

Name of disease	Orphacode	Representative	Nationality	Website	Facebook Page	Email
1p36 deletion	ORPHA:1606	Asociacion de Afectados de Delecion 1p36	Spain	https://www.delecion1p36.es/	https://www.facebook.com/delecion1p36/	info@delecion1p36.es
8p inverted duplication/deletion syndrome	ORPHA:96092	La Maison 8p	France	https://laison8p.com	https://www.facebook.com/LaMaison8p	laison8p@gmail.com
Batten disease	ORPHA:79264	NCL - Gruppe Deutschland e. V.	Germany	https://www.ncl-deutschland.de/	https://www.facebook.com/NCLDeutschland?ref=ts&fref=ts	kontakt@ncl.info
Batten disease	ORPHA:79264	Batten disease family association	UK	http://www.bdfa-uk.org.uk/about-batten-disease/	https://www.facebook.com/bdfauk	admin@bdfa-uk.org.uk
Brain injury		Headway	UK	https://www.headway.org.uk/	https://www.facebook.com/headwayuk	enquiries@headway.org.uk
CASK-related disorders	ORPHA:163937, ORPHA:1934	Association Enfants CASK France	France	https://www.aecf-france.fr/	https://www.facebook.com/AssociationEnfantsCASKFrance	associationenfantscaskfrance@gmail.com
CASK-related disorders	ORPHA:163937, ORPHA:1934	CASK Research UK	UK	https://caskresearch.org/	https://web.facebook.com/Caskresearch	
CHD2 related epilepsy		Coalition to cure CHD2	USA (Global)	https://www.curechd2.org/	https://www.facebook.com/curechd2.org	info@curechd2.org
CLCN4-related X-linked intellectual disability syndrome	ORPHA:485350	Cure CLCN4	UK	https://curecln4.org/	https://www.facebook.com/CureCLCN4/	info@curecln4.org
CSWSS	ORPHA:725	Asociacion de Afectados de Delecion 1p36	Spain	https://sindromePOCS.org	https://www.facebook.com/sindromePOCS	info@sindromePOCS.org
GABAAR variants		Cure GABA-A	US (global)	https://curegaba.org/	https://web.facebook.com/profile.php?id=100091946025243&_rdc=1&_rdr#	info@cureGABA.org
Down syndrome related epilepsy	ORPHA:870	European Down syndrome association	Europe	http://www.edsa.eu/down-syndrome/	https://www.facebook.com/edsachannel/	info@edsa.eu

Dup15q	ORPHA:3306	Dup15q France	France	https://www.dup15qfrance.fr	https://www.facebook.com/dup15qFrance/	info@dup15qfrance.fr
Dup15q	ORPHA:3306	Dup15q e.V.	Germany	https://dup15q.de/	https://www.facebook.com/dup15qeV	kontakt@dup15q.de
Dup15q	ORPHA:3306	Nonsolo15	Italy	http://www.idic15.it/		idic15@idic15.it
Dup15q	ORPHA:3306	Asociacion Nacional del sindrome idic15	Spain	https://www.idic15q.com/	https://www.facebook.com/profile.php?id=100064702145298&fref=ts	info@idic15q.com
ECHS1	ORPHA:653880	Cure ECHS1	USA (global)	https://cure-echs1.com/		contact.echs1@gmail.com
Encephalitis	ORPHA:97275	Encephalitis Society	UK	www.encephalitis.info	https://www.facebook.com/EncephalitisSociety/	mail@encephalitis.info
FIRES	ORPHA:163703	Association Paratonnerre	France	https://associationparatonnerre.org	https://www.facebook.com/AssociationParatonnerre/	contact@associationparatonnerre.org
FOXG1 syndrome	ORPHA:561854	Association FOXG1 France	France	https://foxg1france.fr/	https://www.facebook.com/FOXG1France/	contact@foxg1france.fr
FOXG1 syndrome	ORPHA:561854	FoxG1 Deutschland e.V.	Germany	https://www.foxg1.de/	https://www.facebook.com/foxg1de	info@foxg1.de
FOXG1 syndrome	ORPHA:561854	FOXG1 UK Charity	UK	http://www.foxg1.uk	https://www.facebook.com/foxg1uk/	information@foxg1.uk
Fragile X related epilepsy	ORPHA:908	European FRAGILE X Network	Europe	https://fragilex.eu/	https://www.facebook.com/EuropeanFragileXNetwork	
GRIA, GRID, GRIK, and GRIN		GRI France	France	https://gri-france.org/		
Hypothalamic Hamartoma	ORPHA:86906	Hope for Hypothalamic Hamartomas	UK	www.hopeforhh.org	https://www.facebook.com/hopeforhhuk	infouk@hopeforhh.org
Infantile spasms syndrome	ORPHA:3451	LES ENFANTS DE WEST	France	https://famwest.hypotheses.org/	https://www.facebook.com/groups/52683715391/	enfantsdewest@hotmail.fr
Infantile spasms syndrome	ORPHA:3451	FUNDACION SINDROME DE WEST	Spain	http://www.sindromedewest.org/	https://www.facebook.com/fundacionsindromedewest/	fundacion@sindromedewest.org

