MEMBERS OF THE ADVOCACY COMMITTEE

1	Yann Le Cam	EURORDIS – Rare Diseases Europe
2	Paul Melmeyer	National Organization for Rare Disorders
3	Maureen Smith	Canadian Organization for Rare Disorders
4	Angela Chaves	Federación Colombiana de Enfermedades Raras
5	Migdalia Denis	Pulmonary Hypertension Latin Society
6	Ramaiah Muthyala	Indian Organization for Rare Diseases
7	Chiara Ciriminna	BLACKSWAN Foundation
8	Ritu Jain	DEBRA International
9	Kawaldip Sehmi	International Alliance of Patient Organisations –IAPO
10	Lara Bloom	Ehlers-Danlos Society
11	Simone Boselli	EURORDIS - Rare Diseases Europe
12	Sanja Peric	Rare Diseases Croatia
13	Kelly du Plessis	Rare Diseases South Africa
14	Mark Brooker	World Federation of Hemophilia
15	Lieven Bauwens	International Federation for Spina Bifida & Hydrocephalus
16	Alba Ancochea	Federación Española de Enfermedades Raras

RARE DISEASES INTERNATIONAL

ADVOCACY COMMITTEE

Yan Lee Cam
Eurordis
Chair

Maureen Smith
CORD-Canada

Angela. Chaves
FECOER-
Colombia

Kelly du Plessis
RDSA-South Africa

Ramaiah
Muthyala
IORD-India

Paul Melmeyer
NORD-EEUU

Ritu Jain
DEBRA
International

Lara Bloom
EDS-UK

Sanja Peric
RD-Croatia

Migdalia Denis
PHLS

Lieven Bauwens
IFSBH

Alba Ancochea
FEDER-Spain

Mark Brooker
WFH

Chiara
Ciriminna
BLACKSWAN
FOUNDATION

Kawaldip
Sehmi
IAPO

Simone Boselli
EURORDIS –
Rare Diseases
Europe

1. Yann Le Cam, EURORDIS – Rare Diseases Europe

Yann Le Cam is a patient advocate who has dedicated 25 years of professional and personal commitment to health and medical research non-governmental organisations in France, Europe and the United States in the fields of cancer, HIV/AIDS and rare diseases.

He has three daughters, the eldest of whom is living with cystic fibrosis. Yann is one of the founders of EURORDIS in 1997 and the organisation's Chief Executive Officer since 2001.

He has participated in the revision and adoption of European regulations that impact the lives of rare disease patients, including the EU Regulation on orphan medicinal products.

He was one of the first patient representatives appointed to the Committee for Orphan Medicinal Products (COMP) at the European Medicines Agency (EMA), where he served for 9 years and was its vice-chair for 6 years. He served on the Management Board and Executive Committee of the French HTA agency for 5 years, on the DIA Advisory Committee Europe for 3 years.

He was the Vice Chairman of the EU Committee of Experts on Rare Diseases (EUCERD) from 2011 to July 2013, and he is nominated on the current Commission Expert Group on Rare Diseases.

Yann Le Cam is also a member of and immediate past Chair of the Therapies Scientific Committee of IRDIRC (the International Rare Diseases Research Consortium).

In June 2016, Yann Le Cam was elected to the Management Board of the European Medicines Agency.

2. Paul Melmeyer, National Organization for Rare Disorders

Paul currently serves as the Director of Federal Policy at the National Organization for Rare Disorders. In this role, Paul leads the Federal policy operations in developing and advocating for the enactment and implementation of pro-rare disease patient policy. Paul also holds a Master of Public Policy (MPP) from the George Washington University. Paul is constantly seeking opportunities to make a difference in the lives of the less fortunate through compassionate and effective policy change

3. Maureen Smith, Canadian Organization for Rare Disorders

Maureen Smith's interest in patient advocacy stems from numerous years as a patient, subsequent to being diagnosed with a rare disorder at the age of eight. She has a long history of active collaboration with the health care community. In 2000, Maureen was the first layperson on the federal government's inaugural Inter-Agency Panel on Research Ethics (PRE) and served on the committee that was responsible for PRE's online research ethics tutorial. She was the public member of the National Placebo Initiative and chaired the Public Involvement Committee that conducted public consultations across the country.

Maureen has been a member of the Board of Directors of the Canadian Organization for Rare Disorders (CORD) since 2008 and its Secretary for the past 6 years. Maureen serves on a steering committee on patient collaboration in research for the Institut du savoir Montfort (ISM), affiliated with the University of Ottawa, and represents the ISM on the Council of Academic Hospital's Patient Engagement in Research Committee. She is a co-investigator on two CIHR multi-year pan-Canadian research projects on paediatric inherited metabolic diseases.

