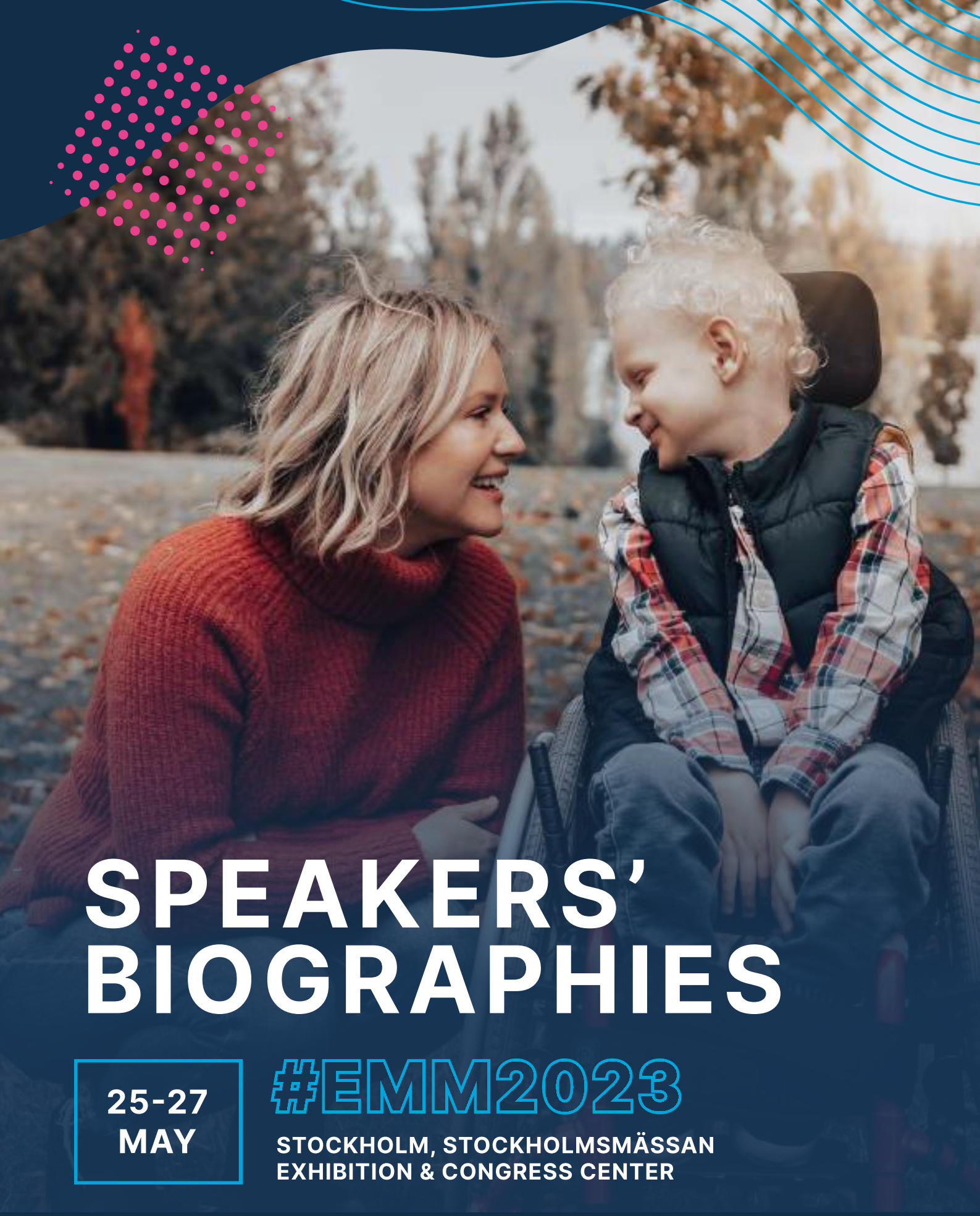

SPEAKERS' BIOGRAPHIES

25-27 MAY

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Anna Arellanesová

Anna Arellanesová is the chairperson of Rare Diseases Czech Republic, a Czech patient-led umbrella association for rare diseases. She is also a member of the board and former chairperson of the Czech Cystic Fibrosis Association. Being in the role of a vice-chair of the newly – established Patient Council of the Ministry of Health, where she represents rare diseases, she was able to participate in the creation of new legislation for an orphan drug reimbursement approval system, one which counts on the active participation of patients as decision makers.

She is also a member of the working group for rare diseases at the Czech Ministry of Health. Thanks to long-term cooperation with experts, she prepared and fulfilled the National Strategy for Rare Diseases as well as National Action Plans for Rare Diseases. She has a bachelor's degree in humanities from Charles University and lives in Prague.

Moderator: Living with a rare disease in childhood

Susanne Blichfeldt

Dr. Susanne Blichfeldt is a highly experienced and respected clinical professional with more than 30 years of experience in the diagnosis and clinical treatment of children with Prader-Willi Syndrome (PWS). She is also a trusted advisor on the treatment of adults with PWS.

