



# IRDiRC

INTERNATIONAL  
**RARE DISEASES RESEARCH**  
CONSORTIUM

## **CONFERENCE 2013**

**Dublin, 16-17 April 2013**



IN ASSOCIATION WITH



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Programme









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RARE DISEASES RESEARCH  
CONSORTIUM

## CONFERENCE 2013

### Dublin, Tuesday 16 April

08:00-09:00	Welcome coffee and registration	
09:00-10:30	<p><b>Official Conference Opening</b>  <b>Patricia REILLY</b>, European Commission</p> <p><b>Welcome address</b>  <b>Ruxandra DRAGHIA-AKLI</b>, European Commission</p> <p><b>topic tbc</b>  <b>Christopher AUSTIN</b>, National Center for Advancing Translational Sciences, NIH, USA</p> <p><b>The Haystack is Made of Needles: A Global View of Rare Disease</b>  <b>Sharon TERRY</b>, Genetic Alliance, USA</p>	Plenary room
10:30-11:00	Coffee Break	
11:00-12:00	<p><b>Partnerships in Rare Diseases</b>  <b>Hans SCHIKAN</b>, Prosensa, the Netherlands</p> <p><b>Panel discussion:</b>  <b>International collaboration in rare disease research - opportunities and challenges</b>  <b>Christopher AUSTIN</b>, Director, National Center for Advancing Translational Sciences, NIH, US  <b>Sharon TERRY</b>, Genetic Alliance, USA  <b>Hans SCHIKAN</b>, Prosensa, the Netherlands  <b>Philip J. VICKERS</b>, Shire Human Genetic Therapies, USA  <b>Moderator: Ruxandra DRAGHIA-AKLI</b>, European Commission</p>	Plenary room
12:00-13:30	Lunch break	

13:30 - 15:30	<p><b>THERAPIES TRACK</b></p> <p><b>Deploying Existing Compounds: Repurposing Strategies</b> The session is devoted to the identification and development of new uses of existing or abandoned pharmacotherapies. The following topics will be discussed: Current experience and future trends on drug repurposing, Reviewing the US experience in drug-repositioning, Use of validated biomarkers for orphan development and marketing approval and Views of patients and academics.</p> <p><b>Chair: David PEARCE</b>, Sanford School of Medicine of the University of South Dakota, USA</p> <p><b>Speakers:</b> <b>Repositioning drugs for rare diseases</b> <b>Kevin LEE</b>, Pfizer, UK <b>NCATS/NIH Repurposing project</b> <b>Christine COLVIS</b>, National Center for Advancing Translational Sciences, NIH, USA <b>Experience from academic network on rare inherited kidney disease</b> <b>Franz SCHAEFER</b>, University of Heidelberg, Germany</p> <p><b>Panel discussion:</b> <b>Biomarkers discovery in rare diseases and implications for therapy</b> <b>Alessandra FERLINI</b>, University of Ferrara, Italy <b>Repurposing Nitisinone for Black Bone Disease: lessons from the EC-funded DevelopAKUre project</b> <b>Nick SIREAU</b>, AKU Society, Cambridge, UK</p>	Herbert room
13:30 - 15:30	<p><b>DIAGNOSTICS TRACK</b></p> <p><b>The Depth of Rare Diseases</b> There are estimated to be 7000 rare diseases. This session will review the number of rare diseases that remain unsolved, the impact this has on patient care, and will highlight the results of large-scale projects that are currently underway to identify the cause of these remaining 3500 rare diseases.</p> <p><b>Chair: Lu WANG</b>, National Human Genome Research Institute, NIH, USA</p> <p><b>Speakers:</b> <b>topic tbc</b> <b>Yann LE CAM</b>, EURORDIS, France <b>FORGE Canada: A nation-wide effort to identify genes for rare childhood disorders</b> <b>Kym BOYCOTT</b>, Children's Hospital of Eastern Ontario, University of Ottawa, Canada <b>A Large Scale Initiative to Identify the Genes Underlying Rare Mendelian Conditions</b> <b>Michael BAMSHAD</b>, University of Washington School of Medicine and Seattle Children's Hospital, USA <b>Deciphering Developmental Disorders (<a href="http://www.ddduk.org">www.ddduk.org</a>) - a UK wide project to transform the diagnosis of rare childhood disorders</b> <b>Helen FIRTH</b>, Cambridge University, UK</p>	Lansdowne room
13:30 - 15:30	<p><b>INTERDISCIPLINARY TRACK</b></p> <p><b>Building the Tools</b> This session is focusing on tools (genomics, registries, databases, trial networks, novel cell models). The lectures will focus on a particular success story, where significant breakthroughs have been achieved or are expected in a RD by using a research tool. This session should help researchers to get a better understanding of tools that they could use towards reaching IRDiRC objectives in their RD area.</p> <p><b>Chair: Ségolène AYMÉ</b>, INSERM, France</p> <p><b>Speakers:</b> <b>A mouse for every gene: Large-scale mouse model production and phenotyping for functional annotation of the genome for biology and drug discovery</b> <b>Colin MCKERLIE</b>, The Hospital for Sick Children, Toronto, Canada <b>Animal models in mitochondrial disorders</b> <b>Massimo ZEVIANI</b>, Istituto Neurologico "C. Besta", Milan, Italy &amp; MRC Mitochondrial Biology Unit, Cambridge, UK <b>Using pluripotent stem cells to model genetic disease of the heart</b> <b>Chris DENNING</b>, University of Nottingham, UK <b>The Rare Diseases Clinical Research Network as a Model Structure for Advancing Rare Diseases Research</b> <b>Jeffrey KRISCHER</b>, University of South Florida College of Medicine, USA</p>	Pembroke room
15:30-16:00	Coffee Break	