Maureen was appointed to Ontario's Committee to Evaluate Drugs as a patient member in 2014 and joined Health Quality Ontario's Health Technology Advisory Committee as a patient member in 2016. Maureen is a member of the Office of the Chief Health Innovation Strategist Consultation Group. She is a member of the US's Patient-Centred Outcomes Research Institute Advisory Panel on Rare Disease and a PCORI Ambassador. Maureen is honoured to be joining forces with patient advocates from around the world on Rare Disease International's Advocacy Committee.

4. Angela Chaves, Federación Colombiana de Enfermedades Raras

Born in Bogota, Colombia. Dentist and Professional in Information Sciences. CEO of Colombian Rare Disease National Alliance (FECOER) Member of the Board of ISPOR Colombia, leader of opinion and defender of the Rights of RD patients and families. Founding member and participant of the National RD Work Table in Colombia. Lecturer and Professor, she achieved concrete actions related to legislative and public policies in favor of the Rare Disease (RD) population in Colombia. Her life experience with Rare Diseases began with her daughter Lucia, thing that with her professional training, studies, and her social commitment, allow she to lead projects related to this topic. She is passionate for working with all the stakeholders to change the reality of those affected with RD in Latin America.

5. Migdalia Denis, Pulmonary Hypertension Latin Society

Honorary Founding President and Executive Director, Pulmonary Hypertension Latin Society

Independent senior consultant specialized in Strategic planning, evaluation, advocacy and communications. Speaker & Life Coach.

Strong background in Patient Advocacy/Government Affairs with over 10 years of experience in healthcare advocacy and policy. Active participant within Non-Profit and Pharmaceutical industry with a comprehensive perspective on healthcare issues and the needs of key stakeholders.

Highlights:

- Honorary Founding President and CEO of the SLHP. Member of the Board of Directors for the International Alliance of Patients Organization (IAPO). Member and representative of the Rare Diseases International (RDI) committee.
- Member of the Board of Trustees IAPO in Latin America,
- Member of the inaugural NGO committee for Rare Diseases at the United Nations Headquarters in New York, 2016.
- Guest member of the 55th Directing Council of PAHO, and the 68th Session of WHO Regional Committee for the Americas.
- Member of Working Group 13 on "Patient Perspectives" at the 2018 World Symposium on PH.
- Guess member of DESC spaces jointly with the office of the UN High Commissioner for Human Rights of patients promoting laws regarding rare diseases for the LatAm region.
- Advisor of the Health Committee in the Latin Parliament. Responsible for developing the legal framework related to Socio-Sanitary Care of rare diseases in LatAm.
- Co-author of chapter "Sociological Impact of Pulmonary Hypertension in Latin America" in the book "Una Enfermedad Oculta" (A Hidden Disease - first book about PH in LatAm).

6. Ramaiah Muthyala, Indian Organization for Rare Diseases

Ramaiah Muthyala, Ph.D. is the founder of Indian Organization for Rare Diseases, (IORD) (2003) and serves as its first President. It is an umbrella organization representing all rare diseases and patients in India. Its mission is to raise the awareness of rare diseases, advocate public policy and promote orphan drug development. Due to IORD efforts, the phrase - rare diseases is no longer foreign in India. He adopted a school (serving ~80 mentally retarded children and adolescents (boys and girls) in 2010. To advocating for the passing of orphan drug act in India a white paper "Rare Diseases and Orphan Products in India" (2014) was submitted to the Central Government of India.

He introduced an amendment to Indian Drugs & Cosmetic Act of 1954 to include "Orphan Drugs" (2017). Because of his efforts, State Governments (AP and Telangana) have adopted the surveillance of new-born melodies in primary health care centres. Ramaiah also serves as a committee member for establishing "Minnesota State Advisory council for Rare Diseases" and it is second of this kind in the United States.

Ramaiah serves as co-chair of Working Group on Chemically-Derived Products including Repurposing, Therapies scientific committee of IRDiRC. Also, Ramaiah was founding member of "In need of Diagnosis" (2006) whose mission is to assist patients who need of diagnosis of rare diseases. He has 25+ years of experience in orphan drug discovery and development in pharma as well as academic institutions.

