ERN ITHACA 2025 ePags, PAB

carousel

"More united and motivated to collaborate together"

European Commission

EUROPEAN REFERENCE NETWORKS

Helping patients with rare or low-prevalence complex diseases

European Reference Networks ITHACA

- Marta Balula

marta.balula@raramente.org

- Raramente, portugal

- Rare syndromes

- Empowering to advance Care, Knowledge, and Advocacy for Rare Lives

- Rui Barbosa Guedes

rui@phelan-mcdermid.pt

- APMP Associçao, Portugal
- Phelan-McDermid

- Beacon of Hope for brighter future

Claudia Beard

claudiabeard2020@gmail.com

Undiagnosed

SWAN UK - Genetic Alliance UK

- Monica Bertoletti

monica@aibws.org

- AIBWS Odv Italia, Association
- Beckwith-Wiedemann Syndrome

- Sharing, collaboration, better perspective

Samantha Carletti

info@pksitalia.org

Pallister-Killian syndrome

PKS Italia - Associazione Sindrome di Pallister-Killian APS

Helene Cederroth

helene@wilhelmfoundation.org

Undiagnosed

Wilhelm Foundation, Sweden

- Lara Chappell

lara.chappell@angelmanalliance.eu

- Angelman Syndrome Alliance, EU

- Collaborating together to bring more useful solutions to our loved ones and families.

- Zhana Chokheli

zhanachokheli02@gmail.com

- Georgian Alliance for Rare Diseases (GARD)

- I fight for the world where rare matters !

European Reference Networks ITHACA

- Dorica Dan

dorica.dan@eurordis.org

- Prader Willi Association, Romanian National Alliance for Rare Diseases

- Living together, learning together!

- Silvia de la Flor

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- KAT6 France

- Syndrome KAT6A & Resilience

Emma Del-Rey

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Mitochondrial diseases

L'AMMi FRANCE

- Ioel Detton

assonoonan@gmail.com

- Noonan France

- Alone, we go faster; together, we go further

- Hanna Dianow

hanna.dianow@cureangelman.pl

- Foundation for Angelman Syndrome Therapeutics (FAST) Poland

- Advancing Angelman syndrome awareness and support

- Sergey Dmitriev

sergio_row08@hotmail.com

- Angelman Syndrome Greece // Rett Syndrome Greece

- No one stands alone, No one is left behind !

Alejandro Doval

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KIF1A-Associated Neurological Disorder (KAND)

Asociación KIF1A España

- Andrej Drdul

andrej.drdul@gmail.com

- IF - International Federation for Spina Bifida and/ or Hydrocephalus
- Slovak Association for Spina Bifida and/ or Hydrocephalus

- We transform fear from the unknown for friendship and trust. A live a full-fledged, joyful life.

Samantha Eisenhauer

samantha.eisenhauer@gmail.com

Angelman syndrome

France, FAST-Foundation Angelman Syndrome

- Antje Enekwe

a.enekwe@gmail.com

- SLO Deutschland e.V, Smith-Lemli-Opitz Syndrome

- Let us make a difference for people with rare diseases.

- Ines Fernandez Ulibarri

ifernaul80@hotmail.com

- Asociación Kleesfstra España
- Kleefstra syndrome

- Together to Change the Future of Rare

- Benoit Fourcroy

benoit.f@spina-bifida.org

- ASBH - spina-bifida.org

- Advances in healthcare and peer support

- Suzàn Genova
  suzan.m.genova@gmail.com

- FAST Bulgaria - Angelman syndrome; Nataliya Foundation - Isovaleric acidemia

- A shared path to hope

- Ana González Hernández
ana@ctnnb1-foundation.org
- Extra rare diseases

- Extraordinarily united

European Reference Networks ITHACA

- Tomasz Grybek

tomek@fundacjabb.pl

- FBB - Foundation of Borys the Hero.

- True Partners in the rare disease world.

- Carole Herman

caroleheman2012@gmail.com

- ADNP France

- Hope with amis de ADNP

- Nina Knight

ninasknight@hotmail.co.uk

- Acrodysostosis Support & Research UK World

- Together we accelerate the science and create a pathway to treatments

- Gerritjan Koekkoek

gerritjan@cdlsworld.org

- CdLS-World
- Cornelia de Lange syndrome

- Everyone knows something, together we know everything

- Ellen Koekoeckx

ellen.koekoeckx@cureangelman.org

- FAST - Foundation for Angelman Syndrome Therapeutics

- Working together towards a better life for our loved ones living with a rare disease

- Anne Lawlor

alawlor4@gmail.com
- 22q11 Ireland

- Together for Rare Families

- Cristina López García

relacionesinternacionales@aacdkl5.org

- Asociación de Afectados CDKL5

- Support families, impulse research and raise awareness

- Jessica McAndrew

jessica@kabukisyndromefoundation.org

- Kabuki Syndrome Foundation

- Building brighter futures for those affected by rare disease

- Juan Antonio Mesonero Escuredo

admin@kbgsindrome.org

- Asociacion Espagnola Syndrome KBG

- Health is a state of complete physical, mental and social well-being and not merely the absence of disease or infirmity.

- Sophie Muir

sophie@timothysyndrome.org

- Timothy Syndrome Alliance (TSA)
- CACNA1C-related disorders, including TSA and LongQT8

- Diagnosis unlocks understanding progress takes all of us

- Daniele Palumbo

dp@champ1foundation.eu

- CHAMP1 Foundation – Europe

- If you don't try, you'll never succeed."

