Directory of ePAG [European Patient Advocacy Groups] representatives

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European Patient Advocacy Groups (ePAGs) are patient forums comprised by ePAG Patient Advocates designated by Patient Organisations wishing to engage in a specific European Reference Network (ERN). Each European Reference Network has one European Patient Advocacy Group.

Rare disease Patient Organisations, EURORDIS members and non-members established in Europe, may endorse ePAG Patient Advocates to be actively involved in the activities and governance structure of a European Reference Network. Each of the 24 ePAGs coordinates the recruitment of new ePAG Patient Advocates in collaboration with the ERN Network Coordinator. The application process and eligibility criteria is detailed in the **ePAG Constitution and Rules of Procedure.**

Rare Bone ePAG

- **Inês Alves**, Fundación ALPE Acondroplasia
- **Elisabeth Martin**, Association Ollier-Maffucci Europe
- **Tenna Toft Olesen**, XLH Patientenforeningen
- **Rebecca Tvedt Skarberg**, Osteogenesis Imperfecta Federation Europe

Rare Cancer ePAG

- **Catherine Bouvier**, NET patient Foundation UK
- **James Caldwell,** Northern Ireland Rare Disease Partnership
- **Iain Galloway**, MPNE Ocular/Rare
- **Stefan Gijssels,** Digestive Cancers Europe
- **Emma Kinloch,** Adenoid Cystic Carcinoma
- **Teodora Kolarova**, International Neuroendocrine Cancer Alliance (INCA)
- **Estelle Lecointe**, ASSOCIATION FRANÇAISE DES PATIENTS DU GIST - ENSEMBLE CONTRE LE GIST
- **Kathy Oliver**, International Brain Tumour Alliance (IBTA)
- **Roberto Persio,** Associazione Italiana Laryngectomizzati (AILAR)
- **Judith Taylor,** Thyroid Cancer Alliance
- **Markus Wartenberg ,** Das Lebenshaus e.V. - Die Organisation fürPatienten mit seltenen Soliden Tumoren:GIST/Sarkome/Nierenkrebs
- **Petya Zyumbileva,** Melanom Info Deutschland

Rare connective tissue and musculoskeletal diseases ePAG

- **Silvia Aguilera**, Asociación Española Síndrome Antifosfolipídico
- **Alain Cornet,** Lupus Europe
- **Charissa Frank,** Vlaamse Vereniging voor Erfelijke Bindweefselaandoeningen
- **Ilaria Galetti,** Gruppo Italiano per la Lotta alla Sclerodermia ONLUS
- **Juergen Grunert,** Deutsche Ehlers-Danlos Initiative e. V.
- **Ana Vieira,** Liga Portuguesa contra as Doenças Reumáticas
- **Lisa Matthew,** Relapsing Polychondritis Narrative

Rare craniofacial anomalies and ENT disorders ePAG

- **Gareth Davies**, European Cleft Organisation
- **Barbara Lieuwen**, Voorzitter Laposa
- **Thomas Luck,** Netzrverk Goldenhar-Syndrom undOhrmuscheldysplasie e.V.
- **Sandra Mösche,** Elterninitiative Apert-Syndrom und verwandte Fehlbildungen e. V
- **Sara Perez**, Asociación Nacional Sindrome de Joubert

- # Rare endocrine diseases ePAG

- **Johan Beun,** Bijniervereniging NVACP
- **Petra Bruegmann**, European MEN Alliance
- **Manuela Brösamle,** AGS- Eltern- und Patienteninitiative e.V.
- **Patricia Carl**, Bundesverband Kleinüschsige Menschen und ihre Familien e.V.
- **Johan de Graaf,** Nederlandse Hypofyse Stichting (Dutch Pituitary Foundation)
- **Carole Delorme,** Association surénale
- **Elisabetta Freo**, L'Associazione Famiglie di Soggetti con Deficit dell'Ormone della Crescita ed altre Patologie
- **Jo Grey**, Association for Multiple Endocrine Neoplasia Disorders
- **Jette Kristensen,** Addison Foreningen i Danmark
- **Martha Kirchhoff,** Phosphatdiabetes e. V.
- **Arlene Smyth**, Turner Syndrome Support Society