16:00 - 18:00	<p><b>THERAPIES TRACK</b></p> <p><b>Developing Innovative Orphan Therapies</b> This session is focusing on innovative drugs and advanced therapies. Specific topics to be discussed include:</p> <ul style="list-style-type: none"> <li>▪ The cell and tissue therapies: current experience and future perspectives</li> <li>▪ Developing in-vivo/ ex-vivo gene therapies: opportunities and challenges</li> <li>▪ Perspective of oligo-nucleotides based therapies</li> <li>▪ Patient needs and collaboration to foster clinical research in this area</li> </ul> <p><b>Chair: Ruxandra DRAGHIA-AKLI</b>, European Commission</p> <p><b>Speakers:</b> <b>Hematopoietic Stem Cell Gene Therapy for Rare Diseases: Current Experience and Future Perspectives</b> <b>Luigi NALDINI</b>, Institute for Gene Therapy Milano, San Raffaele Telethon Institute, Italy <b>AAV-mediated gene therapy for rare disorders: New possibilities for treatment of genetic disease</b> <b>Katherine HIGH</b>, University of Pennsylvania, USA <b>Antisense-mediated exon skipping – Applying lessons learnt from Duchenne muscular dystrophy to other rare diseases</b> <b>Annemieke AARTSMA-RUS</b>, University of Leiden, the Netherlands <b>Establishing multicenter international gene therapy trials for blood genetic diseases</b> <b>Anne Galy</b>, Genethon, France</p>	Herbert room
16:00 - 18:00	<p><b>DIAGNOSTICS TRACK</b></p> <p><b>Diagnosis of Rare Diseases</b> One of the goals of IRDiRC is to enable a diagnostic test for the majority of rare diseases by 2020. This session will focus on the development of best practices to ensure that genomic sequencing can deliver this reality in a clinical setting.</p> <p><b>Chair: Rafael DE ANDRES-MEDINA</b>, National Institute of Health Carlos III, Spain</p> <p><b>Speakers:</b> <b>Quality assurance and guidelines for validation of next-generation sequencing tools</b> <b>Gert MATTHIJS</b>, University Hospital Leuven, Belgium <b>Coupling sequencing and functional studies to improve outcomes in neonates</b> <b>Nico KATSANIS</b>, Duke University Medical Center, USA <b>Clinical Whole Exome Sequencing for the Evaluation of Genetic Disorders</b> <b>Christine ENG</b>, Baylor College of Medicine, USA <b>Complexity of NGS approaches in neurological disorders: New role of Medical Genetics in clinical guiding</b> <b>Olaf RIESS</b>, University of Tübingen, Germany</p>	Lansdowne room
16:00 - 18:00	<p><b>INTERDISCIPLINARY TRACK</b></p> <p><b>Ensuring the Collaboration</b> This session is focusing on collaborations (clinicians and scientists from academia, scientists from industry, patient organizations, regulators). The lectures will focus on a particular success story, where significant breakthroughs have been achieved or are expected in a RD through a collaborative mechanism. This session should help researchers to get a better understanding of collaborations that they could use towards reaching IRDiRC objectives in their RD area.</p> <p><b>Chair: Luc DOCHEZ</b>, Prosensa, The Netherlands</p> <p><b>Speakers:</b> <b>Changing outcomes in cystic fibrosis; the importance of working together</b> <b>Stuart ELBORN</b>, Queens University Belfast, UK <b>Rare Disease Partnerships: A Model for a New Era of Drug Development</b> <b>Cristina CSIMMA</b>, Cydan Development Inc., USA <b>Developing Rational Therapies for Inherited Neuropathies: A collaborative effort between clinical investigators, patient advocacy groups and industry</b> <b>Michael SHY</b>, Carver College of Medicine at the University of Iowa, USA <b>Telethon and Shire: a public-private partnership to translate the results of basic research to therapies for patients</b> <b>Philip J. VICKERS</b>, Shire Human Genetic Therapies, USA and Lucia MONACO, Fondazione Telethon, Italy</p>	Pembroke room
18:00 - 21:00	Poster session, cocktail and dinner	





