

ERN ITHACA Patient Advisory Board (PAB) (ePAGs) 2024 -2025



	Forename	Website	Country	Disorder(s)
ePAG	Marta Balula	raramente.org	Portugal	PLWRD RERE syndrome
ePAG	Rui Barbosa Guedes	www.phelan-mcdermid.pt	Portugal	Phelan-McDermid syndrome
ePAG Associate	Claudia Beard	https://www.undiagnosed.org.uk	UK	Undiagnosed
ePAG	Monica Bertoletti	https://www.aibws.org/	Italy	Beckwith–Wiedemann syndrome (BWS)
ePAG	Samantha Carletti	https://www.pksitalia.org	Italy	Pallister-Killian syndrome
ePAG Partnership	Helene Cederroth	https://wilhelmfoundation.org/	Sweden	Undiagnosed
ePAG Chair	Dorica Dan	http://www.apwromania.ro	Romania	Prader Willi syndrome
ePAG	Silvia de la Flor	https://www.kat6france.org	France	KAT6A, KAT6B syndrome
ePAG	Ioel Detton	https://www.assonoonan.fr	France	Noonan syndrome
ePAG Associate	Alejandro Doval	www.kif1a.es	Spain	KIF1A- Neurological Disorder (KAND)
ePAG	Andrej Drdul	https://www.ifglobal.org/	Slovakia	Spina Bifida & Hypdrocephalus
ePAG Associate	Iuliana Dumitriu	http://www.coffin-lowry.ro/	Romania	Coffin Lowry syndrome
ePAG	Samantha Eisenhauer	https://www.fastfrance.org/	France	Angelman syndrome
ePAG	Antje Enekwe	https://slodeu.wixsite.com/home	Germany	Smith-Lemli-Opitz syndrome
ePAG	Inés Fernández-Ulibarri		Spain/Germany	Kleefstra syndrome
ePAG	Benoit Fourcroy	http://www.spina-bifida.org/	France	Spina Bifida
ePAG	Suzan Genova	cureangelman.bg	Bulgaria	Angelman syndrome
ePAG	Ana González Hernández		Spain	CTNNB1 syndrome
ePAG SC	Tomasz Grybek	https://fundacjabb.pl/	Poland	metachromatic leukodystrophy
ePAG	Carole Herman	https://adnp-france.fr/	France	ADNP
ePAG Associate	Nina Knight	https://www.acrodysostosis.org	UK	Acrodysostosis
ePAG	Gerritjan Koekkoek	https://www.cdslworld.org	Netherlands	Cornelia De Lange syndrome
ePAG Associate	Ellen Koekoekx	www.cureangelman.org	USA	Angelman syndrome

ePAG	Anne Lawlor	https://www.22q11ireland.org	Ireland	22q11.2
ePAG	Christina López García's		Spain	CDKL5-deficiency
ePAG Associate	Annette Maughan	https://www.kbgfoundation.org	USA	KBG syndrome
ePAG Associate	Jessica McAndrew	https://www.kabukisyndromefoundation.org/	USA	Kabuki syndrome
ePAG-Alternate	Marcos Mengual Hinojosa	www.syngap.de	Germany	SYNGAP1 syndrome
ePAG	Juan Antonio Mesonero Escuredo	https://kbgsindrome.org/	Spain	KBG syndrome
ePAG Associate	Sophie Muir		UK	CACNA1C -Timothy Syndrome (TSA); LongQT8
ePAG-Alternate	Maria Palia		Greece	Angelman syndrome
ePAG	Daniel Palumbo	www.champ1foundation.eu	EU	CHAMP1
ePAG	Gábor Pogány	https://www.williams.org.hu; https://eurowilliams.org	Hungary	Williams-Beuren syndrome
ePAG-Alternate	Sylvia Roozen	www.ifglobal.org	Belgium	Spina Bifida & Hypdrocephalus
ePAG SC	Sue Routledge	www.pitthopkins.org.uk	UK	Pitt Hopkins syndrome
ePAG	Annalisa Scopinaro		Italy	Williams-Beuren syndrome
ePAG	Olga Sophocleous		Greece/Cyprus	Angelman syndrome
ePAG	Erika Stariha	www.satb2europe.org	Slovenia	SATB2 syndrome
ePAG	Ammi Sundqvist-Andersson	https://stockholm.rbu.se/	Sweden	Spina Bifida & Hypdrocephalus
ePAG SC	Katarzyna Swieczkowska	www.psoni.gda.pl	Poland	Rare diseases ID syndromes
ePAG	Eszter Szabo	https://www.agykeregtest.hu/	Hungary	Corpus Callosum Dysgenesis
ePAG	Mary Vasseghi	https://tscireland.org/	Ireland	Tuberous Sclerosis Complex
ePAG Associate	Kelly Verbruggen		Belgium	ADNP
ePAG	Verena Schmeder		Germany	SYNGAP1 syndrome
ePAG Associate	Vesna Vujičić		Serbia	22q11
ePAG-Alternate	Lyndsey Walsh	www.rdi.ie	Ireland	22q11.2 and RD / ultra rare condition
ePAG	Tanja Zahlten	https://www.w-b-s.de/	Germany	Williams-Beuren syndrome
ePAG Co-Chair	Tanja Zdolšek Draksler	https://idefine-europe.org/kleefstra-syndrome/	Slovenia	Kleefstra syndrome