



Directory of ePAG [European Patient Advocacy Groups] representatives

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1. Rare Bone ePAG

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

2. Rare Cancer ePAG

- **Kathy Oliver**, International Brain Tumour Alliance
- **Isabelle Manneh-Vangramberen**, European Cancer Patient Coalition (ECPC)
- **Estelle Lecointe-Artzner**, Sarcoma Patients EuroNet (SPAEN)
- **Markus Wartenberg**, Sarcoma Patients EuroNet (SPAEN)
- **Teodora Kolarova**, International Neuroendocrine Cancer Alliance (INCA)
- **Catherine Bouvier**, International Neuroendocrine Cancer Alliance (INCA) & NET Patient Foundation UK
- **Jo Grey**, AMEND UK
- **Iain Galloway**, MPNE Ocular/ Rare

3. Rare connective tissue and musculoskeletal diseases ePAG

- **Irene Pitsillidou**, Cyprus League Against Rheumatism
- **Kirsten Lestrom**, Lupus Europe
- **Sara Badreh**, Lupus Europe
- **Charissa Frank**, Bindweefsel.be (Vlaamse Vereniging voor Erfelijke Bindweefselaandoeningen vzw)
- **Jürgen Grunert**, Deutsche Ehlers-Danlos-Initiative e.V.
- **Marianne Riviere**, Association Française du Lupus et autres Maladies Auto-Immunes (AFL +)
- **Ilaria Galetti**, Sosteni La Ricerca per la Sclerosi Sistemica Sclerodermia

4. Rare craniofacial anomalies and ENT (ear, nose and throat) disorders ePAG

- Sara Perez, Asociación Nacional Síndrome de Joubert
- Gareth Davies, European Cleft Organisation
- Barbara Lieuwen, Voorzitter Laposa

5. Rare endocrine diseases ePAG

- **Johan de Graaf**, Nederlandse Hypofyse Stichting
- **Jette Kristensen**, Addison Foreningen i Danmark
- **Johan Beun**, Bijniervereniging NVACP
- **Petra Bruegmann**, Netzwerks Hypophysen- und Nebennierenerkrankungen e. V.
- **Diana Vitali**, Associazione Italiana Displasia Setto Ottica e Ipoplasia del Nervo Ottico
- **Marika Porrey**, Schildklier Organisatie Nederland
- **Manuela Brösamle**, AGS-Eltern- und Patienteninitiative e.V.
- **Elisabetta Freo**, A Fa DOC
- **Carole Delorme**, Association Surrénales France
- **Patricia Carl**, BKMF e.V.
- **Jo Grey**, AMEND UK

6. Rare eye diseases

- **Christina Fasser**, Retina International
- **Avril Daly**, Retina International
- **Michael Längsfeld**, PRO RETINA Deutschland
- **Paula Morandi**, MITOCON ONLUS
- **Gaëlle Jouanjan**, France & Aniridia Europe
- **Daniela Brohlburg**, Pro Retina Deutschland
- **Russel Wheeler**, Leber's Hereditary Optic Neuropathy Society

7. Rare gastrointestinal diseases

- **Anke Widenmann-Grolig**, EAT, The federation of Esophageal Atresia and Tracheo-esophageal fistula support groups e.V.
- **Graham Slater**, EAT, The federation of Esophageal Atresia and Tracheo-esophageal fistula support groups e.V.
- **Joanne Fruithof**, EAT, The federation of Esophageal Atresia and Tracheo-esophageal fistula support groups e.V.
- **Frederic Armand**, EAT, The federation of Esophageal Atresia and Tracheo-esophageal fistula support groups e.V.
- **Nicole Schwarzer**, SoMA for Hirschsprung Disease and Anorectal Malformation
- **Fanny Cauvet**, Association APEHDia
- **Bailly Villette**, LEONORE
- **Antje Feldtmann-Korn**, KISE
- **Benoit Decavele**, La Vie par un Fil
- **Beverley Power**, CDH UK

