European Commission

2nd CONFERENCE ON

European Reference Networks

SPEAKERS' AND EXPERTS' BIOGRAPHIES

8-9 OCTOBER 2015, LISBON

GOVERNO DE PORTUGAL | MINISTÉRIO DA SAÚDE

Presidency of the Council of the European Union · GRAND DUCHY OF luxembourg

Health

Table of Content

Lydia MUTSCH – Minister of Health of Luxembourg

Paulo MACEDO – Minister of Health of Portugal

Xavier Prats MONNÉ – Director General of DG SANTE

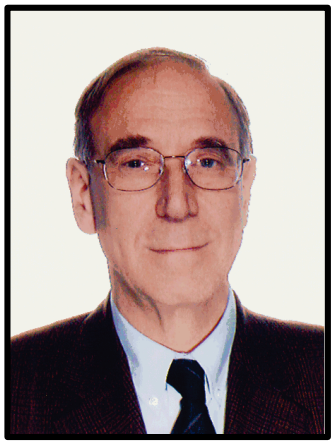
Xavier Prats Monné is the Director-General for Health and Food Safety of the European Commission since September 2015. He previously served as Director-General for education and culture, and as Director for employment policy. He holds degrees in Social Anthropology from the *Universidad Complutense* (Madrid, Spain); in Development Cooperation from the *International Centre for Advanced Mediterranean Agronomic Studies* (CIHEAM; Paris, France); and in European Studies from the *College of Europe* (Bruges, Belgium), where he graduated first of the Class of 1981-82 and served as assistant professor. He is from Spain and fluent in Spanish, English, French, Italian and Catala

Katharine BUSHBY – EUCRD Joint Action

Kate Bushby is a Clinical Academic Professor with joint appointments with Newcastle University and the Newcastle Upon Tyne Hospitals. She is a leader of the team at the John Walton Muscular Dystrophy Muscular Dystrophy Research Centre and MRC Centre for Neuromuscular Diseases based at the Institute of Genetic Medicine.

Working in the field since 1989, Kate's interests are focused on inherited muscular dystrophies from gene identification, disease characterisation and care standards to therapy development and delivery. Kate has a substantial grant portfolio from the EU, MRC and other funding organisations and over 200 publications. She has acted as PI on several industries and academic led clinical trials and natural history studies. This expertise in NMD has developed into a broader leadership role in Rare Disease Policy. Kate was a coordinator of the TREAT-NMD network and continues to work on its scientific secretariat, developing trial and therapy readiness in neuromuscular diseases. She led the EUCERD Joint Action on Rare Diseases and continues as policy coordinator on a new Rare Disease Joint Action 2015-2018. She is an NIHR Senior Investigator and 2015 recipient of the Eurordis Award for Research into Rare Diseases.

Charles BRUNEAU – PACE-ERN Consortium



Charles Bruneau is a specialist in Internal Medicine and Rheumatology. He joined the National Agency for Accreditation and Evaluation in Health (ANAES) in 1997 where he participated in the development of the French national accreditation program for healthcare organisations. From 2006 to 2013, he worked as a scientific advisor at Haute Autorité de Santé in France on the evolution of the accreditation process, on national strategies for patient safety and on the impact of quality improvement initiatives.

He has participated in European projects of DG Sanco (SIMPaTie, EuNetPaS) and of DG Research (MARQuIS and DUQuE).

He was a member of the Board of the International Society for Quality in Health Care (ISQua) and of the Council of the International Accreditation program of ISQua.

He is a member of the consortium of the PACE-ERN project representing the European Hospital and Healthcare Federation (HOPE).

Barbara BRUNMAIR – ExPO-r-Net pilot Network (rare cancers in children)

Barbara Brunmair is grant manager at the Research Support Office (RSO) of the Children´s Cancer Research Institute (CCRI) since 2010. She has specialised expertise in national and international Grants to support the investigators with the day-to-day project management, monitoring milestones and fulfilment of project deliverables, staff and facility administration, punctual scientific and financial reporting to granting agencies, and publication of grant-financed results. She is also engaged in supporting students and postdocs working at CCRI to develop their career development plans and to apply for a wide range of individual grants. Her profound scientific background in molecular biology and preclinical research as well as her experience in medical writing as an editorial journalist of one of the major Austrian publishers, Medizin Medien Austria, benefits her constant assistance to researchers and collaborators in generating high-quality proposals and grant acquisition. Since 2014 she is project manager of the multinational pilot network of cooperation ExPO-r-Net (European Expert Paediatric Oncology Reference Network for Diagnostics and Treatment).