- **Marina Valenti,** Italian Association for Alstrom Syndrome
- **Diana Vitali,** SOD Italia - Associazione Italiana Displasia Setto Ottica e Ipoplasia del Nervo Ottico

Rare eye diseases ePAG

- **Christina Fasser,** Retina International
- **Gaelle Jouanjan,** FRANCE & ANIRIDIA EUROPE
- **Michael Längsfeld,** PRO RETINA Deutschland
- **Paula Morandi,** MITOCON ONLUS
- **Petia Stratieva,** Retina International
- **Dominique Sturz**, Usher Deafblind Forum Austria
- **Russel Wheeler,** Leber's Hereditary Optic Neuropathy Society

Rare gastrointestinal diseases ePAG

- **Duccio Cavalieri,** Associazione Italiana Morbo di Hirschsprung
- **Fanny Cauvet,** European Society of Pediatric Gastroenterology Hepatology and Nutrition
- **Antje Feldtmann-Korn,** Kise e.V
- **Joanne Fruithof,** The Federation of Esophageal Atresia and Tracheo-Esophageal Fistula Support Groups e.V.
- **Annette Lemli**, Selbsthilfeorganisation für Menschen mit Anorektalfehlbildungen
- **Beverley Power**, CDH UK
- **Nicole Schwarzer**, Selbsthilfeorganisation für Menschen mit Anorektalfehlbildungen
- **Graham Slater,** The Federation of Esophageal Atresia and Tracheo-Esophageal Fistula Support Groups e.V.
- **Anke Widenmann Grolig**, Patienten- und Selbsthilfeorganisation für Kinder und Erwachsene mit kranker Speiseröhre

Rare Pediatric Cancer ePAG

- **Luisa Basset**, Federación Española de Padres de Niños con Cáncer & Childhood Cancer International – Europe
- **Anne Goeres,** Fondatioun Kriibskrank Kanner & Childhood Cancer International – Europe
- **Lejla Kamerić,** Heart for kids with cancer in FBiH (Srce za djecu koja boluju od raka u FBiH) & Childhood Cancer International – Europe
- **Anita Kienesberger**, Austrian Childhood Cancer Organization & Childhood Cancer International – Europe

- # Rare hematological diseases ePAG

- **Pierre Aumont**, Association de Soutien et d'Information à la Leucémie Lymphoïde Chronique et la maladie de Waldenström
- **Dag Erling Stakvik,** European Federation of Associations of Patients with Haemochromatosis
- **Jan Geissler**, Leukemia Patient Advocates Foundation
- **Angelo Loris Brunetta**, Associazione Ligure Thalassemici Onlus
- **Ananda Plate**, Myeloma Patients Europe
- **Maria Piggin,** PNH Support UK
- **Sophie Wintrich**, MDS UK Patient Support Group
- **Baiba Ziemele,** Latvia Hemophilia Society

Rare hepatic diseases ePAG

- **Tess Harris**, PKD UK
- **Uwe Korst,** PKD Familiäre Zystennieren e.V.
- **Ernst Leitgeb,** Hepatitis Aid Austria
- **Lone McColaugh**, Leverforeningen
- **Biljana Mirceska**, NGO SLAP - Save Liver Association of Patients
- **Alison Taylor**, Children's Liver Disease Foundation
- **Camille Thrond-Charles,** AMFE Association Maladies FOIE
- **Martine Wallsmley,** PSC Support, UK
- **Jose Willemse,** Nederlandse Leverpatienten Vereniging

Rare hereditary metabolic disorders ePAG*

- **Renza Barbon**, UNIAMO
- **Lut de Baere**, BOKS
- **Julia Boonak,** CGD UK
- **Covadonge Diaz Martinez**, Asociación por la vida con GLUT-1
- **Beatruice Casado Verrrier,** Familial GA1
- **Tanaya Collin**, European Gaucher Alliance
- **Dorinda Dams Silva,** APCDG-RMD (CDG)
- **Hanka Dekker,** VKS, The Netherlands
- **Marie Devaux,** Phenylcetonurie, France
- **Luigi Distefano**, Association Française Niemann Pick