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## CONFERENCE 2013

Dublin, Wednesday 17 April

08:00-09:00 Welcome coffee and registration

09:00 - 10:30 **THERAPIES TRACK**

### Regulatory Dialogue to Optimise Orphan Drug Development

Orphan Drug Designation has proved to be a powerful mechanism in US and EU by providing incentives and stimuli for the preclinical and clinical development of Orphan Drugs. However, the pathway for conducting clinical trials in is not free of hurdles. The following topics will be discussed:

- Hurdles and challenges in orphan drug development: what we have learnt?
- A continuing dialogue with regulatory agencies: perspectives from FDA and EMA
- Academic and Patients: role and participation in planning orphan drug development

**Chair:** Lucia MONACO, Fondazione Telethon, Italy

#### Speakers:

#### Elements to optimising Orphan Drug Development – industry perspective

**Wills HUGHES-WILSON**, EFPIA-EBE, Belgium

#### The EMA policy in fostering Orphan development

**Jordi LLINARES**, European Medicines Agency

#### The FDA policy in fostering Orphan development

**Katherine NEEDLEMAN**, U.S. Food and Drug Administration

#### Accelerating biotechnology innovation for rare diseases: challenges and solutions

**Emil KAKKIS**, ULTRAGENIX Pharmaceutical Inc Novato, USA

#### Panel discussion:

#### Canadian Policy in Fostering Orphan Development

**David K. LEE**, Health Products and Food Branch, Canada

#### Patients experience and views

**Avril DALY**, Eurordis, Genetic & Rare Disorders Organisation, Ireland

Herbert room

09:00 - 10:30	<p><b>DIAGNOSTICS TRACK</b></p> <p><b>The Human Phenome</b>          Interpretation of variation in the human genome in the context of disease is a challenging but essential component of patient care. To dramatically move this forward requires large-scale international collaboration. In this session we will discuss possible ways to assemble data and information in this domain and make it available for research and healthcare.</p> <p><b>Chair: Pierre MEULIEN</b>, Genome Canada, Canada</p> <p><b>Speakers:</b>  <b>The need for comprehensive, standardized phenotyping in the era of genome-wide sequencing</b>  <b>Ada HAMOSH</b>, Johns Hopkins University School of Medicine, USA  <b>An Ontological Foundation for Rare Disease Diagnostics and Novel Disease-Gene Discovery</b>  <b>Peter ROBINSON</b>, Charité – Universitätsmedizin Berlin, Germany  <b>An Ecosystem Approach to Connecting and Collecting Rare Disease Data</b>  <b>Anthony BROOKES</b>, University of Leicester, UK  <b>Translating Allelic Heterogeneity to Clinical Practice: The CFTR2 Project</b>  <b>Garry CUTTING</b>, Johns Hopkins University School of Medicine, USA</p> <p><b>Panel discussion:</b>  <b>The Human Phenome Project: Next Steps</b>          Speakers of the Session and <b>Milan MACEK</b>, Institute of Biology and Medical Genetics, Charles University Prague, the Czech Republic and <b>Ivo GUT</b>, National Genome Analysis Centre, Barcelona, Spain</p>	Lansdowne room