Anne CALTEUX – Ministry of Health Luxembourg

Anne Calteux is Senior Policy Advisor to the Luxembourg Minister of Health in charge of EU Coordination and social security issues having an impact on public health since 2013. Prior to holding this position, she has followed EU initiatives in the field of public health, pharmaceuticals and social security at the Luxembourg Permanent Representation to the EU in Brussels for nearly 10 years. During this period, Anne Calteux has among others been actively involved in the negotiation of the cross-border healthcare Directive since the publication of the proposal in 2008 until its adoption in 2011 and thereafter as a member of the Cross-border Healthcare Committee. She has been a member of the Luxembourg Bar from 1999 until 2003 after having graduated in European and International Law in France and the UK.

Dr. Louise CLEMENT – Accreditation Europe Team leader PACE-ERN Consortium

Dr Louise Clement is the team leader for the PACE_ERN consortium, and represents Accreditation Europe in the consortia. She has lead a number of high profile clinical projects, most recently the implementation of a comprehensive stroke network in a national health system. Her areas of expertise are quality improvement,

physician engagement and enabling systems of care that promote and deliver patient-centered approach.

Louise holds a Medical Doctorate from Université de Montréal, a Bachelor of Science in Physiology from McGill University, and a Physician Leadership diploma from the University of Toronto. Louise has held a number of leadership positions managing multidisciplinary teams and working with a wide range of stakeholders, including patient organizations, health policymakers, clinicians, and health administrators. Many of her projects have involved continuous quality improvement activities that have brought sustainable changes to organization wide policies, procedures and guidelines to improve the care delivery process.

Louise has an in-depth knowledge of healthcare quality improvement and assessment. As a medical advisor and surveyor for Accreditation Canada International and Accreditation Europe, she actively contributed to the development and innovation to ACI's Qmentum International TM program. Louise is an active 'assessor' conducting surveys, evaluations and assessments of quality and patient safety at a variety of medical organisations.

Piotr CZAUDERNA – SIOPEL and Expo-R-Net member

Education:

- Graduated in the Medical University of Gdansk (1986)
- Training in pediatric surgery: Gdansk (Poland) + multiple foreign rotations (USA, Austria, Germany) – completed in 1994
- European Fellow of Pediatric Surgery (FEBPS) - 2002
- Obtained Master of Business and Administration (MBA) – 2014

Positions:

- Currently working full time as the consultant pediatric surgeon and Head of the Department of Surgery and Urology for Children and Adolescents, Medical University of Gdansk, Poland. Former Chairman of the SIOPEL (International Childhood Liver Tumors Strategy Group): 2006-2012.
- Currently member of the SIOPEL Core Committee. Presently also working as the national chair of the Polish Pediatric Liver Tumors Study Group and Surgical Coordinator of the Polish Group for Germ Cell Tumors in Children.
- Past member of the Executive Board of the International Society of Pediatric Oncologic Surgeons (2002-2006).
- President of the Polish Association of Pediatric Surgeons (2012-2014), currently member of the Board. Secretary of the Section and Board of Pediatric Surgery of the UEMS (2013-2015), currently President elect.
- Member of the SIOP (International Society of Pediatric Oncology) Scientific Committee. Member of the European Clinical Research Council in pediatric oncology

Degrees:

1. Medical University of Gdansk - MD - 1986

2. Doctor's dissertation (Ph.D. thesis): „Role of surgery in multidisciplinary treatment of selected solid tumors in children." - 1994.
3. Habilitation thesis: „Venoocclusive disease of the liver in the course of the therapy of Wilms' tumor – clinical and experimental research." - 2001.
4. Full academic Professor's title – 2013.

Silvia DEANDREA – Joint Research Center, European Commission

Silvia Deandrea obtained her degree of Medical Doctor and specialisation in Public Health and Preventive Medicine at University of Pavia, and obtained her Biostatistics PhD at University of Milano in 2011. Before joining the Joint Research Centre in 2012, she worked in healthcare quality consultancy for the Joint Commission International, in cancer epidemiology research at Mario Negri Institute of Pharmacological Research (Milano, Italy) and in population-based cancer screening programmes organisation and evaluation at Cancer Prevention Unit of Milano Local Health Authority. She is now coordinating the Quality Assurance scheme working group for the European Commission Initiative on Breast Cancer (ECIBC). Her current research interests include quality assessment and standardisation in breast and colorectal cancer screening, and Bayesian methods for evidence synthesis. She is author of more than 20 articles published in peer-reviewed international journals.

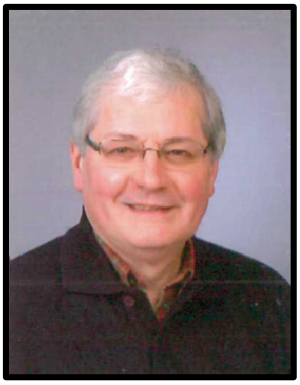
J. Alexandre DINIZ – Ministry of Health Portugal

J. Alexandre Diniz has a degree in Medicine by the School of Medicine of University of Coimbra and a master degree in Theology and Health Ethics by the Portuguese Catholic University. He has competences in Health Services Management by the Portuguese Medical Association.