Infantile spasms syndrome	ORPHA:3451	UK Infantile Spasms Trust-UKIST	UK	https://ukinfantilespasmstrust.org/	https://www.facebook.com/UKInfantileSpasmsTrust/	ukinfantilespasmstrust@gmail.com
Jacobsen syndrome	ORPHA:2308	European Chromosome 11 Network	Europe	https://chromosome11.org/en/home/	https://www.facebook.com/EuropeanChromosome11Network/?fref=ts	board@chromosome11.org
KCNB1	ORPHA:442835	Association KCNB1 France	France	https://kcnb1-france.org/	https://www.facebook.com/kcnb1.france/	contact@kcnb1-france.org
KCNQ2-related epileptic encephalopathy	ORPHA:439218	European KCNQ2 Association	Europe	https://www.europeankcnq2association.com	https://www.facebook.com/europeankcnq2association	europeankcnq2association@gmail.com
KCNQ2-related epileptic encephalopathy	ORPHA:439218	KCNQ2 France Developpement	France	https://bit.ly/3IKPMQN	https://www.facebook.com/kcnq2France/	kcnq2francedev@gmail.com
KCNQ2-related epileptic encephalopathy	ORPHA:439218	KCNQ2 e.V.	Germany	https://kcnq2.de/	https://www.facebook.com/KCNQ2-eV-222465818324480	info@kcnq2.de
KCNQ2-related epileptic encephalopathy	ORPHA:439218	Asociacion KCNQ2 Espana	Spain	https://kcnq2espana.com/	https://www.facebook.com/Kcnq2/	kcnq2spain@gmail.com
KCNT1	ORPHA:293181	Une Etincelle Pour Isaure	France	https://uneetincellepourisaure.com/	https://www.facebook.com/profile.php?id=100081013027978	contact@uneetincellepourisaure.com
Ketogenic diet		Matthew's Friends	UK	https://www.matthewsfriends.org/	https://www.facebook.com/matthewsfriends/	enq@matthewsfriends.org
Leigh disease with myopathy	ORPHA:70474	Stowarzyszenie Dzieci z Miopatią Mitochondrialną pod postacią zespołu Leigha Mali Bohaterowie	Poland	https://www.malibohaterowie.org/		kuc.jolanta@interia.pl
Lennox Gastaut syndrome	ORPHA:2382	Associazione Famiglie LGS Italia	Italy	https://www.associazionelgs.it	https://www.facebook.com/LGSITALIA/	info@associazionelgs.it
Lennox Gastaut syndrome	ORPHA:2382	LGS Foundation	USA	https://www.lgsfoundation.org/	https://www.facebook.com/lgsfoundation1	info@lgsfoundation.org

Lissencephaly	ORPHA:95232	LISS - Anlaufstelle für Eltern und Angehörige an Lissenzephalie leidender Kinder e.V.	Germany	https://lissenzephalie.de/	https://www.facebook.com/Lissevkontakt/	kontakt@lissenzephalie.de
Metabolic diseases including ones that cause epilepsy		Association of Patients with Inherited Metabolic Diseases "ASPIDA ZOIS"	Cyprus	https://www.aspidazois.com/en/		info@aspidazois.com
Migrating partial seizures of infancy	ORPHA:293181	Asociacion Espanola de Afectados por Encefalopatía Epiléptica con Punta Onda Continua durante el Sueño - POCS	Spain	https://sindromePOCS.org	https://www.facebook.com/sindromePOCS	info@sindromePOCS.org
Neurotransmitter diseases		De Neu - Asociacion de Enfermedades de los Neurotransmisores	Spain	https://www.deneu.org/	https://www.facebook.com/asoc.deneu/	info@deneu.org
NEXMIF related developmental and epileptic encephalopathy		Unione Italiana Nexmif	Italy	https://www.nexmif.it/		info@nexmif.it
Development and epileptic encephalopathy 66 / PACS2 syndrome	ORPHA:442835	PACS2 Research Foundation	Poland	https://www.pacs2research.org/	https://www.facebook.com/pacs2research	pacs2research@gmail.com
PCDH19	ORPHA:101039	PCDH19 France	France	https://pcdh19france.fr/	https://www.facebook.com/pcdh19france	pcdh19france@gmail.com

PCDH19	ORPHA:101039	Insieme per la ricerca PCDH19 - Onlus	Italy	http://www.pcdh19research.org/index.html	https://www.facebook.com/insiemepcdh19/	insiemepcdh19@yahoo.it
PCDH19	ORPHA:101039	Asociacion Epilepsia Rosa Pcdh19	Spain	http://www.pcdh19.net	https://www.facebook.com/profile.php?id=100064857304148	
Progressive Myoclonic Epilepsies		AMARMYUL - Association contre la maladie rare myoclonique d'Unverricht-Lundborg	France	http://www.amarmyul.com/index.php		amarmyul@wanadoo.fr
PURA syndrome	438216	Pura Polska	Poland	in progress	in progress	monika.przetocka@fundacijapura.pl
Rare chromosomal anomaly		Valentin APAC- Association de Porteurs d'Anomalies Chromosomiques	France	https://www.valentin-apac.org/	https://www.facebook.com/AssoValentinAPAC/	contact@valentin-apac.org
Rare chromosomal anomaly		LEONA e.V.	Germany	https://www.leona-ev.de/start/	https://www.facebook.com/leonaverrein/	geschaefsstelle@leona-ev.de
Rare chromosomal anomaly		Unique-Rare Chromosome Disorder Support Group	UK	https://rarechromo.org/	https://www.facebook.com/Uniquerarechromosomedisordersupportgroup/	info@rarechromo.org
RASopathy (MEK2 mutation)	ORPHA: 638	MEK2 Research Foundation	Poland	in progress	in progress	mek2research@gmail.com
SLC13A5		Tess Research Foundation	USA (global)	https://www.tessresearch.org/	https://www.facebook.com/TESSresearch/	info@tessfoundation.org
SUDEP		SUDEP Action Day	UK	https://sudepactionday.org	https://www.facebook.com/SUDEPAction.org	info@sudep.org