Throughout her career, Dr Blichfield has been a dedicated researcher and educator in the field of PWS, presenting at conferences for the International Prader-Willi Syndrome Organization (IPWSO) and leading numerous educational sessions throughout Europe. Similarly, her Scandinavian study on the use of Growth Hormone (GH) resulted in the recommendation of GH treatment for children with PWS in Europe in 2000.

Dr. Blichfeldt is a co-founder of the Danish PWS association and has been a leader of the advisory board within the organization since 1986. She has been involved with IPWSO since 1991, co-organizing parent programs for several IPWSO congresses. She served as a board member from 2001 to 2004 and was a member of the PWS Professional and Scientific Board (PPSB) from 2010 to 2014. Dr. Blichfeldt also advises on the Clinical and Scientific Advisory Board (CSAB) and is a member of the FAMCARE.

Dr. Blichfeldt is also a devoted family member, with a husband, children, and grandchildren. Her 43-year-old son has PWS, which has given her a unique perspective on the syndrome and further fueled her passion for helping families affected by PWS.

Speaker: Enhancing Mental Wellbeing in the Rare Disease Community

Matt Bolz-Johnson

Matt Bolz-Johnson joined EURORDIS in 2014 as Healthcare and Research Director, later becoming Mental Health Lead & Healthcare Advisor. At EURORDIS Matt has led advocacy for the rare disease community by shaping the development and delivery of European Reference Networks (ERNs). Matt has ensured a patient centred approach is at the heart of ERNs, through developing European Patient Advocacy Groups formally linked to each ERN. Matt also leads the advocacy work on mental health and the development of a new EURORDIS Mental Health Partnership Network.

Prior to joining EURORDIS, Matt worked for 14 years in the National Health Service in England, working as a hospital manager in a mental health trust and as a health commissioner. He was a National Commissioner for highly specialised mental health services and ultra-rare diseases. Matt holds a Master of Art in Fine Art, at The Slade School of Fine Art, University College London and worked as a production designer before changing career in 1999, when he started working in an acute mental health unit in London.

Recently, Matt worked for Rare Diseases International as a Programme Director, leading the development of the Concept Model & Operational Framework for a new informal WHO hosted network – Global Network for Rare Diseases. Since arriving at EURORDIS, Matt has been working on two Joint Actions for Rare Diseases and leads on European Reference Networks. He developed the Partnership for Assessment of Clinical Excellence in the European Reference Networks (PACE-ERN) consortium and coordinated the technical proposal for the Assessment Manual & Technical Toolbox for ERN applications, which was the blueprint for all 24 ERNs. Matt was also a co-lead for EURORDIS Rare Impact II initiative, leading the research on the processes and criteria of selection of Centres of Expertise for ATMPs.

Moderator: Enhancing Mental Wellbeing in the Rare Disease Community. Moderator: How is my/my family member's Rare Disease impacting my mental health?

Guillaume Byk

Guillaume Byk is a legislative and policy officer at the European Commission, DG Health and Food Safety, Unit C1 on Digital Health. He is a lawyer specialised in data protection and biomedical law. Prior to joining DG SANTE, Guillaume worked at the office of the European Data Protection Supervisor and at the national data protection authority of Luxembourg on the implementation of the GDPR. Previously, he worked for more than 10 years as a data protection officer and legal counsel in a public biomedical research centre in Luxembourg.

Contributor: Training on digital safety and literacy in the context of the ongoing healthcare digitalisation

Raquel Castro

Raquel Castro rejoined EURORDIS in October 2022 as Social Policy and Initiatives Director. A member of the European and International Advocacy team, Raquel leads EURORDIS' advocacy, projects, and initiatives in the fields of integrated health and social care and of social policy, including disability, employment and non-discrimination rights. Raquel has previously worked with EURORDIS, from 2012 to 2021, coordinating its activities to advance holistic care for rare diseases and to support the integration of rare diseases into social policies.

As part of this work, she was involved in the EU-funded project INNOVCare and in the European Joint Actions for Rare Diseases, supporting the elaboration and the implementation of the "Commission Expert Group on Rare Diseases Recommendations to Support the Incorporation of Rare Diseases into Social Services and Policies". From 2018 onwards, she was also the Open Academy Director, overseeing the delivery of EURORDIS' training programmes. Besides her missions with EURORDIS, Raquel has worked as Senior Operations Manager at the European Federation for Spinal Muscular Atrophy – SMA Europe and has coordinated the Portuguese national help line for rare diseases.

Moderator: What barriers do we face while living with a rare disease and visible or invisible disability?

Moderator: Living with a rare disease in adolescence.

Avril Daly

Avril Daly has been President of EURORDIS-Rare Diseases Europe since November 2022. She was elected to the EURORDIS Board of Directors in 2009 and served as Vice-President of EURORDIS from 2012 to 2022. She is also CEO of Retina International, the global patient-led umbrella group, is dedicated to the promotion of research into Retinitis Pigmentosa (RP), Usher Syndrome, Macular Degeneration and allied retinal dystrophies. It acts as a voice for patients on policy issues and fosters cooperation among the medical and scientific communities.

Avril previously held the role of CEO at the Irish charity Fighting Blindness where she was responsible for raising awareness of retinal degenerative diseases among the general public, health care professionals and policy makers as well as the implementation of the development plan.