He attended the Global Management Program for Senior Managers at École des Hautes Etudes Commerciales de Paris (HEC Paris) and undertook an internship in palliative care at the International Hospital of the University of Paris. He is qualified in Teamwork Dynamics and Public Health Policies within the framework of the Council of the European Union as well as in Committees and Comitology in the European Community Political Process by the European Institute of Public Administration. He has lecturing in several postgraduate courses in a number of Portuguese universities. He is a senior leader at the Portuguese Ministry of Health since 1986. He has participated in national health strategic papers and he has been the representative member of the Ministry of Health and of the Directorate-General of Health in several national and inter-ministerial committees as well as in several working groups and committees of the European Commission. Since 2009, as Director of the

Department of Quality in Health of the Portuguese Directorate-General of Health, he coordinates the planning of the national policy for quality in the health system, approved by the Portuguese Government. In 2012, he was awarded with the gold medal of the Portuguese Ministry of Health for distinguished service.

Patrice DOSQUET – Ministry of Health France



Patrice Dosquet is a medical doctor, medical specialist in Nephrology, with a master degree in Immunology. Since more than 20 years, he has been working in medical evaluation, clinical practice guidelines development, quality of care and certification of hospitals, in particular as a member of the French National Authority for Health. Since 2011, he has been working in the General Directorate for Health (DGS) in the French Ministry of Health (Ministry of Social Affairs, Health and Women's' Rights) as project leader for the 2nd French National Plan for Rare Diseases (2011- 2016) in close relation with all the stakeholders of the Plan. He is the French representative in the EC expert group for rare diseases.

Teresinha EVANGELISTA – EUCERD Joint Action, Newcastle University

Dr Teresinha Evangelista is a Consultant Neurologist with over 20 years of clinical experience, specialising in the fields of Neuromuscular Diseases & Neuropathology. During her 15 years at the Hospital de Santa Maria in Lisbon, Teresinha set-up a fully equipped Neuromuscular Laboratory and built an international reputation for Neuromuscular expertise (cemented in 2009 when she was named President of the Portuguese Society for The Study of Neuromuscular Diseases). Teresinha moved to the UK in 2012 to enhance her clinical and research expertise in the John Walton Muscular Dystrophy Research Centre (based in the Institute of Genetic Medicine, Newcastle University.) For the last three years Teresinha has been a core member of the specialised clinical team providing neuromuscular services to the North of England and delivering the UK's highly specialized service for genetically-determined muscle diseases. Teresinha also plays a key role in the research activities of the Centre, uniting clinical genetics and molecular biology via international Next Generation Sequencing projects. Teresinha's clinical expertise has informed her policy research role in the EUCERD Joint Action for Rare Diseases, 2012-2015: she will continue to provide expert rare disease support in the new Joint Action 'RD-Action'.

Pascale FLAMANT – UNICANCER

Pascale Flamant is a graduate of ENA (Ecole nationale d'administration, the French management school for high-level civil servants). Since 2011, she has been the General Delegate of UNICANCER, the Group of French Cancer Centres. These private non-profit health institutions are the leading edge in cancerology in France, and international key players. They are exclusively devoted to healthcare, research, education and training in oncology. Being General Delegate of UNICANCER, her responsibilities include the representation and upholding of the Cancer Centres' model as well as the development of strategies to enable them to stay ahead and innovate together for the benefit of patients. Previously, she worked as the General Director of the French National Cancer Institute.

Irene GLINOS – European Observatory on Health Systems and Policies

Paula GRECO – PACE-ERN Consortium

Victoria HEDLEY – EUCERD Joint Action

Victoria Hedley is a rare disease policy expert with experience and understanding of a wide range of topics across the rare disease 'spectrum'. As Thematic Coordinator in the new Joint Action for Rare Diseases, RD-Action, she is responsible for facilitating the translation of the needs and priorities of the field into meaningful policies at the European level whilst maximising impact at the national/regional level. From 2012-2015 Victoria project-managed the EUCERD Joint Action working for Rare Diseases, as the main point of contact in the Newcastle University Coordinating team. Over the last three years this role entailed building and maintaining relationships with stakeholders from Competent National Authorities, academia, patient organisations and Industry, to ensure the specificities and unique needs of those living and working with rare diseases are addressed in expert policy guidance. Since 2013 Victoria has supported the National Rare Disease conferences and provided guidance to optimise national activities for rare diseases. She has organised and delivered workshops on the European level with a particular emphasis on cross-border healthcare, particularly on the topics of European Reference Networks and Cross-Border Genetic Testing for Rare Diseases.