09:00 - 10:30	<p><b>INTERDISCIPLINARY TRACK</b></p> <p><b>Facing the Challenges</b>          This session focus on “challenges” that go beyond the “pure science”, but are relevant and interesting for scientists. It will debate some of the most important ethical, economic and political questions that are directly related to IRDiRC research including incidental findings and economic impact of RD research. The way these challenges are addressed will, at least in part, determine whether the ambitious IRDiRC objectives can be reached and implemented. Session is jointly programmed with Eurordis.</p> <p><b>Chair: Tarun WEERAMANTHRI</b>, Department of Health, Government of Western Australia</p> <p><b>Speakers:</b>  <b>Delivering on the promise: the clinical application of new diagnoses and treatments for rare diseases</b>  <b>Kate BUSHBY</b>, Newcastle University, UK  <b>“Big” Pharma: From Data Hoarding to Data Sharing?</b>  <b>Bartha KNOPPERS</b>, McGill University, Montreal, Canada  <b>The patients’ perspective</b>  <b>Maria MAVRIS</b>, EURORDIS, France</p> <p><b>Debate:</b>  <b>What to do with incidental findings from next-generation-sequencing: disclose or not disclose?</b>  <b>Jack GOLDBLATT</b>, Genetic Services and the Familial Cancer Programme of Western Australia and <b>Alastair KENT</b>, Genetic Alliance UK</p>	Pembroke room
10:30 - 11:00	<p><b>Coffee Break</b></p>	
11:00 - 12:00	<p><b>THERAPIES TRACK</b>  <i>session continued</i></p> <p><b>DIAGNOSTICS TRACK</b>  <i>session continued</i></p> <p><b>INTERDISCIPLINARY TRACK</b>  <i>session continued</i></p>	

12:00 - 13:30	Lunch break
13:30 - 16:30	<p data-bbox="280 338 644 367"><b>FORWARD LOOK SESSION</b></p> <p data-bbox="280 383 1382 472">This session will illustrate real success stories that have emerged from molecular characterisation of rare disease genes - stories where information about the gene led to an effective therapy for patients.</p> <p data-bbox="280 510 1401 539"><b>Chair: Paul LASKO</b>, Institute of Genetics of the Canadian Institutes for Health Research, Canada</p> <p data-bbox="280 555 408 584"><b>Speakers:</b></p> <p data-bbox="280 591 1366 645"><b>Inherited disorders of neuromuscular transmission – from gene discovery to tailored treatments</b></p> <p data-bbox="280 651 1027 680"><b>Hanns LOCHMUELLER</b>, University of Newcastle upon Tyne, UK</p> <p data-bbox="280 687 1385 741"><b>Newfoundland's Sudden Death Riddle Solved...How a Successful Gene Hunt is Saving Lives, Changing Practices and Influencing Policies</b></p> <p data-bbox="280 748 1315 777"><b>Terry-Lynn YOUNG</b>, Memorial University, St. John's, Newfoundland and Labrador, Canada</p> <p data-bbox="280 784 1378 837"><b>Alternating Hemiplegia of Childhood: the journey to the gene discovery and effective treatments</b></p> <p data-bbox="280 844 1270 873"><b>Tsveta SCHYNS</b>, European Network for Research in Alternating Hemiplegia (ENRAH)</p> <p data-bbox="280 880 1299 934"><b>Children with rare diseases of the immune system: from therapeutic orphans to pioneers of personalized medicine</b></p> <p data-bbox="280 940 1043 969"><b>Christoph KLEIN</b>, Ludwig Maximilian University, Munich, Germany</p> <p data-bbox="280 976 1219 1005"><b>Unravelling the jargon- c.402_405CCAA p(Ans135Val*fs): What the blip?</b></p> <p data-bbox="280 1012 746 1041"><b>Lesley MURPHY</b>, Rare Voices Australia</p> <p data-bbox="280 1048 1254 1102"><b>Ten years of translational research in rare progeroid diseases – lessons and perspectives for rare diseases</b></p> <p data-bbox="280 1108 970 1137"><b>Nicolas LEVY</b>, INSERM Research Centre Marseille, France</p> <p data-bbox="280 1144 1401 1198"><b>Unexpected genetic architectures underlying rare blood diseases: lessons learned from the BRIDGE project</b></p> <p data-bbox="280 1205 868 1234"><b>Christel VAN GEET</b>, University of Leuven, Belgium</p>
16:30	End of conference coffee