7. Chiara Ciriminna, BLACKSWAN Foundation

Chiara is Project Coordinator & External Relations at BLACKSWAN Foundation. Her responsibilities include planning and implementation of the Foundation's program and management of different initiatives, including the RAREvolution Project that aims at encouraging the recognition of rare diseases as a public health and research priority and advocate for new and more effective international and national policies.

Before joining the BLACKSWAN Foundation, she has directed the activities of a Swiss organization supporting Mental Health in Africa. As representative of this non-governmental organization she has contributed to the discussions at the World Health Organization on the promotion of mental health policies and projects in low and middle-income countries. She has also served the United Nations Peacekeeping Operations in Ivory Coast (ONUCI) evaluating the electoral process and the political context of the region.

Chiara holds a Master degree in Law from the University of Florence and a post-master degree in International Affairs from the Institute of International Political Studies (ISPI) of Milan. She has also attended training courses in project management at HEI-Graduate Institute in Geneva.

8. Ritu Jain, DEBRA International

A representative of DEBRA International, Dr Ritu Jain has been an engaged member of the RDI council since 2017. She has actively contributed to drafting RDI's strategy plans, objectives and bylaws and is working towards strengthening and extending the Rare network in the Asian regions.

Ritu is also president of DEBRA Singapore, the Epidermolysis Bullosa (EB) patient advocacy and support organization that she helped establish in 2015. Within the DEBRA network, she serves as treasurer of DEBRA International and as regional ambassador for EB Without Borders, a Debra International workstream. In all these roles, her focus has been to offer emotional and material support to patients and families, organise local and regional conferences for capacity building and improved delivery of EB care, and advocate for policy changes. She remains committed to fostering collaboration of local and international clinicians/researchers as well as pharmaceutical organisations for enhanced research initiatives and clinical trials for improvement in the quality of lives of individuals living with EB. Ritu's familiarity with the culture, conditions, and challenges of those with rare diseases in Asia make her a suitable voice within Rare Diseases International for those with rare diseases.

Beyond her volunteer roles, Ritu is a sociolinguist and teaches graduate and under-graduate students at a local university. A PhD from the National University of Singapore, Ritu's research and publications explore the impact of language policies on languages of immigrant minorities in multilingual sites.

9. Kawaldip Sehmi, International Alliance of Patient Organisations –IAPO

Kawaldip previously held the position of CEO at Richmond Psychosocial Foundation International and worked as Managing Director of Coram Children's Legal Centre. He has European and international public health experience as Director of the Global Health Inequalities Programme and as Chairman of the European Network of Quitlines.

Kawaldip's qualifications include an MSc in the Public Health International Programme from the London School of Hygiene and Tropical Medicine, an MBA in Business Administration from the London Business School and Open University, and an LLB (Hons) from the London College of Law.

10. Lara Bloom, Ehlers-Danlos Society

Lara Bloom is the international Executive Director of The Ehlers-Danlos Society and responsible for globally raising awareness of The Ehlers-Danlos syndromes and hypermobility spectrum disorders (HSD). She manages coordinated medical collaboration, raising funds for research and focusing on the progression of EDS and HSD throughout the world. In Lara's previous role at EDS UK, in just five years she went from being the only member of staff to managing a team of six staff and 45 volunteers and increasing their membership, social media presence and income substantially.

Lara speaks at conferences all over the world, lectures to medical students and professionals, and supports specialists in the field by offering her experience as a leading patient expert in rare diseases.

From 2013–2015 Lara was part of the specialized rheumatology CRG (Clinical Reference Group) working with the NHS and she regularly works with umbrella organizations lobbying in parliament. She is a member of the Patient Empowerment Group for Rare Disease UK and in 2016 Lara graduated after completing expert-level training in medicines research and development and became a fellow of the European Patients Academy EUPATI.

Lara played a key role in the recent international effort to re-classify EDS and create management and care guidelines. She was a published author on the subsequent classification publication in the AJMG, March 2017 and serves on the steering committee of the International EDS Consortium. Lara lives in London with her wife and their dog Ripley.

11. Simone Boselli, EURORDIS - Rare Diseases Europe

Simone joined EURORDIS in April 2017 as a Public Affairs Director.

As a member of the European and International Advocacy team, Simone specifically represents EURORDIS in policy discussions on access to therapies, with a focus on existing delays and inequalities observed across Europe, on the underlying challenges in the field of the value assessment and pricing and reimbursement of orphan medicines, and on current efforts/initiatives towards improved access (e.g. MoCA). He will be supporting the follow up on the reflection paper 'Breaking the Access Deadlock to Leave No One Behind' and will also lead the multi-stakeholder negotiations to develop the 'One-text' for improving access to therapies for people with rare diseases.