Michele HILTON-BOON – University of Glasgow

Michele Hilton Boon has ten years of experience as a programme manager and information scientist in the UK National Health Service, where her work included evidence review, guideline development, standards, quality indicators, and care pathways on a range of clinical topics, primarily cancer and blood-borne viruses, as well as participation in the RARE-Best practices project. She is currently part of the Informing Healthy Public Policy programme in the Medical Research Council/Chief Scientist Office Social and Public Health Sciences Unit (SPHSU) at the University of Glasgow. Her PhD research relates to systematic review of natural experiments to evaluate public health policy and interventions. She holds a Master of Public Health with Distinction from the University of Glasgow, Master of Library and Information Studies from Dalhousie University (Canada) and Master of Arts from Acadia University (Canada).

Reinhard HOLL – Network on Rare Diabetes in children

Reinhard W. Holl is a pediatric endocrinologist and diabetologist working at the University of Ulm, Germany, in the Institute of Epidemiology and medical Biometry. He is head of the working-group on computer-assisted quality monitoring in medicine and has a 20-year experience in standardized prospective documentation for patients with diabetes mellitus (DPV software), obesity (APV-software) as well as connatal hypothyroidism and congenital adrenal hyperplasia. He studied medicine at the universities of Ulm and Hannover, followed by internal medicine and pediatric training and sub-specialization in pediatric endocrinology/diabetology. After 25 years in inpatient and outpatient care at the Universities of Ulm and Gießen, he switched to patient-centered research as a primary focus. Main research interests are health care research in children with chronic conditions, medical quality management, multicenter observational studies using advanced methodology, focusing on the longterm course in chronic disorders (diabetes, obesity, endocrine). His research-group, including a mathematician, two statisticians, nutrition and physical activity experts as well as medical doctors and data experts, is part of the German Government-funded Competence Network Diabetes mellitus and the German Center for Diabetes Research (DZD). Data management for the SWEET initiative, aimed at European centers of reference for diabetes, is provided in Ulm, Germany. In addition to patient-centered research, Reinhard Holl sees pediatric patients with endocrine disorders, obesity or diabetes in a private practice. He is 57 years of age, married, and has a son. In 2013, he was awarded the ISPAD prize for innovation.

Matt JOHNSON – Partnership for Assessment of Clinical Excellence in European Reference Networks

Matt Johnson worked for 14 years in the National Health Service in England, in both hospital and commissioning trusts, where he was responsible for strategy development, clinical turnaround, quality assessment and contracting the full spectrum of healthcare.

He was a National Commissioner for ultra-rare conditions. He developed national clinical networks to improve equitable access, cost efficiency and clinical effectiveness, and to promote innovation for rare cancers, genetic conditions, rare paediatric conditions, transplants and highly specialised mental health conditions.

Since 2014, Matt has worked as the Healthcare and Research Director at EURORDIS. He works on the Joint Action for Rare Diseases and leads on European Reference Networks. He was responsible for developing Partnership for Assessment of Clinical Excellence in European Reference Networks (PACE-ERN) consortium and coordinating the technical proposal for the Assessment Manual & Technical Toolbox for European Reference Network application

Ruth LADENSTEIN – Expo-R-Net pilot network

Professor in Paediatrics and Senior Consultant in Paediatric Oncology.

- Head of the Clinical Trials Unit S2IRP (Studies & Statistics for Integrated Research and Projects) at the Children's Cancer Research Institute (CCRI) of the St. Kinderkrebsforschung e.V.

Recent project related activities are the coordination of the EU FP7 funded Network of Excellence: "EUROPEAN NETWORK for CANCER research in CHILDREN and ADOLESCENCE", the EU funded network: ExPO-r-NeT "European Expert Paediatric Oncology.

- Research Network for Diagnostics and Treatment" (http://www.expornet.eu/) and the Austrian Medicine for Children Research Network OKIDS (**http://www.okids-net.at/**).

- Board member SIOP EUROPE (http://www.siope.eu/);

- SIOPE president period September 2009 -October 2012 and chair of the SIOPE European Paediatric Research Council since 2012.

- Advisory board member of the SIOP Europe Neuroblastoma Group since May 2011

- SIOPEN president from May 2007 – May 2011;

- Principle Coordinating Investigator of SIOPEN High Risk Neuroblastoma Trials since 2002.

-Member of Oncology Advisory Board of the Ministry of Health Austria Chair of the Austrian Group for Paediatric Haematology-Oncology (AGPHO member - Arbeitsgruppe für Pädiatrische Haematology-Onkologie) www.docs4you.at

Denis LACOMBE – EORTC

Denis Lacombe graduated with his MD from the University of Marseilles (France) in 1988 and obtained a Master Post Doctoral Fellowship at The Roswell Park Cancer Institute (Buffalo, NY USA) for research in pharmacology and pharmacokinetics from 1989 to 1991. From 1991 to 1993, he worked as a Clinical Research Advisor in charge of the development of a new drug in oncology in the pharmaceutical industry.