She is the chairperson of Rare Diseases Ireland, the Irish National Alliance for Rare Diseases, and in 2011 was appointed by the minister of health to the steering committee working towards the development of the Irish National Plan for Rare Diseases, published in 2014. Avril now sits on the oversight committee reviewing the implementation of that plan and on the working group of the Clinical Programme for Rare Diseases at the Irish Health Service Executive (HSE). Avril is co-chair of the National Vision Coalition in Ireland.

Avril represents Retina International at the European Patients Forum and was a founding board member of the Medical Research Charities Group (MRCG) and, the Irish Platform for Patients' Organisations, Science and Industry (IPPOSI). She was diagnosed with the rare retinal condition Retinitis Pigmentosa in 1998.

Speaker: Lifelong holistic approach and full inclusion in Society

Dorica Dan

Dorica Dan was appointed Vice-President in 2022. She was elected to the Board of Officers in 2011 and has been a member of the EURORDIS Board of Directors since 2007. Dorica is the mother of a daughter who was diagnosed with Prader Willi Syndrome at the age of 18. Dorica initiated RPWA (Romanian Prader Willi Association) in 2003, established RONARD (Romanian National Alliance for Rare Diseases) through a project funded by CEE Trust in 2007 and Romanian Rare Cancers Association in 2011.

In June 2011 she has opened the Pilot Reference Center for Rare Diseases “NoRo” through a project implemented in partnership with Frambu Norway and funded by Norway Grants. Today she is the chair of the Romanian Prader Willi Association, Romania (RPWA); president of the Romanian National Alliance for Rare Diseases(RONARD); president of the Romanian Association for Rare Cancers and the coordinator of the Centre for Information about Rare Genetic Diseases and NoRo Center. She is member of IPWSO (International Prader Willi Syndrome Organization) board, and is currently a EURORDIS EUROPLAN advisor.

Dorica represents EURORDIS at International Conferences throughout Europe and beyond, and is part of the Commission Expert Group on Rare Diseases. She is a member of the Programme Committee for the EURORDIS Conference on Rare Diseases and Orphan Products 2016 Edinburgh. Dorica is also part of the interim working group for the European Year for Rare Diseases. She is the Work Package leader of the EUCERD Joint Action Working for Rare Diseases (EJA) Work Package 6 focusing on Specialised Social Services, and is a member of the Council of National Alliances.

Panellist: Ageing with a rare disease

Laura de Graaff

Laura de Graaff (MD, PhD) is associate professor Internal Medicine for Rare Genetic Syndromes (RGS) and founder of the Erasmus MC Center for adults with RGS in Rotterdam, the Netherlands. As an internist-endocrinologist, she leads both clinical research and fundamental research lines investigating biomolecular pathways and cellular mechanisms involved in RGS. She finished her PhD in 2008, based on genetic studies in patients with congenital hypothalamic and pituitary disorders.

In 2015 she finished her medical training in Internal Medicine-Endocrinology and launched the Center for RGS. Its multidisciplinary team takes care of over 1100 adults with RGS from 19 national reference centres, including neurodevelopmental disorders; Rare Growth Disorders; Disorders of Sex Development and Prader-Willi (like) Syndrome. These expert centres are all part of ERN-ITHACA (Intellectual Disability and Congenital Malformations) or Endo-ERN (rare endocrine conditions). The Center for RGS aims to improve quality of healthcare for adults with RGS, combining specialised multidisciplinary care with innovative basic and clinical research.

Panellist: Ageing with a rare disease

Danielle Drachmann

Danielle Drachmann is a highly accomplished professional who has made significant contributions to the field of international health and rare disease policy. With a Bachelor's degree in Social Education and a Master's degree in Anthropology of Health, Danielle has a unique academic background that has informed her work as a patient advocate and researcher. After Danielle and her children were diagnosed with idiopathic ketotic hypoglycemia, she founded Ketotic Hypoglycemia International (KHI), a patient organization aimed at advancing research into the etiology, prevalence, and treatment of the disease.

Through KHI, Danielle facilitated patient-driven research and identify gaps in medical knowledge. Their work resulted in a shared publication in the international medical journal Orphanet Journal of Rare Diseases. Danielle's work with KHI led her to become a patient representative in the European Medicines Agency (EMA), where she helped draft health policy in Europe. She also served as a committee member at the European Health Parliament for the committee of "Telemedicine and Patient Centered Care" and as a "Young Citizen" in the Rare 2030 project in EURORDIS. In recognition of her contributions to patient advocacy, Danielle was awarded the Young Patient Advocate Award 2022 at the EURORDIS Black Pearl Awards.

Panellist: Lifelong holistic approach and full inclusion in society
Panellist: Living with a rare disease in Childhood

Jessie Dubief

Jessie Dubief joined EURORDIS and the Rare Barometer Programme in August 2019 to coordinate the H-CARE Pilot Survey on rare disease patients' experience. The aim of this pilot was to understand how to best operationalise a robust patient feedback mechanism across the 6000+ rare diseases and across the 24 European Reference Networks. Jessie is now a Rare Barometer Research Executive and develops the H-CARE project while participating in other Rare Barometer surveys and projects. She is specialised in multi-methodology and multilingual survey coordination, statistics for social sciences and in multi-stakeholders consultations, both in the health sector and in urban planning.

