



“UBUNTU ”

*“More united and motivated to collaborate together”*



*(ITHACA’s ePags - PAB carousel)*



European Commission

**EUROPEAN REFERENCE NETWORKS**

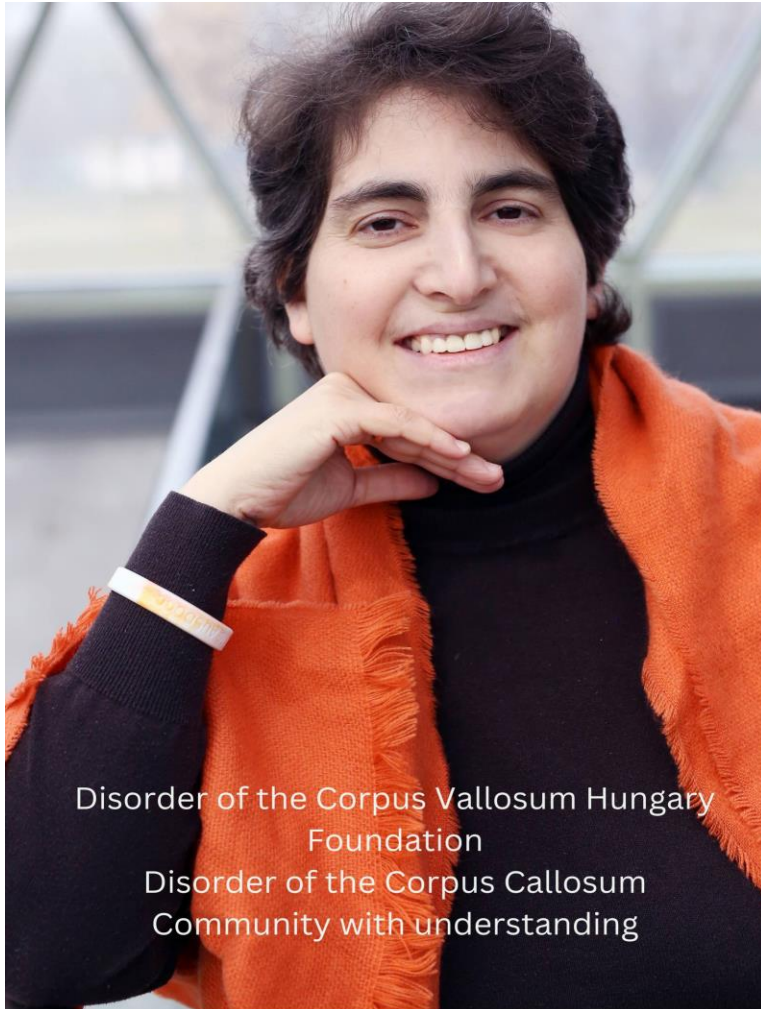
Helping patients with rare or low-prevalence complex diseases



- Tomasz Grybek  
[tomek@fundacjabb.pl](mailto:tomek@fundacjabb.pl)
- FBB - Foundation of Borys the Hero.
- True Partners in the rare disease world.



- Juan Antonio  
[admin@kbgssyndrome.org](mailto:admin@kbgssyndrome.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.



- Eszter Szabóné Katona  
[szabo.esther@gmail.com](mailto:szabo.esther@gmail.com)
- Disorder of the Corpus Callosum  
Hungary Foundation
- **Community with understanding**



- Ellen Koekoeckx  
[ellen.koekoeckx@cureangelman.org](mailto: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



- Daniele Palumbo  
[dp@champ1foundation.eu](mailto:dp@champ1foundation.eu)
- CHAMP1 Foundation – Europe
- If you don't try, you'll never succeed."



- Katarzyna Świeczkowska  
[katarzyna.swieczkowska@psoni.gda.pl](mailto:katarzyna.swieczkowska@psoni.gda.pl)
- Polish Association for Persons with Intellectual Disability.
- Developing networks, networks for development.



- Silvia de la Flor  
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- KAT6 France
- Syndrome KAT6A & Resilience





- Gerritjan Koekkoek  
[gerritjan@cdlsworld.org](mailto:gerritjan@cdlsworld.org)
- CdLS-World
- Cornelia de Lange syndrome
- Everyone knows something,  
together we know everything



- Carole Herman  
[caroleheman2012@gmail.com](mailto:caroleheman2012@gmail.com)
- ADNP France
- Hope with amis de ADNP



- Ana González Hernández  
[ana@ctnnb1-foundation.org](mailto:ana@ctnnb1-foundation.org)
- Extra rare diseases
- **Extraordinarily united**



- Andrej Drdul  
[andrej.drdul@gmail.com](mailto:andrej.drdul@gmail.com)
- Slovak Association for Spina Bifida and/ or Hydrocephalus
- IF - International Federation for Spina Bifida and/ or Hydrocephalus
- We transform fear from the unknown for friendship and trust. Our belief is that every person with a disability and their families as well may live a full-fledged, joyful life.