Dr. Lacombe joined the EORTC in 1993 as a research fellow and quickly became a very active and productive Clinical Research Physician involved in the conduct of clinical research from protocol development through publication for a number of oncology indications from phase I to phase III. Dr Lacombe contributed to the strategic evolution of the EORTC pan-European clinical and translational research infrastructure by setting up various supportive assets such as regulatory and pharmacovigilance expertise as well as partnership models with the pharmaceutical industry. Dr. Lacombe rose to the position of Director EORTC Headquarters in 2010, and in April 2015 was appointed EORTC Director General. In his current position, Denis Lacombe is now involved in the coordination and administration of all EORTC activities in order to promote the EORTC as a major European organization in Cancer Clinical and Translational Research and is responsible for the organization of scientific activities, public relations and medium term strategies as defined by the EORTC Board as well as for internal and external communications. Dr. Lacombe is the author of well over 100 peer reviewed publications and communications that have had a positive impact on the future of cancer therapy.

Paola LARICCHIUTA – Istituto Superiore di Sanita, National Center for Rare Disease

Paola Laricchiuta is a researcher at the National Centre for Rare Diseases (CNMR) of the Italian National Institute of Health (ISS). She has a background in library and information science. In 2008 Paola joined the ISS, National Centre for Epidemiology, Surveillance and Health Promotion. She served on the Italian National Guideline System (http://www.snlg-iss.it) and participated in the development process of several guidelines with different roles in the guideline development group (supporting the activities of the coordination team, as a coordinator, as a member of the writing committee). Within the CNMR her work focuses on rare disease guideline development and promotion and on running courses on the development and evaluation of guidelines for rare diseases. She is involved in the EU FP7 project RARE-Best practices - A platform for sharing best practices for the management of rare diseases (www.rarebestpractices.eu) as a member the coordination team and task leader in the work package on dissemination.

Yann LE CAM – EURODIS patient organisation

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 has cystic fibrosis. Yann is one of the founders of EURORDIS in 1996-1997 and its Chief Executive Officer since 2001. He has participated in the revision and adoption of European regulations having an impact on rare disease patients' life, including the EU Regulation on Orphan Drugs, December 1999. He was one of the first patient representatives appointed to the Committee for Orphan Medicinal Products (COMP) at the European Drug Agency (EMA) where he served for 9 years and was its Vice Chairman 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 Experts Group on Rare Diseases. In November 2013, Yann Le Cam was elected Chair of the Therapies Scientific Committee of the IRDIRC – International Rare Diseases Research Consortium.

Suszy LESSOF – European Observatory on Health Systems and Polices

Suszy Lessof is the Director of Management of the European Observatory on Health Systems and Policies.

She is based in Brussels and is responsible for work planning, budget management, staffing, reporting to partners and supporting the implementation of policy and strategy. Prior to joining the Observatory Suszy worked on national standards in public health and in evaluation and had managed capacity-building initiatives in eastern Europe as well as the Health Services Management MSc at the London School of Hygiene & Tropical Medicine. Her background is in community economic development and urban regeneration, and she has an MBA from City University.

Henrique MARTINS – EXPAND project coordinator

Professor Martins is an Internal Medicine Specialist. He obtained his PhD degree from the Judge Business Scholl University of Cambridge with a thesis on 'The use of Mobile ICT in clinical Settings". He holds a Master in Management from the University of Cambridge and a Masters in HIV/AIDS from the University of Barcelona. He has several publications in the area of Mobile computing in healthcare and many conference presentations/keynotes in the area of eHealth. He worked as a CMIO-Chief Medical Information Officer at the Hospital Fernando Fonseca, a 730-bed hospital, between 2010 and 2013 where he set-up the new Electronic Health Record, and created the Center for Investigation and Creativity in Informatics (www.cu2.pt), where he supervised projects in robotics, mobile computing and database exploration and intelligent systems. From 2011 to 2013 he worked at the Ministry of Health as Adjunct for Health IT to the Health Secretary of State, and was responsible for the new Health Information Sharing Platform for Electronic Health informatics. In April 2013 he became president of the Shared Services of the Ministry of Health (SPMS), national agency responsible for medical products and IT Central Purchase body as well as ehealth and healthcare IT for the Portuguese National Health Service. He is the representative for Portugal at the European eHealth Network, was the National EpSOS project Coordinator until its end, present coordinator of EXPAND follow project. He publishes and teaches health management, leadership and medical informatics in Portugal and abroad.