- **Rita Francisco**, APCDG
- **Alan Finglas,** MSD Action Foundation
- **Anne Hugon**, Association Francophone des Glycogénoses
- **Marija Joldic**, Association For Help And Support People With Hunter Syndrome in Serbia
- **Anne Gregoriades,** VML
- **Anne Kalweit,** SLO Deutschland
- **Enrique Landilo Contreras Pulido,** GSD Spain
- **Anne Grethe Lauridson,** European Gaucher Alliance
- **Daniel Lewi,** Cure & Action for Tay-Sachs Foundation
- **Nuno Marques,** Rarissimas (LSD)
- **Davidonis Marynas,** FIN (Fabry)
- **Toni Mathieson,** Niemann-Pick UK
- **Anna Merilouto,** Fabry International Network

- **Alexander Niehaus,** Asociación por la vida con GLUT-1
- **Laura Rossi Brunori,** AIG (Gaucher)
- **Thomas Schaller**, Pompe Deutschland e.V.
- **Silvia Sestini,** Associazione Italiana Malati di Alcaptonuria
- **Stefania Tobaldini,** Onlus Italy
- **Fernanda Torquati,** Gaucher, Italy
- **Erica Van der Mheen,** Fabry Support & Informatie Groep Nederland
- **Paula Videira**, APCDG -NMR (CDG)
- **Leona Wagner,** DSAKU

This list is incomplete and will be updated in due course

Rare immunological & auto inflammatory diseases ePAG

- **Zoi Anastasia,** Vasculitis UK
- **Eugenia Durante,** Associazione Pazienti della Sindrome di Churg Strauss
- **Jose Drabwell**, IPOPI - International Patient Organization for Primary Immunodeficiencies
- **Andrea Gressani,** Associazione per le immunodeficienze primitive Onlus
- **Jaana Grönholm,** Suomen Vaskuliittiyhdistys Ry
- **Saara Kiema**, Ipopi - International Patient Organization For Primary Immunodeficiencies
- **Diana Marinello**, Associazione Italiana Sindrome e Malattia di Behcet
- **Paul Morgan**, FMF & AID
- **Martine Pergent**, IPOPI - International Patient Organization for Primary Immunodeficiencies
- **Julie Power,** Vasculitis Ireland
- **Johan Prévot**, IPOPI - International Patient Organization for Primary Immunodeficiencies

- **Rachel Rimmer**, RACC – UK
- **Stephanie Skeffington,** Irish Children’s Arthritis Network
- **Leire Solis**, IPOPI - International Patient Organization for Primary Immunodeficiencies
- **Mailis Suhonen,** Suomen Vaskuliittiyhdistys Ry
- **Romana Torracca,** Associazione Pazienti della Sindrome di Churg Strauss
- **Malena Vetterli**, FMF & AID
- **Carlota Villar,** Barcelona PID Foundation

Rare malformations / developmental anomalies/and rare intellectual disabilities ePAG

- **Claudio Ales,** Associazione Italiana per la lotta alle PHTS
- **Lieven Bauwens,** International Federation for Spina Bifida and Hydrocephalus
- **Dorica Dan,** Romanian National Alliance for Rare Diseases
- **Sandrine Daugy**, Génération 22
- **Carole Herman**, Amis de ADNP France
- **Renée Jopp**, International Federation for Spina Bifida and Hydrocephalus
- **Gerritjan Koekkoek,** Cornelia De Lange Syndrome World Federation
- **Pietro Marinelli,** ASSOCIAZIONE SMITH-MAGENIS ASM17 ITALIA ONLUS
- **Yvonne Milne,** Rett Syndrome Europe
- **Gabor Pogany,** FEWS - Federation of European Williams Syndrome
- **Sue Routledge**, Pitt Hopkins UK

- **Annalisa Scopinaro,** APWItalia
- **Ammi Sundqvist**, International Federation for Spina Bifida and Hydrocephalus