She started to develop this expertise while preparing for her PhD and teaching survey design and statistics at the University of Lyon (Lyon, Saint-Etienne), and while participating in research on past multi-methodology surveys.

Prior to working for EURORDIS, she worked in French urban planning agencies (multi-stakeholder NGOs in charge of urban planning and monitoring) where she directed and conducted studies using first-hand and second-hand data on well-being and health, social policies, housing, economy, mobility and land use.

Panellist: Living with a rare disease in Childhood

Katja Ekholm

Katja is a nurse, midwife and genetic counsellor working at the Karolinska Centre for Rare Diseases. She is also a PhD student in the Rare Diseases Group at Karolinska Institutet, Stockholm, Sweden. Her PhD studies focus on psychosocial aspects in parents of children diagnosed with cancer predisposition and/or rare congenital syndromes. At the Centre for Rare Diseases, Katja gives guidance to caregivers, patients and their next of kin. Among many other things she is involved in the work of developing common European minimal set of requirements for transition from child to adult care and has been part of Rare Diseases Sweden's transition project.

Panellist: Living with a rare disease in adolescence

Mette Grentoft

Mette Grentoft is a nurse anesthetist who has been working in the Danish health system since 1997. She has developed expertise in managing airways and ensuring patient comfort during surgical procedures. Grentoft is married and has three children, one of whom, her 23-year-old daughter, was born with Williams syndrome. n addition to her professional work, Grentoft has dedicated much of her spare time to volunteering for various associations.

She has previously served on the board of Rare Diseases Denmark and is currently the chairman of the European Federation for Williams Syndrome. Grentoft's mission in life is to advocate for people living with rare diseases and to help ensure that they have access to the best possible opportunities and resources to lead fulfilling lives on par with everyone else.

Panellist: Living with a rare disease in adolescence

continue

Karin Högvall

Karin Högvall works as a project manager at Rare Diseases Sweden. She joined the organization five years ago after working as an assistant nurse at a hospice. Due to her rare disease, she found it challenging to continue with her previous job. Karin's primary objective at Rare Diseases Sweden is to work towards improving and providing more equitable care for everyone living with a rare disease. She enjoys collaborating with her colleagues and meeting wonderful people along the way. Currently, Karin leads a project called "Sällsynt mitt I livet - Rare in the midst of life."

She finds this project fun and meaningful, and she is excited to share more about it during the EMM. During her free time, Karin loves spending time with her Icelandic horse, Krafla. She finds Krafla to be the sweetest soul, and the horse never fails to bring a smile to Karin's face.

Panellist: Adult life with a rare disease

Anne Hugon

Ms Anne Hugon has a background in Strategic Management of Health and Medical Services and a speciality in Documentary Resources Management and Databases. As Project Manager of ERN ITHACA (APHP Paris Hospital), she is involved in the general coordination and oversees all activities that involve interactions with patients and patient support groups/ePAGs. Anne is responsible for supporting the work of the two major “Share, care and cure” Work Packages dedicated to intellectual disability, neurodevelopmental disorders and malformative syndromes, and assists the WG Teaching and Training.

As a parent of a child living with GSD (Glycogen Storage Disease), the Founder President of AFG (Association Francophone des Glycogénoses - https://www.glycogenoses.org/), and a professional in the field of Rare Diseases, she is in a unique position to explore new perspectives and build relationships between HCP and governmental institutions. She is an ePAG volunteer with EURORDIS, advocating for patients’ rights and access to diagnostic and quality care for rare diseases, and solidarity and engagement are some of her core values.

Panellist: Ageing with a rare diseases

Kristine Jansone

Kristine joined the EHC in 2015, with ten years of work experience in the not-for-profit sector focusing on youth engagement and volunteering. She has a background in education, communication, and marketing. Kristine has an academic background in theology, pedagogy, and culture management. With the EHC Kristine's main focus has been the European Inhibitor Network that has transformed into the European Rare and Inhibitor Network in 2022, as well as the annual Leadership Conference and work with National Member Organisations with the focus on community development and organisational health. Kristine speaks Latvian, English, French and Russian.

Panellist: Ageing with a rare disease

Lene Jensen

For the past 10 years, Lene Jensen has been – and is - the director/CEO of the secretariat of Rare Diseases Denmark with app. 10 employees. Before that, Lene was a CEO at the Danish Haemophilia Society and even before that, a member oft he Danish „Folketinget" – the national parliament of Denmark. Lene holds an academic degree as master of science in economics and has for the past 10 years been engaged in various EURORDIS-activities as project leader, program committee member, moderator and more.

Lene has also been representing EURORDIS in various ways, counting the now closed EUCERD – European Commission Expert Group of Rare Diseases

Moderator: National actions which had an impact: What has worked in my country?

Kirsten Johnson

Kirsten Johnson is a member of the Board of The Fragile X Society (UK) and Chair of the Board of Fragile X International. Fragile X Syndrome is a rare disease, and carriers of the FMR1 gene can experience their own issues, Fragile X Premutation Associated Conditions. Kirsten is a carrier of the Fragile X FMR1 gene and has two daughters who live with Fragile X Syndrome. In 2022, Kirsten co-authored an article in Cells which called for the eradication of 'mental retardation' in the Fragile X gene and protein nomenclature.