Peter O'DONNELL – Healthcare Journalist at POLITICO

Peter O'Donnell is an EU-accredited Brussels-based journalist who has been covering European and international affairs for over twenty years. He has worked for The Financial Times, The Sunday Times, Reuters, the Economist Intelligence Unit, UPI, Euronews, and many other media in Europe and beyond. He is a former editor-in-chief of Europe Information Service, a former associate editor of European Voice, and is currently associate editor of Politico. In addition to his work as a journalist, he acts as editor, editorial adviser and speechwriter for numerous clients in the corporate, political and academic world. He frequently chairs EU-level policy debates, and lectures on EU affairs.

Willy PALM – Health Observatory of Health Systems

Willy Palm is the Dissemination development Officer of the European Observatory on Health Systems and Policies. He is responsible for organizing and leading policy dialogues with policy-makers in European countries on various health systems related issues. As a lawyer specialized in European social security law, he has been working mainly in the field of statutory health insurance and has been more specifically looking at the impact of EU integration on health systems. Before joining the Observatory in April 2006, Willy was the Managing Director of the International Association of Mutual health funds (AIM).

Samantha PARKER – Lysogene

Samantha Parker's expertise has evolved from her central role in the development and implementation of rare disease networks in the porphyrias, Wilson's disease pilot networks and most recently in the European network of intoxication type metabolic diseases (E-IMD and E-HOD). Ms Parker has an expert role at the European Commission on the EUCERD (now EGRD) and IRDiRC committees. Ms Parker represents the industry in her role on the expert boards and networks. She is employed by an SME biotech company Lysogene. Lysogene is developing AAV gene therapy in MPS IIIA and GM1 gangliosidosis

Jorge PENEDO – Ministry of Health Portugal

- General Surgeon consultant at Centro Hospitalar de Lisboa Central

- Competence in Health Services Management by the Portuguese Medical Association

- Deputy Clinical Director of the Centro Hospitalar de Lisboa Central

- Surgery Professor at Nova Medical School

- Senior Adviser of the Portuguese Health Minister

Member of the Board of member States for European Reference Network

- Member of the Portuguese Commission for Reference Centre

- Chief Editor of the "Revista Portuguesa de Cirurgia"

- Member of the National Ethic Committee to Clinical Investigation.

Karen RITCHIE – Healthcare Improvement Scotland

Karen originally studied cell biology and has a BSc from University of Glasgow and a PhD from University of Surrey. She worked on lab-based cancer research projects before completing a Masters in Public Health and developing a career in health services research. Following research posts at the University of Glasgow and the MRC Institute of Hearing Research, Karen joined the National Health Service in Scotland in 2002 to produce health technology assessments (HTA) of healthcare interventions. Karen now leads teams within Healthcare Improvement Scotland, the national healthcare quality improvement body, to provide research and knowledge management support for the development of HTAs, clinical guidelines and national clinical standards and indicators. The team also provides bespoke support to programmes for sharing knowledge and learning through use of a wide range of tools including social media. Karen holds an honorary research post at University of Glasgow and is responsible for the development and implementation of the HIS research strategy. Karen's research interests include the evaluation of complex improvement programmes and the provision of support for best practice in the management of rare disease through the RARE-Best Practices project.

Andrzej RYS – Health Systems and Products Director, Health and Food Safety DG, European Commission

- Medical doctor (radiology and public health) graduated from Jagiellonian University (JU) Krakow (PL).

- 1991 established School of Public Health at the JU. SPH's Director till 1997.

- 1997-99 Krakow's city Health Department Director. 1999-02 Health Deputy Minister (PL).

- Member of the Polish accession negotiators team. 2003 established and ran as Director the Center for Innovation and Technology Transfer at JU.

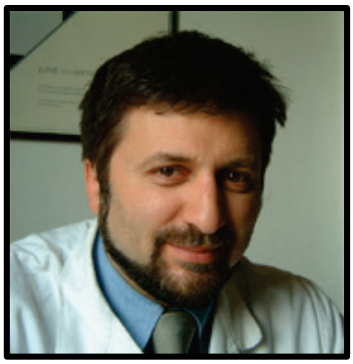
- 2006 joined the EC as Public Health and Risk Assessment Director in the Directorate-General for Health and Consumers in LUX.

- 2011 appointed Director for Health Systems and Products in the DG for Health and Consumers in BE.