**Dr. Annemieke Aartsma-Rus** played an important role in the development of the antisense mediated exon skipping therapy for Duchenne muscular dystrophy during her PhD research (2000-2004), at the Leiden University Medical Center, Department of Human Genetics (the Netherlands). Since September 2004 she continued this research as a post doc. As of December 2007 she became leader of the “DMD exon skip group” first, as assistant professor and currently as associate professor at the Department of Human Genetics. In 2013 she did a visiting professorship at the Institute of Genetic Medicine of Newcastle University (UK).



Thus far, she has published over 60 peer-reviewed papers and 9 book chapters, as well as 11 patents and has edited one book. She has given many invited lectures at meetings, symposiums and workshops as well as patient/parent organizations meetings. She has created and maintains a website on exon skipping, containing a dedicated section for patients and parents as well as patient-friendly pages on therapeutic approaches for the TREAT-NMD website. In 2011 she received the Duchenne Award from the Dutch Duchenne Parent Project in recognition of this work and her dedication to the Duchenne field.

She has successfully applied for numerous grant applications, including a prestigious VIDI award (€800,000) from the Dutch government in 2009. She is chair of the COST Action “Networking towards clinical application of antisense-mediated exon skipping for rare diseases”, vice chair of the executive board of the TREAT-NMD alliance and member of the board of directors of the Oligonucleotide Therapeutics Society.

**Christopher Austin** is Director of the National Center for Advancing Translational Sciences (NCATS) at the U.S. National Institutes of Health. NCATS' mission is to catalyze the generation of innovative methods and technologies that will enhance the development, testing, and implementation of diagnostics and therapeutics across a wide range of human diseases and conditions. Before becoming NCATS Director in September 2012, he was director of the NCATS Division of Preclinical Innovation, which focuses on translating basic science discoveries into new treatments, particularly for rare and neglected diseases, and developing new technologies and paradigms to improve the efficiency of therapeutic and diagnostic development. In this role, he founded and directed numerous initiatives including the NIH Chemical Genomics Center (NCGC), the Therapeutics for Rare and Neglected Diseases (TRND) program, and the Toxicology in the 21st Century (Tox21) program. Before joining NIH in 2002, Dr. Austin directed research programs genomics-based target discovery, pharmacogenomics, and neuropsychiatric drug development at Merck, with a particular focus on schizophrenia. Dr. Austin received his A.B. in biology from Princeton and M.D. from Harvard Medical School. He completed clinical training in internal medicine and neurology at the Massachusetts General Hospital, and a fellowship in genetics at Harvard.



**Ségolène Aymé** is a medical geneticist and Emeritus Director of Research at the French Institute of Health and Medical Research (INSERM). She was the founder of Orphanet in 1997 ([www.orpha.net](http://www.orpha.net)) and its Executive Manager from 1997 up to 2011. Orphanet is the webportal dedicated to rare diseases and orphan drugs which is currently funded by the French Ministry of Health, the INSERM and the European Commission as a Joint Action (DG Public Health). She chairs the European Union Committee of Experts on Rare Diseases ([www.eucerd.eu](http://www.eucerd.eu)) and the WHO Topic Advisory Group for Rare Diseases. She serves as Editor-in-Chief of the Orphanet Journal of Rare Diseases ([www.ojrd.com](http://www.ojrd.com)). She is the project leader of "Support IRDiRC", which provides the services of a scientific secretariat to the International Rare Diseases Research Consortium ([www.irdirc.org](http://www.irdirc.org)).