8. Rare Pediatric Cancer ePAG

- **Anita Kienesberger**, Austrian Childhood Cancer Organization & Childhood Cancer International – Europe
- **Stephanie Shremmer**, Austrian Childhood Cancer Organization & Childhood Cancer International – Europe
- **Luisa Basset**, Federación Española de Padres de Niños con Cáncer & 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
- **Anne Goerens**, Fondation Kribskrank Kanner

9. Rare hematological diseases ePAG

- **Amanda Bok**, European Haemophilia Consortium
- **Jan Geissler**, Leukemia Patient Advocates Foundation
- **Sophie Wintrich**, MDS UK Patient Support Group
- **Angelo Loris Brunetta**, Associazione Ligure Talassemici Onlus
- **Ananda Plate**, Myeloma Patients Europe
- **Pierre Aumont**, Association de Soutien et d'Information à la Leucémie Lymphoïde Chronique et la maladie de Waldenström

10. Rare hepatic diseases ePAG

- **Marleen Kaatee**, PSC Patients Europe
- **Robert Mitchell Thain**, PBC Foundation
- **Lone McColaugh**, Leverforeningen
- **Alison Taylor**, Children's Liver Disease Foundation
- **Biljana Mirceska**, NGO SLAP - Save Liver Association of Patients
- **Sandrine Lefrancois**, Alpha 1 France
- **Angela Leburge**, albi-france
- **Joan Bedlington**, Liver North
- **Ingo van Thiel**, Deutsche Leberhilfe e.V.
- **Clémentine Duflo**, AMFE
- **Helen Morement**, AMMF
- **Virginie Tsilibaris**, FILFOIE
- **Luis Torres Alonso**, HePA
- **Stephane Coutin**, AMVF
- **Valerie Wheeler**, Wilson's Disease Support Group – UK
- **Norbert Klusen**, Yael Stiftung
- **Anna Holgerssen**, PSC Sweden
- **Ernst Leitgeb**, Hepatitis Hilfe Österreich
- **Hanna Eloranta**, Finnish Kidney and Liver Association
- **Catarina Lindström**, Leverförbundet
- **Karen Topaz Druckman**, HHT-Swiss

11. Rare hereditary metabolic disorders ePAG

- **Anne-Sophie Lapointe**, Vaincre les Maladies Lysosomales
- **Anne Hugon**, Association Francophone des Glycogenoses
- **Cinzia Arbellino**, MITOCON ONLUS
- **Lut de Baere**, Belgische Organisatie voor Kinderen en Volwassenen met een Stofwisselingsziekte
- **Rita Francisco**, Associacao Portuguesa CDG e outras Doencas Metabolicas

12. Rare immunological & auto inflammatory diseases ePAG

- **Diana Marinello**, Associazione Italiana Sindrome e Malattia di Behcet
- **John Mills**, Vasculitis UK
- **Peter Verhoeven**, Vasculitis Stichting
- **Richard West**, Behcets International
- **Malena Vetterli**, FMF & AID
- **Leire Solis**, IPOPI - International Patient Organization for Primary Immunodeficiencies ????
- **Andrea Gressani**, Associazione per le Immunodeficienze Primitive Onlus
- **Carlota Villar**, Barcelona PID Foundation

13. Rare malformations / developmental anomalies/and rare intellectual disabilities ePAG

- **Tobias Arndt**, European Dysmelia Reference Information Centre
- **Dorica Dan**, Romanian Prader Willi Association
- **Gabor Pogany**, Hungarian Williams Syndrome Association
- **Yvonne Milne**, Rett Syndrome Europe
- **Ammi Sundqvist**, International Federation Spina Bifida & Hydrocephalus
- **Becky Jenner**, Rett UK
- **Cor Oosterwijk**, VSOP
- **Paul Kavanagh**, ASSERT UK
- **Claudio Ales**, PTEN Italia
- **Pietro Marinelli**, Associazione Smith-Magenis ASM17 Italia onlus
- **Lisa Jaynecourt, Andrea Baines, Rachel Martin, Katie Cunnea**, Angelman Syndrome UK
- **Nick Meade**, Genetic Alliance UK
- **Anne-Sophie La Pointe**, Vaincre les Maladies Lysosomales
- **Françoise Neuhaus**, 22Q10 association
- **Sue Routledge**, Pitt Hopkins UK
- **Luis Quaresma**, ASBHIP