Rare multi-systemic vascular diseases ePAG

- **Romain Alderweireldt,** Association Belge du Syndrome de Marfan ASBL
- **Maria Barea,** Vascular Anomaly Patient Association
- **Luisa Botella,** Asociación HHT España
- **Claudia Crocione,** Associazione Italiana Teleangectasia Emorragica Italiana - HHT ONLUS
- **Elena de Moya Rubio,** MARFAN HILFE DEUTSCHLAND E.V.
- **Karen Druckman,** HTT Swiss
- **Valentina Favalli**, Magica Onlus
- **Paolo Federici,** Associazione Fondazione Italiana HHT "Onilde Carini"
- **Charissa Frank,** Vlaamse Vereniging voor Erfelijke Bindweefselaandoeningen
- **Christina Grabowski,** HHT Europe
- **Juergen Grunert,** Deutsche Ehlers-Danlos Initiative e. V
- **Peter Hall,** Svenska Ödemförbundet

- **Pernille Henriksen,** Dansk Lymfødem Forening
- **Lex van der Heijden, Cutis Marmorata Telangiectatica Congenita and other Vascular Malformations**
- **Eline Hoogstra,** Nederlands Netwerk voor Lymfoedeem & Lipoedeem (NLNet)
- **Juan Lameiro,** Asociación Galega de Linfedema
- **Manuela Lourenço Marques,** National Association of Sufferers of Lymphatic Disorders Portugal **Lise Murphy,** Svenska Marfanföreningen
- **Elena Matta, Lymphido ONLUS**
- **Rafaella Restaino,**
- **Françoise Steinbach,** Association Marfans
- **Caroline Van Den Bosch,** HEVAS
- **Aaike van Oord**, LGD Alliance Europe

Rare neurological diseases ePAG

- **Astri Arnesen**, European Huntington Association
- **Monika Benson**, Dystonia Europe
- **Mary Kearney**, Friedreich's Ataxia Research Alliance Ireland (FARA)
- **Lori Renna Linton**, Euro-HSP
- **Lubomir Mazouch**, Spolek pro Atypické Parkinsonské syndromy
- **Marek Parowicz**, Association AHC18+ e. V.

Rare neuromuscular diseases ePAG

- **Bobby Ancil**, Muscular Dystrophy UK
- **Dimitrios Athanasiou,** MDA Hellas/UPPMD
- **Patrizia Blomkwist Markens**, Spierziekten Nederland - Dutch Patient Society of Neuromuscular Diseases
- **Joaquim Brites,** Associacao Portuguesa de Neuromusculares
- **Ria Broekgaarden**, Dutch Patient Society of Neuromuscular Diseases
- **Nic Bungay,** Muscular Dystrophy UK
- **Fernanda de Angelis**, Duchenne Parent Project Italy
- **Ingrid de Groot**, Spierziekten Nederland - Dutch Patient Society of Neuromuscular Diseases
- **Mencía de Lemus Belmonte,** Fundame Spain/SMA Europe
- **Emma Del Rey**, French Association for Mitochondrial Diseases
- **Marguerite Friconneau**, AFM-Téléthon
- **Rosanna Fodera**, MITOCON ONLUS
- **Patrizia Garzena,** CIDP Italia Onlus
- **Jérémie Gautreau,** French Association against Peripheral Neuropathies

- **Filippo Genovese,** ACMT-Rete
- **Madelon Kroneman,** Spierziekten Nederland
- **François Lamy,** AFM-Téléthon
- **San Jose Leticia,** ASEM Cataluna
- **Massimo Marra**, CIDP Italia Onlus
- **Alexandre Mejat**, AFM-Téléthon
- **Marisol Montolio,** Duchenne Parent Project Spain
- **Michela Onali**, Gli Equilibristi-HIBM
- **Marie-Christine Ouillade**, AFM-Téléthon
- **Françoise Pelcot**, French Association Against Amyloidosis
- **Alejandra Pereda**, Duchenne Parent Project Spain
- **Francisco Javier Pérez-Martinez,** Fundación Ana Carolina Díez Mahou/ AEPMI / Federación
- **Jean-Philippe Plançon,** French Association against Peripheral Neuropathies
- **Evy Reviers,** ALS Liga Belgium
- **Jacques Salama,** Former General Secretary of the Myology Institute
- **Sandrine Segovia-Kueny**, AFM-Téléthon