- Pogány Gábor

pogany@williams.org.hu

- Hungarian Williams Syndrome Association (HWSA), (FEWS) – (HUFERDIS, Rare Diseases Hungary)
- Williams Syndrome

- Start by doing what's necessary then do what's possible and suddenly you are doing the impossible” (Saint Francis of Assisi)

- Sue Routledge

sue@pitthopkins.org.uk

- Pitt Hopkins UK and Stichting
- Pitt-Hopkins syndrome

- Learning and going forward together

- Annalisa Scopinaro

annalisa.scopinaro@yahoo.it

- APW Italia
- Williams Syndrome

- Together we can

- Olga Sophocleous

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- Angelman syndrome Greece

- Common vision for our rare patient’s better futur

- Erika Stariha

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- SATB2 Europe

- Hope Becomes real when we come together

- Ammi Andersson

ammi.andersson@cordnode.se

- RBU Rörelsehindrade Barn och Ungdomar Stockholm
- Spina Bifida and Spinal Dyphrasism

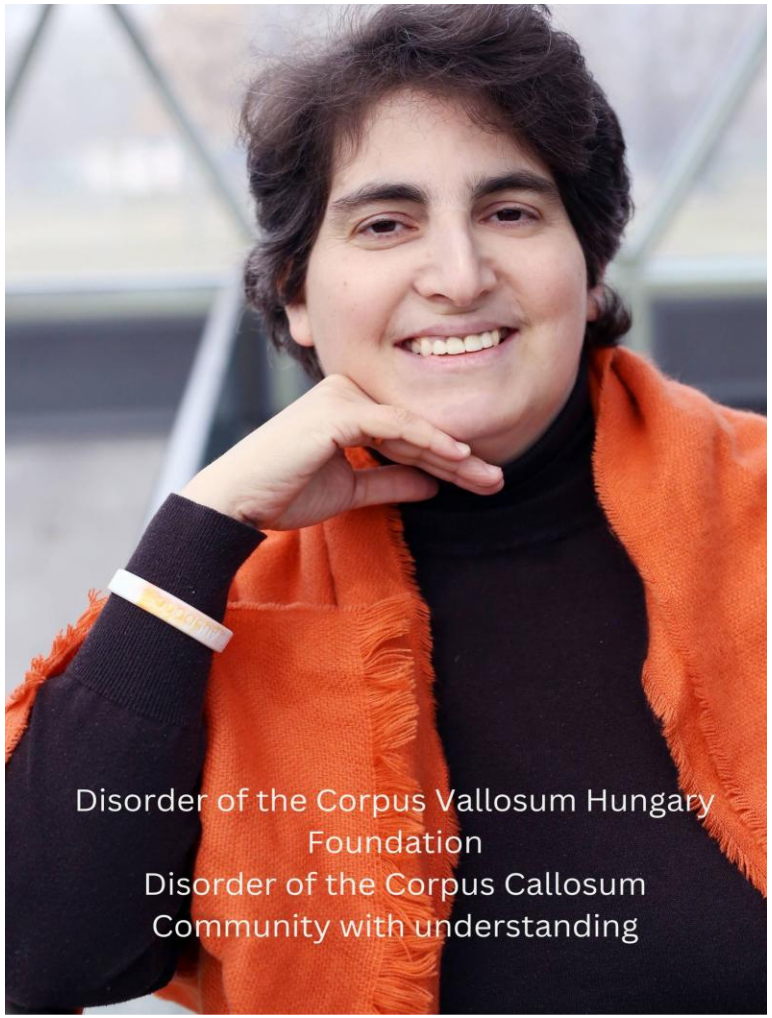
- Cooperate with clinicians to create an equal specialist care for rare conditions in every country within EU

- Katarzyna Świeczkowska

katarzyna.swieczkowska@psoni.gda.pl

- Polish Association for Persons with Intellectual Disability.

- Developing networks, networks for development.



- Eszter Szabóné Katona

szabo.esther@gmail.com

- Disorder of the Corpus Callosum Hungary Foundation

- Community with understanding

Termander

- Birgitta Termander

birgitta.termander@sallsyntadiagnoser.se

- Rare Diseases Sweden

A better Rare life in all areas

Mary Vasseghi

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Tuberous Sclerosis Complex
TSC, Ireland

- Kelly Verbruggen

adnpkids.hvdasbelgium@gmail.com

- ADNP kids Belgium

- Helsmoortel - Van der Aa syndrome (aka ADNPsyndrome)

- Striving to a better understanding of Helsmoortel Van der Aa syndrome and supporting parents across the world

European Reference Networks ITHACA

Verena Schmeder

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SYNGAP1 syndrome

SYNGAP Elternhilfe, Germany

- Vesna Vujičić

22qexyu@gmail.com

- 22q ex Yu, Serbia

- Together we can change the future

- Justyna Walczuk

mek2research@gmail.com

- MEK2 Research Foundation

- Committed to advancing research on MEK2 mutations and related rasopathies

- Lyndsey Walsh

lyndsey.1@hotmail.com

- Rare Diseases Ireland, 22q11 Ireland and Ultra-Rare Diseases

- Together we are a strong partnership for change

- Tanja Zahlten

tahischle@hotmail.com

- German Bundesverband WBS
- Williams Beuren Syndrom

- Share knowledge and learn from each other

- Tanja Zdolšek Draksler

tanja.zdolsek@ijs.si

- IDefine Europe Foundation
- Kleefstra syndrome

- Data is the key

Thank you for your attention !

https://ern-ithaca.eu

UBUNTU

" I am because we are."

European Reference Networks ITHACA