As a result of this article, the HUGO Gene Nomenclature Committee, the international body which names genes, agreed to change the definition of FRAXA and FMR1. Further, UniProt, the protein database, has agreed to update the definition of the fragile x protein, FMRP. Removing this offensive terminology has been welcomed around the world, from scientists, clinicians and families. Building a more inclusive world for all our families affected by rare disease, so that they do not face stigma and discrimination, is key. In her day job, Kirsten is a pianist and composer. She has a doctorate in music and has released 24 discs of classical piano music on various labels.

Moderator: Lifelong holistic approach and full inclusion in society
Speaker: Enhancing Mental Well being in the Rare Disease Community

Kristian Emil Kristoffersen

Kristian Emil Kristoffersen has been CEO of Frambu Foundation since 2015. He is also a father of person with a rare disorder and has served as director of the board of the patient organisation representing her diagnosis. Kristoffersen is also an adjunct professor of Scandinavian linguistics at Nord University and was formerly professor of linguistics at the University of Oslo (1998 – 2015). His main research interest during these years has been speech and language disorders in children and adults. He has published extensively on speech and language in cri du chat-syndrome, a rare condition in which severe challenges with speech.

Language and communication are prominent. In his present position as professor at Nord university he is mainly involved in teaching and supervision of speech, language, and communication therapy students, in addition to doing research in this field.

Panellist: Living with a rare disease in childhood

Yann Le Cam

Yann was one of the founders of EURORDIS-Rare Diseases Europe in 1997. He is the organisation's Chief Executive Officer since 2000. Yann initiated Rare Diseases International (RDI) in 2009. He is an elected member of the RDI Council and Chair of the RDI Advocacy Committee. He is a founding member of the NGO Committee for Rare Diseases (United Nations, New York) in 2014 and its Vice-Chair. Yann is a Co-Chair of the Global Commission to End the Diagnostic Odyssey for Children with a Rare Disease since its launch in 2018. Yann is a member of the World Economic Forum's Health Stewards Board from 2020 and of its Global Precision Medicine Council since 2019.

Recent past positions include: member of the Management Board of the European Medicines Agency (EMA) 2017-2019; Chair of the Therapies Scientific Committee of the International Rare Diseases Research Consortium (IRDiRC), 2013-2017; Vice-Chairman of the EU Committee of Experts on Rare Diseases (EUCERD), 2011 – 2013; and a member of the Commission Expert Group on Rare Diseases, 2014 – 2017; member of the Committee for Orphan Medicinal Products (COMP) at the EMA, served 9 years, two elected mandate as Vice-Chair for 6 years. Yann holds an MBA from HEC Paris. He has three daughters, the eldest of whom is living with cystic fibrosis.

Moderator: What makes a difference to achieve lifelong integrated care

Åsa Lundin

Åsa Lundin is a registered nurse who has been working in the healthcare industry for almost three decades. She resides with her family in a small village called Berg outside Linköping, Sweden, and is married to Björn with a 13-year-old daughter named Stina. After beginning her career as a nurse in a ward with infectious diseases and psychiatry, she moved to Oslo, Norway, where she continued working in a ward with infectious diseases and at an intensive care unit. Upon returning to Linköping, she took a course to become an intensive care unit nurse and worked at a thoracic intensive care unit for fifteen years.

Since 2016, Åsa Lundin has been serving as a coordinator at the Centre of Rare Diseases, CSD southeast, which aims to provide equal opportunities for diagnosis, treatment, and services to people with rare diseases. Her primary responsibilities include coordinating the ward and other stakeholders for patients with rare diseases, increasing knowledge about rare diseases, and ensuring information exchange between wards and other stakeholders for patients with rare diseases. For example, she convenes Multidisciplinary Conferences and meetings with patients' healthcare providers to discuss their illness and troubles. Åsa also leads meetings to inform families, nursing staff, personal assistants, and school staff about specific rare diseases. She participated in a project with "The Swedish Confederation for rare diseases" about the transition from adolescence to adulthood and continued working on this topic. She designed a concept that provides a structured way of working with patients making the transition from adolescence to adulthood.

Panellist: Living with a rare disease in adolescence

continue

Jelena Malinina

Jelena Malinina joined EURORDIS in March 2022 as Patient Data Director. Based in the Brussels office, her role is to lead EURORDIS' work on data and represent rare disease patient voice in digitalisation of healthcare. Prior to joining, Jelena worked for the European Consumer Organisation, the World Health Organisation, healthcare consultancy, patient organisations and governmental public health organisation. Through her work she developed expertise in healthcare digitalisation, personal health data protection and security, artificial intelligence, medical devices, healthcare systems innovation and public health advocacy.

Jelena is a former member of the European Commission's ehealth Stakeholder Working Group and a former member of the European Medicines Agency Patient and Consumer Working Party. Jelena holds a Bachelor's Degree in Public Administration from the Mykolas Romeris University in Lithuania and a Master Degree in European Studies from the University of Gothenburg in Sweden.