- **Inge Schwersenz,** Deutsche Gesellschaft für Muskelkranke
- **David Stephenson,** Muscular Dystrophy UK
- **Daniel Tanesse**, Charcot Marie Tooth-France/European CMT Federation
- **Isabela Tudorache**, PPMD
- **Diana van der Meij-Kim**, FSHD EU
- **Judit Varadine Csapo**, Angyalszarnyak Hungarian Muscle Dystrophy Association
- **Dominic Wells**, Muscular Dystrophy UK
- **Gerard Wellenberg**, Myotonic Dystrophy

Rare renal diseases ePAG

- **Alena Blaß**, aHUS
- **Marjolein Bos,** VKS-Cystinose Groep
- **Antonio Cabrera Cantero,** Hypomagnesemia
- **Suzanna Carvajal Arjona**, Hipofam
- **Flavia Galetti,** PKD
- **Tess Harris,** Ciliopathy Alliance
- **Christiane Mockenhaupt,** aHUS
- **Francisco Monfort,** aHUS
- **Daniel Renault,** Alport
- **Claudia Sproedt,** Cystinose Selbsthilfe e.V.
- **Marjolein Storm,** Nierpatienten Vereniging Nederland
- **Evy van Kempen,** Nierpatiënten Vereniging Nederland (Dutch Kidney Patients Association)

- **Marieke van Meel**, NephcEurope

Rare Skin Disorders ePAG

- **Marie-Claude Boiteux,** Cutis Laxa Internationale
- **Laurence Gallu,** Association Pemphigus – Pemphigoïdes France
- **Ulrike Holzer**, Selbsthilfegruppe Ektodermale Dysplasie e.V.
- **Ingrid Jageneau**, Debra Belgium vzw
- **Jose Manuel Montoya Gutierrez,** Asociación de Afectados por Displasia Ectodérmic
- **Diana Perry,** ED Society
- **Ivonne Ronchetti**, PXE-Italy
- **Bente Villumsen,** Hidrosadenitis-Denmark
- **Giulia Volpato,** p63 EEC Syndrome International network word communication
- **Jodi Whitehouse,** Caring Matters Now
- **Anna Wiegandt,** Selbsthilfe Ichthyose e.V.

- **Marjolein van Kessel,** Naevus Global
- **Lex van der Heijden**, Cutis Marmorata Telangiectatica Congenita and other Vascular Malformations

Rare urogenital diseases ePAG

- **Dalia Aminoff**, AIMAR
- **Serena Bartezzati**, AICI Associazione Italiana Cistite Interstiziale
- **Jurgen Hensen,** ICA Deutschland
- **Annette Lemli,** Selbsthilfeorganisation für Menschen mit Anorektalfehlbildungen
- **Christiann Groen**, Dutch Bladder Extrophy
- **June Rogers**, Bladder & Bowel UK
- **Conny Aust**, German Bladder Exstrophy Patient Organisation
- **Michael Männer**, Verein Lichen Sclerosus
- **Richard Stamp**, Orchid UK

- ## Rare Pulmonary ePAG

- **Marta Almagro,** European Lung Foundation
- **Edwin Brekelmans,** Alpha 1 Global
- **Alessandro Carcano,** Associazione Italiana Per La Sindrome Da Ipoventilazione Centrale Congenita
- **Hilde de Keyser,** Association Muco Vereniging
- **Liam Galvin,** Irish Lung Fibrosis Association
- **Carlee Gilbert,** ChILD Lung Foundation
- **Kate Hill,** June Hancock Mesothelioma Research Fund
- **Johann Hochreiter,** Lungenfibrose Forum Austria
- **Dagmar Kauschka,** ERWACHSENEN-HISTIOZYTOSE X E.V.
- **Filippo Martone**, Amici Contro la Sarcoidosi Italia ONLUS
- **Luc Matthysen,** Association de patients souffrant d'hypertension artérielle pulmonaire en Belgique
- **Gergely Meszaros,** Pulmonary Hypertension Association Europe