- Dorica Dan  
[dorica.dan@eurordis.org](mailto:dorica.dan@eurordis.org)
- Prader Willi Association, Romanian National Alliance for Rare Diseases
- Living together, learning together!



- Benoit Fourcroy  
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- ASBH - [spina-bifida.org](http://spina-bifida.org)
  
- **Advances in healthcare and peer support**



- Erika Stariha  
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- SATB2 Europe
  
- Hope Becomes real when we come together



- Nina Knight  
[ninasknight@hotmail.co.uk](mailto:ninasknight@hotmail.co.uk)
- Acrodysostosis Support & Research UK World
- Together we accelerate the science and create a pathway to treatments





- Sue Routledge  
[sue@pitthopkins.org.uk](mailto:sue@pitthopkins.org.uk)
- Pitt Hopkins UK and Stichting
- Pitt-Hopkins syndrome
- Learning and going forward together



- Ioel Detton  
[assonoonan@gmail.com](mailto:assonoonan@gmail.com)
- Noonan France
- Alone, we go faster; together, we go further



- Vesna Vujičić  
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- 22q ex Yu, Serbia
  
- Together we can change the future



- Anne Lawlor  
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- 22q11 Ireland

- Together for Rare Families



- Antje Enekwe  
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- SLO Deutschland e.V, Smith-Lemli-Opitz Syndrome
- Let us make a difference for people with rare diseases.



- Olga Sophocleous  
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- Angelman syndrome Greece
- **Common vision for our rare patient's better futur**



- Tanja Zahlten  
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- German Bundesverband WBS
- Williams Beuren Syndrom
- Share knowledge and learn from each other



- Ines Fernandez Ulibarri  
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- Asociación Kleesfstra España
- Kleefstra syndrome
  
- Together to Change the Future of Rare





- Annalisa Scopinaro  
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- APW Italia
- Williams Syndrome
- Together we can



- Kelly Verbruggen  
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- 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



- Rui Barbosa Guedes  
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  - APMP Associação, Portugal
  - Phelan-McDermid
- 
- Beacon of Hope for brighter future



- Monica Bertoletti  
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- AIBWS Odv Italia, Association
- Beckwith-Wiedemann Syndrome
- Sharing, collaboration, better perspective



- Tanja Zdolšek Draksler  
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- IDefine Europe Foundation
- Kleefstra syndrome
- Data is the key



- Pogány Gábor  
[pogany@williams.org.hu](mailto: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)



- Jessica McAndrew  
[jessica@kabukisyndromefoundation.org](mailto:jessica@kabukisyndromefoundation.org)
- Kabuki Syndrome Foundation
- Building brighter futures for those affected by rare disease



- Marta Balula  
[marta.balula@raramente.org](mailto:marta.balula@raramente.org)
- Raramente, portugal
- Rare syndromes
  
- Empowering to advance Care, Knowledge, and Advocacy for Rare Lives



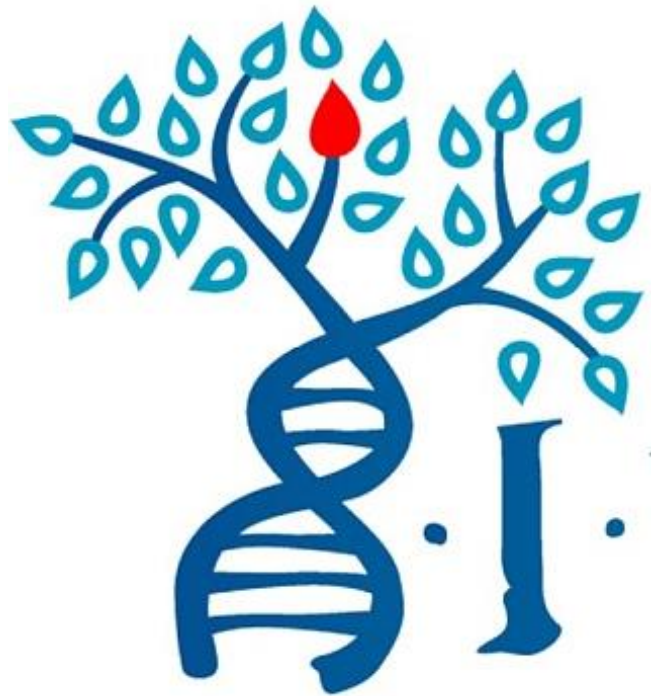


- Ammi Andersson  
[ammi.andersson@cordnode.se](mailto:ammi.andersson@cordnode.se)
- RBU Rörelsehindrade Barn och Ungdomar Stockholm
- Spina Bifida and Spinal Dyphrasism
- Cooperate with clinicians to create an equal specialist care for rare conditions in every country within EU



- Lyndsey Walsh  
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- Rare Diseases Ireland, 22q11  
Ireland and Ultra-Rare Diseases
- Together we are a strong  
partnership for change

Thank you for your attention !



UBUNTU

*"I am because we are."*

S·I·T·H·A·K·A