Philippe RYVLIN – E-Pilepsy pilot Network

Philippe Ryvlin is Professor of Neurology specialized in epilepsy and more specifically in the management of patients undergoing epilepsy surgery. He has been working most of his career in Lyon where he is currently the Director of the Epilepsy Institute IDEE. He is also an affiliated Professor at the University of Copenhagen, Denmark, and chairs the Department of Clinical Neurosciences in Lausanne, Switzerland. Philippe Ryvlin has founded several European organisations dedicated to epilepsy, including the European Epilepsy Monitoring Unit association and European Network for Epilepsy Research. In 2014, he was appointed co-chair of the Joint Task Force of the International League Against Epilepsy (ILAE) and International Bureau for Epilepsy (IBE), an organization in charge of the communication between ILAE/IBE and the European Union and Commission. Based on his experience with European Networks in epilepsy and epilepsy surgery, he developed a pilot project of cooperation with 26 other partners in response to the 2013 call of the European Agency for Health and Consumers (EAHC). This project, E-PILEPSY, is one of the two pilot projects funded by EAHC within the framework of the upcoming European Reference Network.

Luca SANGIORGI – Coordinator of a network on Rare Skeletal Diseases



He is Head of Department of Medical Genetics and Coordinator of Rare Disease Centre at Rizzoli Orthopaedic Institute, Bologna, and Contract professor of Clinical Genetics of Bologna University. He is responsible of 3 National Registers of Rare Diseases (Li-Fraumeni, MHE and OI), a Member of National Coordination Team for Clinical Genetics Department, a Coordinator of many Regional Lab for Bioinformatics and manages the National and Regional Hub and Spoke Network on Skeletal Dysplasia. Contributor of more than 50 articles, he's an active Member of several international medical associations, serving on the Executive Committee of CTOS as President. Since 2013 he's been appointed as Italian government representative for the Assembly of Member States for BBMRI Italy in Europe and he's Member of National Node Coordination Office and coordinator of Rare Disease Interest Group. He's also Member of BBMRI-ERIC Steering Committee and Vice-Chair of Financial Committee. In 2014 he's been nominated as Italian representatives on "Committee for Advanced Therapies" of EMA by Italian Minister of Health. Expert Reviewer for the EU Commission for FP6 and FP7 grants, he's Coordinator of a European Project within ERA-NET on Translational Cancer Research (TRANSCAN). He's been invited speaker in national and international conferences.

Maurizio SCARPA – Expert in clinical trials in rare diseases

Prof. Maurizio Scarpa is the Director of the Centre for Rare Disease at the Helios Horst Schmidt Klinik (HSK) in Wiesbaden, Germany, and Professor of Pediatrics at the University of Padova, Italy. He received his medical degree, pediatric residency and doctorate from the University of Padova, He completed a postdoctoral fellowship at the European Molecular Biology Laboratory (EMBL) Heidelberg, Germany, on Molecular Biology and Gene Expression and at the Howard Hughes Medical Institute, Institute for Molecular Genetics, Baylor College of Medicine, Houston, TX, USA, on genetics and gene therapy. He has extensive expertise as a basic scientist in genetics, biotechnology, metabolic diseases and as a clinician in the diagnosis and treatment of paediatric rare disorders. Together with Prof. David Begley, Kings College London, UK, he is the Founder and President of the Brains for Brain Foundation (www.brains4brain.eu), a Paneuropean Research Foundation, grouping more than 60 international Universities and 10 Biotech Companies, collaborating to the development of therapies crossing the blood brain barrier and the understanding of basic mechanisms of rare pediatric neurodegenerative disease

Arimantas TAMAŠAUSKAS – Lithuanian University of Health Sciences

Professor and Chairman of the Department of Neurosurgery and Director of Neuroscience Institute of University of Health Sciences of Lithuania (National center of Neurosurgery), Chairman of Lithuanian Society of Skull Base Surgeons, Member of executive board of European skull base society, Member of UEMS section of Neurosurgery, Member of JRAAC (Joint residency advisory and accreditation committee) of UEMS and EANS (European association of Neurosurgical Societies). Invited lecturer in TATENA courses, European skull base society (ESBS), European Association of Neurosurgical Societies (EANS) conferences and courses. Evaluator of EC research projects of FP6, FP7, and Horizon 2020.

Adviser of the Minister of Health of Republic of Lithuania on public grounds (since 2014), Member of Advisory Board under the Ministry of Health of Republic of Lithuania (2015), Nominated Member (Lithuania) of ERN Board of MS.

Domenica TARUSCIO – Istituto Superiore di Sanita, National Center for Rare Disease

Domenica Taruscio, born in Cotronei (KR, Italy), is the Director of the National Centre for Rare Diseases at the National Institute of Health (Istituto Superiore di Sanità - ISS, Rome).

She performed her medical studies and specialization in histopathology at Bologna University; post-doctoral studies in human genetics at Yale University (CT-USA); master in bioethics (Roma University).

- Past or present member of the:

Committee for Orphan Medicinal Products - EMA (2000-2009); European Rare Diseases Task Force; EUCERD; European Commission of Expert Group on Rare Diseases; Health Research Advisory Group (DG-Research); Management Board of the European Molecular Genetics Quality Network; Interdisciplinary Scientific Committee of IRDiRC (www.irdirc.org); National Committee for the National Plan for Rare Diseases.