14. Rare multi-systemic vascular diseases ePAG

- **Caroline Van den Bosch**, HEVAS
- **Lise Murphy**, Svenska Marfanföreningen
- **Juergen Grunert**, Ehlers-Danlos Initiative Deutschland e.V.
- **Christina Grabowski**, Morbus Osler Selbsthilfe e.V.
- **Natascha Assies**, NLNet
- **Claudia Crocione**, Associazione Italiana Teleangectasia Emorragica
- **Romain Alderweireldt**, ASBM Association Belge du Syndrome de Marfan
- **Nele Verhaegen**, BeLymph
- **Luisa Maria Botella**, Asociacion HTT Espana
- **Paolo Federici**, Associazione Fondazione Italiana HHT 'Onilde Carini'
- **Ange Van Der Velden**, LGD Alliance Europe
- **Valentina Favalli**, Magica Onlus
- **Maria Barea**, Franziska Haiml, VASCAPA

15. Rare neurological diseases ePAG

- **Cathalijne van Doorne**, euro-Ataxia and European Federation of Neurological Associations
- **Mary Kearney**, Friedreich's Ataxia Research Alliance Ireland (FARA)
- **Tsveta Schyns-Liharska**, European Network for Research on Alternating Hemiplegia, ENRAH
- **Astri Arnesen**, European Huntington Association
- **Robert Scholten Apeldoorn**, Dutch Dystonia Association
- **Lori Renna Linton**, Euro-HSP
- **Amanda Rose**, Rare Dementia Support UK
- **John McFarlane**, EPU, PoPSyCLE

16. Rare neuromuscular diseases ePAG

- **François Lamy**, AFM Téléthon
- **Jean-Philippe Plançon**, French Association against Peripheral Neuropathies
- **Rosanna Fodera**, Mitocon Onlus, Italy
- **Evy Reviere**, ALS Liga Belgium
- **Judit Varadine Csapo**, Angyalszarn yak Hungarian Muscle Dystrophy Association
- **Marisol Montolio**, Duchenne Parent Project Spain

17. Rare renal diseases ePAG

- **Daniel Renault**, FEDERG
- **Claudia Sproedt**, CystinoseSelbsthilfe e.V
- **Susie Gear**, Alport UK
- **Marieke van Mel**, NephcEurope (TBC)
- **Francisco Montfort**, aHUS (TBC)
- **Nacho Nunez**, aHUS (TBC)
- **Etienne Gosyng**, Barrter and Gitelman (TBC)
- **Bruno Woitrin**, Barrter and Gitelman (TBC)
- **Marjoleine Bos**, VKSCystinose Groep (TBC)
- **Tess Harris**, Polycystic Kidney Disease Charity UK
- **Michel Schenkel**, ADPKD

18. Rare skin disorders ePAG

- **Ingrid Jageneau**, Debra Belgium vzw
- **Ulrike Holzer**, Selbsthilfegruppe Ektodermale Dysplasie e.V
- **Ivonne Ronchetti**, PXE-Italy Onlus
- **Jodi Whitehouse**, Caring Matters Now
- **Olivia Gross-Khalifa**, Association Française des Dysplasies Ectodermiques
- **Jose Manuel Montoya Gutierrez**, Asociacion de Afectados por Displasia Ectodermica
- **Sophie Le Pallec**, Amalyste
- **Mike Jaega**, DEBRA International
- **Laurence Gallu**, Association Pemphigus – Pemphigoïdes France
- **Mandy Aldwin**, European Network for Ichthyosis
- **Wafa Chaabi**, Enfants de la Lune
- **Marie-Claude Boiteux**, Cutis Laxa Internationale
- **Jacques Monnet**, Incontinenta Pigmenti France
- **Evanina Morcillo Makow**, DEBRA España
- **Bente Villumsen**, Danish HS Patients' Association
- **Christian Moser**, XP Freunden
- **Flavio Minelli**, Unione Italiana Ittiosi
- **Clare Robinson**, DEBRA UK
- **Cinzia Pilo**, Francesca Sofia, DEBRA Italia ONLUS