With a view to advance rare diseases as a public health priority both at the global level, Simone will also support to the further development of NGO Committee for Rare Diseases, in close collaboration with CoNGO (the Conference of NGOs in Consultative Relationship with the UN) and the Agrenska Foundation.

Simone brings to EURORDIS over ten years of experience in the European public affairs arena, having previously worked for two leading consultancies in Brussels and specialised in health advocacy and government affairs in particular. He has in-depth expertise in healthcare having devised and implemented advocacy campaigns at EU and national level on a range of global health issues from sustainable health systems, chronic diseases, healthy ageing to mental health, neurodegenerative diseases, malaria, TB and hepatitis C.

Simone graduated in Sciences of Communications at the University of Bologna and holds a post-graduate certificate in European Public Relations and Project Management from the Italian Institute of Commerce in Brussels.

Simone speaks Italian, English and French, and has a working knowledge of Spanish.

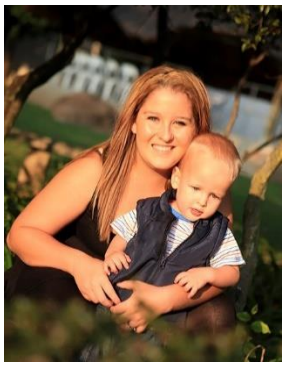
12. Sanja Peric, Rare Diseases Croatia

President of Rare Diseases Croatia, Sanja Perić has dedicated 28 years of active work to improve the life of Phenylketonuria patients in Croatia. She is a member of the Croatian PKU association since 1990, as in 1971 her brother was born with PKU. She started being very active in the association in 1995, when her daughter was born, also with PKU. From 1995 to 2007 she was a member of the Steering Committee, from 2007 to 2016 she was the president of the association and from 2016 she is a member of the Supervisory Board"

Her contribution in the field of rare diseases includes ensuring specific needs of patients are met within the health and social care systems, for example, helping provide medicine for specific diseases through government-funded health insurance. She is active in Rare Diseases Croatia since the beginning and has been a member of the Steering Committee. In July 2016, she became the president of Rare Diseases Croatia. Sanja is Croatian representative in ESPKU from 1990. and member of the commission for new born screening in Ministry of health.

Sanja Perić had the opportunity to collaborate with different institutions on national and international level in relation to rare diseases, in order to improve knowledge about cooperation between patients, doctors, state institutions and other stakeholders. She is also a member of Committee for Rare Diseases in Ministry of Health. During her work in both organizations, she had organised different activities: educational workshops (20), awareness educational workshops in schools (primary and high schools, 150), national/international camps (9), international/national/regional cooking classes (over 50), organized international conference (1), wrote cookbooks for PKU (2), developed educational materials for PKU also lead the campaign for Rare Disease Day and international PKU day. She contributed to raising awareness and developing the necessary response to health and social needs of people with rare diseases.

13. Kelly du Plessis, Rare Diseases South Africa



Kelly du Plessis, wife and mother of 2 children (aged 8 and 6) and CEO & Founder of Rare Diseases South Africa, a registered NPO born out of necessity when her oldest child was diagnosed with Pompe disease at 11 months old. Pompe disease is a rare, neuromuscular disorder which is fatal if left untreated. At the time, treatment for this rare condition was not available in South Africa, and so Kelly's personal journey of patient advocacy started.

Having dedicated her life and career to furthering the plight of those impacted by rare diseases in developing countries, Kelly serves on various boards and committees which focus on improving the quality of life for rare patients.

In 5 years, Kelly has taken Rare Disease policy and patient advocacy to new heights in South Africa, and has presented at various national and international conferences to raise awareness and create a new narrative in terms of treatment and access for rare patients. Understanding the isolation and lack of support surrounding a rare disease diagnosis, providing a safe place for patients as well as families, and improving patient-centred care has become her passion. With over 6500 patients/families impacted by rare diseases having been assisted over the past 5 years, Kelly has ensured that the patients voice is never forgotten.

Kelly was recently awarded the African Leadership Award for healthcare (Dec 2016 – Mauritius) with other recent achievements including the successful launch of RareX (the first Rare Disease conference in Africa), founding Africa-Rare.org (an African alliance for Rare Diseases), as well as the implementation of International Rare Disease Day in South Africa (Public awareness campaign reached over 17million South African's in 2014).

Future plans include further development of Africa-Rare.org across the continent as well as the development and implementation of a multidisciplinary centre of excellence for rare conditions.