Moderator: Training on digital safety and literacy in the context of the ongoing healthcare digitalisation

Fanni-Laura Mäntylä

Fanni-Laura Mäntylä is a Mental Health Activist, Patient Advocate and Mental Health and Substance Abuse Work Professional. In 2020, Mäntylä was Chair of the 5th Edition of European Health Parliament Committee on Mental Health and Healthy Workforce delivering the European Parliament an endorsed Policy Recommendation about implementing actions for a sustainable work life. More recently, Mäntylä was a member in Eurordis Rare 2030 Young Citizens network, and Patient Advisory Group in the IMI Project EU-PEARL.

Mäntylä is a type-1 diabetic and speaks openly about her personal experience of work related burnout and experience with severe depression. Though there is very little "rare" with the illness Mäntylä has lived experience with, it is not too often that somatic/physical and psychosocial wellbeing are brought forth with the effort Mäntylä brings. Mäntylä urges for more discussion about health in a holistic manner and calls for all stakeholders together on the agenda.

Panellist: Adult life with a rare disease

Maria Montefusco

Maria Montefusco has served as Vice-President of EURORDIS since November 2022. She is also the chairperson of Rare Diseases Sweden (2019–), a senior advisor for disability issues at the Nordic Welfare Centre, and the secretary at the Nordic Council of Ministers' disability council. With EURORDIS, she has sat on the board since 2020 and has been a member of its Social Policy Action Group (formerly Social Policy Advisory Group) since 2015. Previously, she was a part of the expert group at the Swedish Infocentre for Rare Diseases (University of Gothenburg), which reported to the National Board of Health and Welfare.

Maria has BAs in Political Science and Social Anthropology (Stockholm University), and a diploma in executive strategic project management from the Berghs School of Communication.

Speaker: Lifelong holistic approach and full inclusion in Society

Adéla Odrihocká

Adéla Odrihocká is a translator and interpreter specializing in healthcare, medicine and pharmaceutical area. Adéla became involved in patient advocacy after being diagnosed with a rare disease after 12 years of being undiagnosed and searching for a diagnosis. During her studies, she led workshops for academic staff members to help improve study conditions for students with chronic illnesses and to talk about accessible education in university settings. She has been an active Adéla ODRIHOCKÁ is a translator and interpreter specializing in healthcare, medicine, and pharmaceuticals area.

Adéla became involved more actively in patient advocacy during her university studies: she was involved in a working group on accessibility and disability in university settings and hosted workshops for academic staff members to help improve study conditions for students living with chronic conditions. She has been a member of ČAVO (Rare Diseases Czech Republic) for 6 years and is now co-chairing a patient organization for individuals living with Ehlers-Danlos syndrome and hypermobility syndrome. In recent years, she has been hosting many rare disease and disability-related workshops for diverse audiences (healthcare professionals, students, pharmaceutical companies, etc.) in the Czech Republic, France, and other European countries. She was invited as a speaker at the One World International Human Rights Documentary Film Festival to raise awareness about invisible disabilities and the social challenges individuals with invisible disabilities face. She has been part of the EURORDIS - 30 million reasons for European action on rare diseases campaign and was invited as a speaker and a young advocate to a panel discussion at the 11th European Conference on Rare Diseases & Orphan Products. She was a speaker at the Expert Conference on Rare Diseases organized in Prague within the Czech Presidency in the Council of the European Union. She participated in the EURORDIS Brussels Rare Disease Week earlier this year.

Panellist: Lifelong holistic approach and full inclusion in Society

Panellist: Adult life with a rare disease

Anders Olauson

Anders Olauson was involved in the founding of the Agrenska Centre in 1989. He served as director until 2004 and since then has been chairman. He is responsible for establishing the Agrenska Virtual International Academy, a research centre for rare disorders. In 2003, the Eesti Agrenska Foundation was inaugurated, and Anders has served as chairman of the Council since then. Anders is a past member of the board of EURORDIS, and was president from 1999 to 2001. During this period, EURORDIS was instrumental in the establishment of COMP (The Committee for Orphan Medicinal Products) at the European Medicine Agency.

Anders equally represented EURORDIS as a member of the board of the European Patients' Forum (EPF) until 2016. He was president of EPF from 2005 to 2016, when he was appointed Honorary President. Agrenska was, in 2005, appointed as a member of ECOSOC within the United Nations, with "special consultative status". Agrenska became a full member of CoNGO within the UN in 2014, and started, alongside EURORDIS, the NGO Committee for Rare Diseases (CfRD) in 2015. Anders has since served as Chairman for CfRD.

Between 2006 and 2012, Anders was a member of the advisory group for Health Research within DG Research at the European Commission. From 2013 to 2018, Anders was appointed member of the Horizon 2020 Advisory group for Societal Challenge - Configuration 'Health, demographic change and well-being'. In 2018, Anders was elected board member of CoNGO and became the chairman of the newly inaugurated RareResourceNet, a network for centres within the EU focussing on rare diagnoses.

The Swedish Government appointed Anders to serve as a member of the Advisory Council at The National Board of Health and Welfare from 2008 to 2014. Anders received HM the King of Sweden's Medal in 2010 and the EURORDIS Lifetime Achievement Award in 2017, to honour his valuable contributions to the field of disability and the rare disease community. He also received the City of Gothenburg's medal in 2014 for outstanding service for the city.