- **Cassidy Nicola,** Irish Lung Fibrosis Association
- **Karen O'Hara**, Alpha 1 Global
- **Stefano Pavanello**, Unione Trapiantati Polmone Di Padova
- **Pippa Powell,** European Lung Foundation
- **Marjo Poulissen**, Longfonds
- **Bernd Quadder,** Deutsche Sarkoidose Vereinigung Gemeinnütziger e.V.
- **Bernhard Rindlisbacher**, KARTAGENER SYNDROM & PRIMÄRE CILIÄRE DYSKINESIE(E.V.)
- **Bernd Stachetzki,** Sarkoidose Netzwerk
- **Stefano Guerini,** ASSOCIAZIONE NAZIONALE ALFA1-AT
- **Patrick Vandorpe**, HALO
- **Anke Widenmann-Grolig,** The Federation of Esophageal Atresia and Tracheo-Esophageal Fistula Support Groups e.V.

Rare Genetic Tumour Risk Syndromes ePAG

- **Claudio Ales**, Associazione Italiana per la lotta alle PHTS
- **João de Sousa e Silva,** NF Patients United
- **Tamara Hussong Milagre,** Evita (Portuguese Association of carriers of genetic mutations related to Hereditary Cancer)
- **Rita Magenheim,** German Li Fraumeni Association
- **Anne Micallef**, Europa Donna
- **Stefania Mostaccioli,** Lega per la Neurofibromatosi 2-OnlusItalian
- **Nicola Reents,** Familienhilfe Darmkrebs e.V. / Semi-Colon (German patient association for Lynch and Polyposis)
- **Claas Röhl**, NF Kinder – Verein zur Förderung der Neurofibromatoseforschung Österreich
- **Tanja Spanic**, Europa Donna Slovenia

Rare Epilepsies ePAG

- **Isabella Brambilla**, Dravet Italia Onlus
- **Sarka Kanova,** Epistop
- **Monica Lucente,** Blue one association
- **Barbara Nicol**, Purple Day Spain
- **Emma Nott,** Hope For Hypothalamic Hamartomas UK
- **Anita Noordhoff,** KCNT1
- **Carol Anne Partridge**, CDKL5
- **Torie Robinson,** Epilepsy Sparks
- **Rosaria Vavassori, Italian patient organization for AHC, A.I.S.EA Onlus**
- **Allison Watson**, Ring 20 UK

Transplantation in Children ePAG*

- **Matilde Correia**, Associação Portuguesa de Insuficientes Renais
- **Pisana Ferrari**, PHA Europe
- **Juan Fuertes**, PHA Europe
- **Nana Gomex Mayoral,** HEPA
- **Sandrine Lefrancois** - Association ADAAT Alpha1-France
- **Daniela Paulo,** Portuguese Children With Liver Disease (HEPATIX)
- **Stefano Pavanello,** Unione Trapiantati Polmone di Padova
- **Alba Santos,** NUPA
- **Alison Taylor**, Children's Liver Disease Foundation Diverse Disease
- **Evy van Kempen,** Beleidsmedewerker Eigen Regie & Ervaringskennis delen
- **Conchita Velázquez-Gaztelu,** NUPA
- **Kristina Zaveckiene**, Lithuanian Children's Cancer Association

This list is incomplete and will be updated in due course

Rare and Low Prevalence Complex Diseases of the Heart ePAG

- **Ruth Biller,** ARVC-Selbsthilfe e.V
- **Edward Callus**, European Congenital Heart Disease Organisation
- **Ester Costafreda**, Asociacion SAMS
- **Steven Cox,** Cardiac Risk in the Young
- **Simone de Vergori,** ACMRC ONLUS Associazione Cardiomiopatie e Malattie Rare Connesse ONLUS
- **Katja Laine,** sydanlapset ja-aikuiset (Finnish Association for Heart Children and Adults)
- **Simone Louisse**, Stichting Hart4Onderzoek
- **Peter Nordquvist**, Swedish Heart Foundation
- **Sophie Pierre**, Association des Maladies héréditaires du Rythme Cardiaque
- **Inge Schalkers,** Haarteraad
- **Lisbeth Vestergaard Andersen,** Danish Heart Foundation
- **Dayenne Zwaagman,** Stichting Hart4Onderzoek