19. Rare urogenital diseases ePAG

- **Serena Bartezzati**, AICI Associazione Italiana Cistite Interstiziale
- **Dalia Aminoff**, AIMAR
- **Massimo Di Grazia**, Associazione Italiana Estrofia Vescicale-Epispadia Onlus

20. Rare Pulmonary ePAG

- **Fillipo Martone**, Amici Contro la Sarcoidosi Italia Onlus
- **Bernd Stachetzki**, Sarkoidose Netzwerk
- **Liam Galvin**, EU-IPFF
- **Bernd Quadder**, Deutsche Sarkoidose-Vereinigung e.V. (DSV)
- **Gergely Meszaros**, PHA Europe
- **Marta Almagro**, ELF, Bronchiectasis Patient Advisory Group
- **Kate Hill**, June Hancock Mesothelioma Research Fund
- **Pisana Ferrari**, PHA Europe
- **Pippa Powell & Sara Mansfield**, European Lung Foundation
- **Patrick Vandorpe**, HALO
- **Nicola Cassidy**, ILFA
- **Dagmar Kauschka**, Lungfibrose e.V.
- **Stefano Guerini**, Alfa₁-AT Italia
- **Hilde De Keyser**, Cystic Fibrosis
- **Alessandro Carcano**, A.I.S.I.C.C.
- **Edwin J. Brekelmans**, Alpha 1 Global
- **Luc Matthysen**, PHA Europe
- **Carlee Gilbert**, ChILD EU
- **Karen O'Hara**, Alpha 1 Global

21. Rare Genetic Tumour Risk Syndromes ePAG

- **Claas Röhl**, NF Kinder – Verein zur Förderung der Neurofibromatoseforschung Österreich
- **Anne Micallef**, Europa Donna
- **Claudio Ales**, Associazione Italiana per la lotta alle PHTS
- **Juergen Seppen**, Lynch-Polyposis
- **Stefania Mostaccioli**, Italian Association for NF2 and Schwannomatosis (NF3)
- **Tamara Hussong Milagre**, EVITA

22. Rare Epilepsies ePAG

- **Isabella Brambilla**, Dravet Italia Onlus
- **Anne-Sophie Hallet**, Alliance Syndrome de Dravet
- **Nathalie Coque**, Alliance Syndrome Dravet
- **Emma Williams**, Matthew's Friends
- **Myra de Groot**, Dravet Nederland/Belgium
- **Allison Watson**, Ring 20 UK
- **Carol Anne**, CDKL5
- **Barbara Nicol**, Purple Day Spain
- **Judette Lancrenon**, The Paratonnerre Association for FIRES
- **Fabrizio Mistretta**, Insieme per la Ricerca PCDH19 Onlus

23. TRANSCHILD

- **Juan Fuertes**, PHA Europe
- **Pisana Ferrari**, PHA Europe
- **Daniela Paulo**, Portuguese Children With Liver Disease (HEPATIX)
- **Conchita Velázquez-Gaztelu**, NUPA
- **Evy van Kempen**, Beleidsmedewerker Eigen Regie & Ervaringskennis delen

24. GUARD HEART

- **Edward Callus**, European Congenital Heart Disease Organisation
- **Sophie Pierre**, Association des Maladies héréditaires du Rythme Cardiaque
- **Brenda Alsemgeest**, Stichting Hart4Onderzoek
- **Patricia Vlasman**, Stichting Hart4Onderzoek
- **Dayenne Zwaagman**, Stichting Hart4Onderzoek
- **Ester Costafreda**, Asociacion SAMS
- **Peter Nordqvist**, Hjärte Barno Fonden
- **Steve Adams**, Cardiac Risk in the Young