Speaker: Lifelong holistic approach and full inclusion in Society

continue

Jan Penfrat

Jan Penfrat is Senior Policy Advisor at European Digital Rights (EDRi) and leads EDRi's work on health data, IT security and big tech regulation. Before joining EDRi, Jan was a freelance technology reporter at the German IT magazine Golem.de and a co-founder of the Privacy Training Center. He holds Master's degrees from the College of Europe (Belgium) and Freiburg University (Germany) and started his career as a policy consultant for the lobbying firm EUTOP. Jan is also co-founder of the Belgian non-profit Privacy Training Center.

Contributor: Training on digital safety and literacy in the context of the ongoing healthcare digitalisation

Claas Röhl

Claas Röhl studied communication science at the University of Vienna after graduating from an engineering school in Vienna. He specialized in advertising and public relations and worked in film production and B2B-marketing. After the diagnosis of his daughter with Neurofibromatosis Type 1, a rare genetic tumor risk syndrome, he began his path into patient advocacy. He founded the Austrian patient organization NF Kinder in December 2013.

After completing several educational programs for patient representatives on a European level (such as the EUPATI patient expert training course, EURORDIS summer school & winterschool), and by continuously building a national and international network and investing in care and research infrastructures in Austria, NF Kinder managed to establish the first Austrian center of expertise for Neurofibromatosis in Austria by forming a partnership with the Medical University of Vienna.

On a European level Claas Röhl continued his ambition to create a better future for people living with Neurofibromatosis by co-founding NF Patients United, a European umbrella organization for NF patient organizations. • Patient Advocate Neurofibromatosis & Rare Diseases • President & Founder NF Kinder Austria • President & Co Founder NF Patients United • President & Founder EUPATI Austria • Vice-chair of Alliance of oncological patient organizations (AUSTRIA) • Chair ePAG group in ERN GENTURIS • Former member scientific committee IMI • Member of REiNS group • Registered patient expert for Neurofibromatosis at EMA • Member of the board Pro Rare Austria • EUPATI fellow • EURORDIS summer school & winter school graduate • ESO patient advocacy master class • WECAN - Evidence based patient advocacy training course.

Speaker: Enhancing Mental Well being in the Rare Disease Community

Lars Nesset Romundstad

Lars Nesset Romundstad has a Master of Science degree in Global Development from the Norwegian University of Life Sciences and is the deputy chair of the Norwegian Osteogenisis Imperfecta Association (NFOI). He has worked in Save the Children, The Norwegian Development Fund and in the United Nations Development Programme (UNDP). He is a graduate from the EURORDIS Young Patient Advocate programme of 2021 and has been a part of most Youth Events organized by Osteogenisis Imperfecta Federation Europe (OIFE) since 2015. Besides a strong interest in the international community, he also likes to ski, play tennis and travel.

Panellist: Living with a rare disease in adolescence

Laura Rosilio Bebeyto

Born in Alicante, in the sunny south-east coast of Spain, Laura has a university degree in English Translation by the University of Alicante, and a master's degree on International Relations by the Autonomous University of Barcelona, with a specialization in the European Union by the Diplomatic School of Barcelona. Laura focused her career on the intersectionality of human rights, advocating now for disability rights, youth rights, and women's rights, as being herself a young woman with Spina Bifida. After growing personally and professionally in the Third Sector, she is working now on disability policies at European level.

Since January 2023, she is the Chair of the Youth Advisory Group of IF – the International Federation of Spina Bifida and Hydrocephalus. This youth group works on the right to live independently while becoming adults with Spina Bifida and Hydrocephalus. The slogan 'Nothing about us without us' for her means not only to consult persons with disabilities in the pertinent political decisions, but for persons with disabilities to take part of the decision-making roles. To achieve that, Laura states that a holistic approach is needed to ensure that young persons with disabilities have the necessary resources and opportunities to be able to develop high-level professional careers.

Panellist: Adult life with a rare disease

Rebecca Tvedt Skarberg

Rebecca lives in Oslo, Norway. Her personal experience living with a rare condition comes from being born with osteogenesis imperfecta (OI). She is a trained social worker with additional degrees in psychology and counseling. She has previously worked for the Norwegian National Welfare office (NAV). From 2014 Rebecca has worked for the Norwegian National Advisory Unit on Rare Disorders (NKSD). In 2022 she was head of the program committee for the Rare Disease Day Norway. Rebecca started volunteering from an early age through the Norwegian OI Organization (NFOI) and The Norwegian Federation of Organizations of Disabled People (FFO)

Rebecca has also been engaged in international volunteer work through Osteogenesis Imperfecta Federation Europe (OIFE). OIFE is an umbrella association for 37 OI organizations. OIFEs vision is for children and adults with OI to live active lives – with access to competent healthcare and necessary social support. OIFEs mission is to connect and empower organizations, professionals and individuals to improve lives of people with OI. She was involved in the establishment of BOND ERN and was elected ePAG when ERNs were launched in 2016. Rebecca has been part of the SC in BOND and has contributed in many working groups in the ERN . She attended EURORDIS Leadership school of 2019. She has been a part of the establishment of the EuRR-Bone registry from 2020, especially working on patient involvement. In 2019-2021 she served on the Rare2030 panel of experts.

