

ERN ITHACA 2025

Patient Advisory Board (PAB) ePAGs



Disorder(s)	Forename	Website	Country
22q11	Vesna Vujičić		Serbia
22q11.2	Anne Lawlor	https://www.22q11ireland.org	Ireland
22q11.2 and RD / ultra rare condition	Lyndsey Walsh	https://www.rdi.ie	Ireland
Acrodysostosis	Nina Knight	https://www.acrodysostosis.org/	UK
ADNP	Carole Herman	https://adnp-france.fr/	France
ADNP-kids-HVDAS	Kelly Verbruggen		Belgium
Angelman syndrome	Sergey Dmitriev	https://www.angelman.gr	Greece
Angelman syndrome	Samantha Eisenhauer	https://www.fastfrance.org	France
Angelman syndrome	Maria Palia	https://www.angelman.gr	Greece
Angelman syndrome	Olga Sophocleous	https://angelman.gr	Greece/Cyprus
Angelman syndrome	Ellen Koekoekx	https://www.cureangelman.org	USA
Angelman Syndrome	Suzan Genova	https://cureangelman.org	Bulgaria
Angelman syndrome	Lara Chappel	https://angelmanalliance.org	Netherlands
Angelman syndrome	Hanna Dianowé	https://cureangelman.org/articles/welcome-fast-poland	Poland
Beckwith–Wiedemann syndrome (BWS)	Monica Bertoletti	https://www.aibws.org	Italy
CACNA1C -Timothy Syndrome (TSA); LongQT8	Sophie Muir	https://timothysyndrome.org	UK
CDKL5-deficiency	Christina López García's	https://www.aacdkl5.org	Spain

CHAMP1	Daniel Palumbo	https://www.champ1foundation.eu	EU
Coffin Lowry syndrome	Iuliana Dumitriu	http://www.coffin-lowry.ro	Romania
Cornelia De Lange syndrome	Gerritjan Koekkoek	https://www.cdlsworld.org	Netherlands
Corpus Callosum Dysgenesis	Eszter Szabo	https://www.agykeregtest.hu	Hungary
CTNNB1 syndrome	Ana González Hernández	https://ctnnb1-foundation.org	Spain
Kabuki Syndrome	Jessica McAndrew	https://www.kabukisynromefoundation.org	USA
KAT6A; KAT6B syndrome	Silvia de la Flor	https://www.kat6france.org	France
KBG Syndrome	Annette Maughan	https://www.kbgfoundation.org	USA
KBG Syndrome	Juan Antonio Mesonero Escuredo	https://kbgsindrome.org	Spain
KIF1A-Associated Neurological Disorder (KAND)	Alejandro Doval	https://www.kif1a.es	Spain
Kleefstra syndrome	Inés Fernández-Ulibarri	https://inesfernandezulibarri.com	Spain/Germany
Kleefstra syndrome	Tanja Zdolšek Draksler	https://idefine-europe.org/kleefstra-syndrome	Slovenia
MEK2 mutation	Justyna Walczuk		Poland
Metachromatic leukodystrophy	Tomasz Grybek	https://fundacjabb.pl	Poland
Mitochondrial diseases	Emma Del-Rey	https://www.association-ammi.org	France
Noonan syndrome	Ioel Detton	https://www.assonoonan.fr	France
Pallister-Killian syndrome	Samantha Carletti	https://www.pksitalia.org	Italy
Phelan-McDermid Syndrome	Rui Barbosa Guedes	https://www.phelan-mcdermid.pt	Portugal
Pitt Hopkins syndrome	Sue Routledge	https://www.pitthopkins.org.uk	UK
PLWRD RERE Syndrome	Marta Balula	https://www.raramente.org	Portugal
Prader Willi syndrome	Dorica Dan	http://www.apwromania.ro	Romania
Rare diseases ID syndromes	Katarzyna Swieczkowska	https://www.psoni.gda.pl	Poland
Rare Diseases	Birgitta Termander	https://www.sallsyntadiagnoser.se	Sweden

SATB2 syndrome	Erika Stariha	https://www.satb2europe.org	Slovenia
Smith-Lemli-Opitz syndrome	Antje Enekwe	https://slodeu.wixsite.com	Germany
SCN2A syndrome	Zhana Chokheli		Georgia
Spina Bifida	Benoit Fourcroy	http://www.spina-bifida.org	France
Spina Bifida & Hypdrocephalus	Andrej Drdul	https://www.ifglobal.org	Slovakia
Spina Bifida & Hypdrocephalus	Sylvia Roozen	https://www.ifglobal.org	Belgium
Spina Bifida & Hypdrocephalus	Ammi Sundqvist-Andersson	https://stockholm.rbu.se	Sweden
SYNGAP1 syndrome	Marcos Mengual Hinojosa	https://www.syngap.de	Germany
SYNGAP1 syndrome	Verena Schmeder		Germany
Tuberous Sclerosis Complex	Mary Vasseghi	https://tscireland.org	Ireland
Undiagnosed	Claudia Beard	https://www.undiagnosed.org.uk	UK
Undiagnosed	Helene Cederroth	https://wilhelmfoundation.org	Sweden
Williams-Beuren syndrome	Gábor Pogány	https://www.williams.org.hu	Hungary
Williams-Beuren syndrome	Annalisa Scopinaro	https://www.apwitalia.org	Italy
Williams-Beuren syndrome	Tanja Zahlten	https://www.w-b-s.de	Germany