBD	p.Met896Ile	2	1	1		GOF (Milligan 2014)
NADBD	p.Met896Lys	1				GOF (Hinckley 2023, McTague 2018)
NADBD	p.Met896Val	1	1	1		
NADBD	p.Ala899Val	1				
NADBD	p.Arg905Gln		1			
NADBD	p.Gln906His		1			GOF (Hinckley 2023)
NADBD	p.Phe909Leu	1				
RCK2	p.Pro924Leu	1	1			GOF (Barcia 2012)
RCK2	**p.Arg928Cys**			**4 (13*)**	**2**	GOF (Hinckley 2023, Milligan 2014)
RCK2	p.Arg929Gln	1				

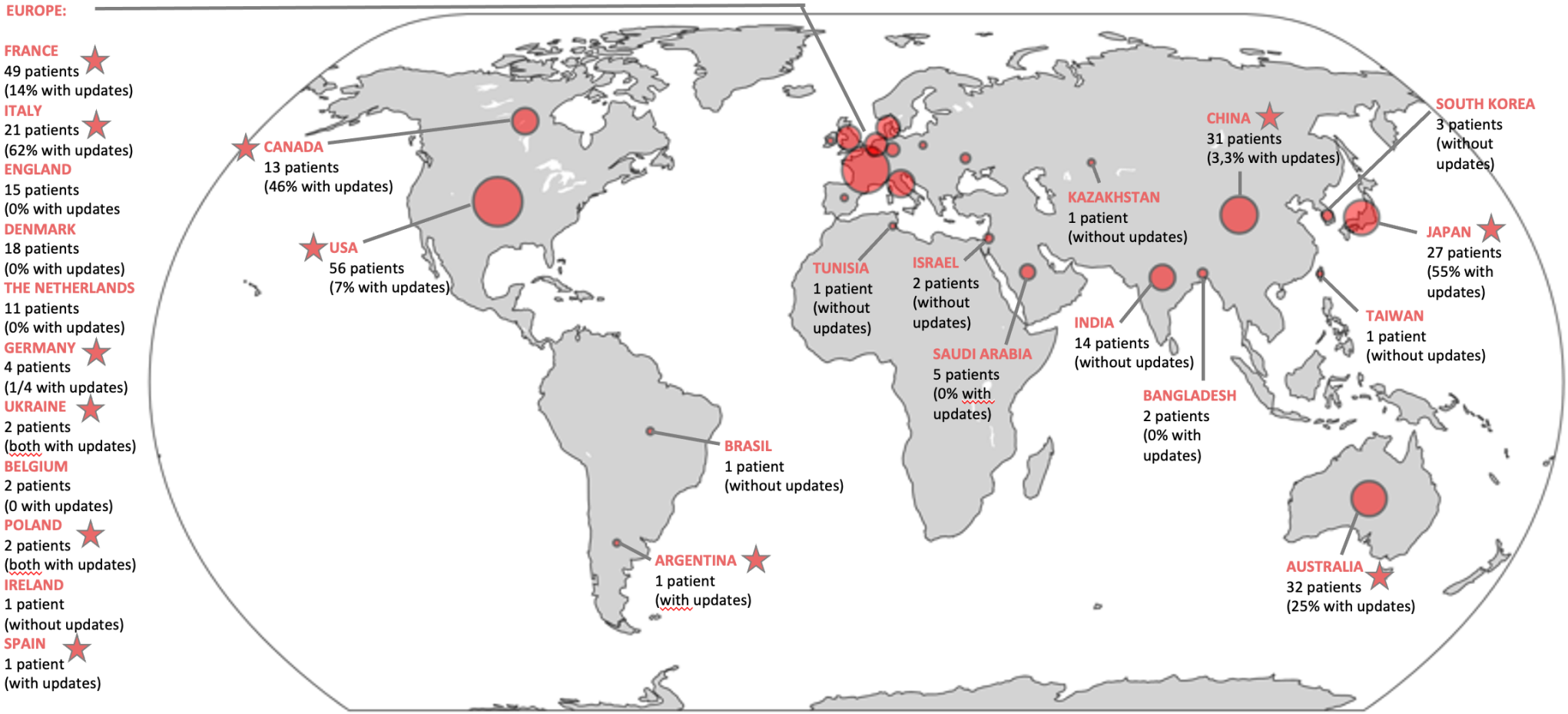
RCK2	p.Phe932Ile	0	1			LOF (Evely 2017[41])
RCK2	p.Phe932Leu	1		1		
RCK2	p.Phe932Ser	2				
RCK2	p.Arg933Cys			1 (2*)		
RCK2	p.Arg933Gly	1				
RCK2	p.Arg933His	1				
RCK2	**p.Ala934Thr**	**23**	**9**	**4**		GOF (McTague 2018, Milligan 2014)
RCK2	p.Ser937Gly				1	GOF (Mosca et al. 2024[35])
RCK2	p.Leu942Phe		0	1		
RCK2	p.Lys947Glu	1	1			
RCK2	**p.Arg950Gln**	**8**	**1**	**3 (6*)**	**2**	GOF (Hinckley 2023)
RCK2	**p.Arg961His**	**1**	**2**	**3**	**1 (3*)**	
RCK2	p.Arg961Ser	1				GOF (Hinckley 2023)
RCK2	p.Leu962Pro	1				
RCK2	p.Ala965Thr			1		
RCK2	p.Ala965Val	1				
RCK2	**p.Ala966Thr**			**3 (4*)**		GOF (Hinckley 2023)
RCK2	p.Ala966Thr (x2)		1			
RCK2	p.Ala989Ser	1				
Cter	p.Arg1106Gln				1	
Cter	p.Arg1106Pro	1				GOF (Hinckley 2023)
	Total général	180 (181*)	59 (59*)	36 (62*)	12 (14*)	

Supplementary Table 5: **Listing of reported KCNT1 variant and associated phenotypes**

One variant per family and phenotype in yellow

* In parentheses, the total number of patients reported, with each case counted, including familial cases.

Abbreviations: DEE: Developmental and Epileptic Encephalopathy, EIMFS: Epilepsy of Infancy with Migrating Focal Seizures, GOF: Gaon Of Function, LOF: Loss Of Function, SHE: Sleep-related Hypermotor Epilepsy



Supplementary Figure 1: Geographic distribution of published patients worldwide and response rate to the follow-up questionnaire

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