Moderator: Adult life with a rare disease

Cees Smit

Cees Smit (1951) is a long time patient advocate in the international patient community with multiple chronic conditions. In 2003, he received an honorary doctorate from the College of Deans of the University of Amsterdam.

He is (co-)author of several books, among them 'Aging with hemophilia: medical and psychosocial impact' (2007). In August, 2020 he published his autobiography 'Surviving hemophilia, a road trip through the world of healthcare'. He is also a painter.

Panellist: Lifelong holistic approach and full inclusion in Society
Panellist: Ageing with a rare disease

Cecilia Stocks

Cecilia has extensive experience working with children and adults with disabilities and their families. Cecilia is a social worker and psychotherapist and she has been with Ågrenska since 2003. Cecilia has a Master's degree in social work from the University of Gothenburg. Her specialist areas include parenting, rare diseases and neuropsychiatric disorders.

At Ågrenska, Cecilia is responsible for planning the specific parental modules during the family programmes. She also holds guidance sessions for school staff who meet children with rare diseases, as well as guidance meetings for parents in various parts of Sweden.

Cecilia plays a vital part in the shaping of Ågrenska's educational programme and frequently gives lectures on parenting and having a child with a rare disease. Cecilia is also responsible for planning the family programme for parents and siblings who has lost a child/sibling in a cancer disease.

Panellist: Living with a rare disease in Childhood

Jane Velkovski

Jane Velkovski is a 15 years old wheelchair user, having been diagnosed with Spinal Muscular Atrophy (SMA). He has been part of UNICEF disability campaigns in North Macedonia and a speaker at a high-level conference in Skopje and the United Nations General Assembly in New York for World Children's Day 2019. He was also part of a UEFA campaign to raise awareness about equality and access issues. His TED's speech "The life-changing power of assistive technologies" has millions of views where he evocatively explains why assistive technology should be available to anyone who needs it: "This chair is my legs -- this chair is my life [...] Freedom of movement, no matter on legs or on wheels, is a human right". He continuously gives speeches advocating for disability rights and unmet needs.

Using a wheelchair has not stopped Jane from playing football at every opportunity. "When playing football, I sometimes forget that I am in my electric wheelchair," Jane said. "I imagine myself scoring goals like Ronaldo, and sometimes I almost forget that I can't walk."

Panellist: Lifelong holistic approach and full inclusion in Society
Panellist: Living with a rare disease in adolescence

Geske Wehr

Geske Wehr was elected to the Board of Directors of EURORDIS in 2012 and currently serves as General Secretary. She has three children. Her eldest son suffers from an autosomal recessive congenital Ichthyosis. Geske has been involved in the German patient organisation Selbshilfe Ichtyose e.V. since 1997. In 2004, Geske started working towards founding a European Network of Ichthyosis (eni) for which she currently serves as Chief Executive on a voluntary basis.
Geske is the president of ACHSE e.V., the German National Alliance for Rare Diseases.

Moderator: Ageing with a rare disease

Maria Westerlund

Maria Westerlund is a 52-year-old social worker and ordained deacon in the Church of Sweden. She resides in the city of Umeå in northern Sweden and was recently elected as the chairperson of Rare disease Sweden. Maria enjoys her work as a social worker and loves meeting people. She finds that she learns something new every week, and this is the same situation when attending rare disease meetings. She is concerned about the growth of segregation and poverty in society and the unequal treatment of people, no matter their background.

Maria was born with the rare disease Klippel Feil syndrome and has experienced struggles with people's prejudice, low self-esteem, and poor knowledge about rare diseases in health care and society. However, she now looks upon many of her difficulties as a resource, despite wishing she never had to deal with them. Maria is also passionate about traveling and spent six months in Zimbabwe when she was a young nineteen-year-old. This experience, along with living with a rare disease, has greatly influenced her as a person and her view of life. She finds music, nature, and close friendships as what keeps her going.

Speaker: Lifelong holistic approach and full inclusion in Society

Kym Winter

Kym is the Clinical Director and Founder of Rareminds, a UK based non-profit organisation providing specialist psychotherapy, counselling and emotional wellbeing resources to rare disease communities. They also provide therapeutic group programmes, training and support for Patient Leaders with respect to the emotional impact of leading from lived experience, and work collaboratively to inform policy and practice at both the national and international level with respect to rare disease and mental health.

A psychotherapist for nearly 30 years across the NHS, higher education and UK third sector, she has worked with patients, families and healthcare professionals impacted by rare diseases since 2013. She also has lived experience of a rare genetic condition in her immediate family. Previously Head of Counselling at the University of Hertfordshire, her former roles also include Chair of a UK Psychotherapy Training Programme and Senior Therapist in a therapeutic community. She is a Consultant/Supervisor to several psychotherapy and mental health trainings, teams, and research studies, and has a long-standing interest in the impact of technology on relationships. She also has a passion for science fiction books and films!

Speaker: Enhancing Mental Well being in the Rare Disease Community