- Scientific leader of the bilateral agreement on rare diseases between ISS-Italy and NIH-USA (since 2003 up to now).

- Coordinator of the following EU projects: RARE-Best practices (www.rarebestpractices.eu); EUROPLAN (European Project for Rare Diseases National Plans Development, www.europlanproject.eu); EPIRARE (European Platform for Rare Disease Registries; www.epirare.eu); WP leader in the following EU projects: RD-Connect (http://rd-connect.eu), Advance-HTA, BURQ-OL, E-RARE, EUROCAT Joint Action; EUCERD Joint Action, WP co-leader in the RD-Action.

- Past President of ICORD (2010-2012, www.icord.se). Her efforts are directed mainly to tackle rare diseases from science to society.

Enrique TEROL – Dir D - Health systems and products, DG SANTE, European Commission

Enrique Terol works as National Expert in DG SANTE unit D2 European Commission and is in charge of the implementation of the European Reference Networks under the framework of the Directive of Cross-border Health care. He is MD, specialized in Family and Community Medicine, MSc and PhD in Public Health. His professional experience includes the clinical practice, managerial positions of Primary and Specialised Healthcare in private and public institutions and healthcare planning. He was Deputy General Director of Quality and Health Planning of the

Ministry of Health of Spain between 2004 and 208. He worked as Health Attaché in the Spanish Permanent Representation to the EU and coordinator of the area of Health in the Spanish Presidency of the EU between 2008 and 2011.

Dr. JLl. VIVES-CORRONS – ENERCA network on rare anameias

Dr. J.L. Vives-Corrons is Professor of Medicine, and Head of the Haematology Laboratory Department /Red Cell Pathology Unit of the Hospital Clinic of Barcelona (University of Barcelona) with a high input in the development of new diagnostic procedures, standardization and quality assessment of Haematology with pioneering activities in the development of an External Quality Assessment Scheme (EQAS) for haematology in Spain. Since 2002, Prof. JL Vives Corrons is the Head of the European Network for Rare and Congenital Anaemias (ENERCA) a Project co-financed by the European Commission (DG-SANCO) for the diagnosis, treatment and epidemiology surveillance of rare anaemias or rare diseases with anaemia as clinical key feature. This Project has been developed in four phases starting in 2002 and finishing in 2016.

Research activities are guaranteed by over 400 publications in scientific journals and haematology books as well as by the coordination of 38 different research projects, focused on; the study of physiopathology, molecular mechanisms and epidemiology of red blood cell (RBC) disorders due to hereditary enzymopathies, haemoglobinopathies, and membrane defects.

Till VOIGTLÄNDER – Co-chair Board of Member States, Clinical Institute of Neurology, Medical University Vienna

Till Voigtländer is associate professor of neurobiology and university lecturer of neurosciences at the Clinical Institute of Neurology, Medical University of Vienna. After studying medicine in Heidelberg, Germany, he received an intensive training in molecular biology, as well as neuropathological, neurochemical and neuroimmunological diagnostics, at different institutions in Heidelberg and Berlin (Germany), Zurich (Switzerland) and Vienna (Austria). Since his board certification as specialist in neurobiology, he leads a specialised clinical laboratory at his institute that focuses on the diagnosis of selected rare neurometabolic, neuroimmunological and neurodegenerative diseases. Beside his diagnostic responsibilities and due to his close link to the topic of rare diseases, he is also actively involved in recent developments in the area of rare diseases on a national, as well as a European level. In Austria, Dr. Voigtländer is country coordinator of Orphanet since 2004 and medical and

strategic head of the National Coordination Centre for Rare Diseases at the Austrian Healthcare Institute, established in 2011. In this context, he was one of the main participants in the elaboration process of the national plan of action for rare diseases, officially published on February 28 this year. At the European level, Dr. Voigtländer was and is the official representative of the Austrian Health Ministry in several European expert groups and committees including the European Commission Cross-Border Healthcare Expert Group and the current Commission Expert Group on Rare Diseases. Since December 2014, he is also member and elected co-chair of the Board of Member States on ERN of the European Commission.

Jaroslaw WALIGORA – DG SANTE

Jarosław Waligóra MD PhD is a policy officer on rare diseases in the Directorate General for Health and Food Safety, European Commission. Jaroslaw joined the Commission in 2006 and he has worked on several health policies: cancer, nutrition and physical activity. Currently he is responsible for rare diseases policy developments in the Commission.

Graduate of the Medical University of Warsaw, he worked the next 10 years at the University's 2nd Department of Paediatrics. Jaroslaw is a specialist in clinical genetics and paediatrics; in 2002 he obtained his PhD in human genetics.

Matthias WISMAR – European Observatory of Health Systems and Policies