Current Positions:

CEO/Founder – **Rare Diseases South Africa** (2013-current)

Founding Member – **Africa-Rare.org** (2015 – Current)

Board Member – **Genetic Alliance South** Africa (Vice – Chair 2014-2016) Previous board member at large, and provincial Chair)

Member of Patient Advocates Constituent - **International Rare Diseases Research Consortium (IRDiRC) –** October 2017 – current

Member of consortium assembly - **International Rare Diseases Research Consortium (IRDiRC) –** October 2017 – current

Member of patient advocacy committee – **Rare Diseases International** – October 2017 - current

Awards:

Finalist – WOS Woman of the year – Healthcare: 2017

Winner – African Leadership Award – Healthcare: 2016

14. Mark Brooker, World Federation of Hemophilia

Mark Brooker has worked for the World Federation of hemophilia since 2002. His current position is Senior Public Policy Officer in the Department of Research & Public Policy. His main areas of responsibility are access to treatment and data collection. For over 50 years, the World Federation of hemophilia (WFH) has provided global leadership to improve and sustain care for people with inherited bleeding disorders, including hemophilia, von Willebrand disease, rare factor deficiencies, and inherited platelet disorders. We save and improve lives by: training experts in the field to properly diagnose and manage patients; advocating for adequate supply of safe treatment products; and educating and empowering people with bleeding disorders to help them live healthier, longer and more productive lives. The World Federation of hemophilia (WFH), an international not-for-profit organization, was established in 1963. It is a global network of patient organizations in 134 countries and has official recognition from the World Health Organization.

Treatment Product Safety, Supply and Access

Through its Coagulation Product Safety, Supply and Access Committee, the WFH monitors developments in product safety and supply and takes proactive action as required. When important issues related to either safety or supply emerge, the CPSSAC posts informative, accurate, and useful information on our website as soon as possible. The WFH has advocated at the World Health Organization, U.S. Food and Drug Administration, the European Commission, European Medicines Agency, national governments, and in support of our members on safety and supply issues.

Data Collection and Advocacy

Every year, the WFH collects demographic and treatment-related data on people with hemophilia, von Willebrand disease, rare factor deficiencies, and inherited platelet disorders throughout the world. The 2015 Report includes data on more than 304,000 people with bleeding disorders in 111 countries. As in past years, the Global Survey Report provides analysis and results for a limited number of the survey questions asked each year. The WFH World Bleeding Disorders Registry (WBDR) is a web-based data entry system that provides a platform for a network of hemophilia treatment centres (HTC) to collect uniform and standardized data on people with hemophilia. This registry brings together privacy-protected patient data from countries around the world, storing it in one central location. The real-world data generated in the registry will be used to advance the understanding and care of people with hemophilia worldwide and support evidence-based advocacy initiatives.

15. Lieven Bauwens International Federation for Spina Bifida & Hydrocephalus

Lieven Bauwens joined the EURORDIS Board of Directors in 2017.

Lieven is the Secretary General of the International Federation for Spina Bifida and Hydrocephalus, and the chair of both Child-Help Belgium and Child-Help International, a charity dedicated to help children with SB/H in developing countries. In those capacities, he is a global advocate for the rights of persons with the disabilities while engaging actively in primary prevention of these impairments. He is a board member of the Food Fortification Initiative , of the International Disability Alliance , and a founding partner of the global alliance for Spina Bifida and Hydrocephalus, PUSH!, and Child-Help in Belgium, Germany, France and the Netherlands. A background in Architecture and Business Administration.

Lieven got involved in IF as brother of a young man with Spina Bifida and Hydrocephalus.

16. Alba Ancochea Federación Española de Enfermedades Raras

Alba Ancochea joined the EURORDIS Board of Directors in May 2017.

Alba has a decade of experience and commitment in implementing and planning policies, programs and socio-health projects in the field of rare diseases. Since 2009, she has been actively involved with the Spanish Federation of Rare Diseases (FEDER) and its Foundation, which gathers 338 patient organisations, and since 2013 is the organisation's CEO. She leads a team of more than 40 people who work to develop 25 specialised projects aimed at improving the quality of life of people living with a rare disease. FEDER has promoted the creation and activities of ALIBER, the Ibero-American Alliance of Rare Diseases.

Alba has completed undergraduate and masters studies in psychology, special needs teaching and management of NGOs.
Alba is a graduate of the EURORDIS Summer School and represents FEDER in Rare Diseases International and the EURORDIS Council of National Alliances. In addition she is a member of the Drug Information, Transparency and Access Task Force (DITA).