**Michael Bamshad** is Professor and Chief of Genetic Medicine in the Department of Pediatrics and Director of the Center for Clinical Genomics at the University of Washington School of Medicine and Seattle Children's Hospital. Dr. Bamshad's research interests are focused on understanding how evolutionary processes and demographic history have shaped patterns of genetic variation among humans, and how such variation influences differences in physical features and disease susceptibility among humans. Currently active projects are investigating the distribution of genetic variation among human populations, developing novel strategies to find disease susceptibility variants based on exome and whole genome sequencing, and characterizing genetic variants influencing an assortment of health-related conditions including both Mendelian disorders and complex chronic diseases of childhood. He also co-authors a popular textbook entitled "Medical Genetics."



**Kym Boycott** is a Neurogeneticist at the Children's Hospital of Eastern Ontario (CHEO) and Investigator at the CHEO Research Institute. She is an Associate Professor and holds a Faculty of Medicine Research Chair in Neurogenetics at the University of Ottawa. She completed her PhD, MD and FRCPC training in Medical Genetics at the University of Calgary. She is the recipient of the Canadian Institutes of Health Research Clinical Investigatorship Award from the Institute of Genetics, the SickKids Foundation Young Investigator Award and the Basil O'Connor March of Dimes Young Investigator Award. Her current research is focused on the identification of genes and molecular pathways for rare disorders and she is the Lead Investigator of the Genome Canada, CIHR funded 'Finding of Rare Disease Genes in Canada' (FORGE Canada) initiative.



Professor **Anthony Brookes** is an expert in genomics and bioinformatics, having contributed to research progress in disease and population genetics, DNA variation analysis, and data management/exploitation systems for biomedical information. He has published over 150 peer reviewed articles and reviews. He has served two 3-year terms on the HUGO Council, co-founded the Human Genome Variation Society and the Human Variome Project, leads an international meeting series on genome variation (HGV200X), and jointly designed the standard Observ-OM data model for gene-disease relationships. In his current position as Professor of Bioinformatics and Genomics at the world-renowned Genetics department (University of Leicester, UK) he continues to focus on 'knowledge engineering' to bridge the divide between research and healthcare. This is reflected, not least, by running the world's largest 'GWAS Central' database ([www.gwascentral.org](http://www.gwascentral.org)), and leadership roles in the EC-FP7 projects GEN2PHEN ([www.gen2phen.org](http://www.gen2phen.org)) and BioSHaRE ([www.p3g.org/bioshare](http://www.p3g.org/bioshare)).



Prof. **Kate Bushby** : MD is a Professor of Neuromuscular Genetics, is coordinator of the EUCERD Joint Action on Rare Diseases, and is former joint coordinator of the FP6 TREAT-NMD Network of Excellence; she is also Deputy Director of the MRC Centre for Neuromuscular Diseases and she has over 100 publications.

Kate Bushby is a clinical academic with joint appointments between Newcastle University and the NHS. She is a member of the Neuromuscular Research Group within the Institute of Genetic Medicine and plays a leading role in the National Commissioning Group (NCG) for rare neuromuscular diseases.

Professor Bushby has a long-standing interest in the molecular genetics of the limb-girdle muscular dystrophies and related disorders and is interested in the best possible development and implementation of care guidelines as well as clinical trials. Her team has developed an extensive programme of research in NMD from basic molecular pathology to clinical studies.



**Christine Colvis** joined NCATS in June 2012 to lead the “NIH-Industry Pilot Program: Discovering New Therapeutic Uses for Existing Molecules” that tests a new model for PPP collaborations including template agreements to shorten the time it takes to establish collaborations between an academic institution and a pharmaceutical company and move more rapidly into the actual research . The pilot involves eight pharmaceutical companies that made 58 assets available for repurposing by the broader research community. Collaborations established between academic institutions and the company will test ideas for new therapeutic uses, with the ultimate goal of identifying promising new treatments for patients. Before joining NCATS, Christine had been a program director at the National Institute on Drug Abuse and Director of Program Integration. She led NIDA's management of the ARRA which resulted in commitments of over \$300,000,000 made by the Institute in 7 months. She has also been a leader and advisor for complex NIH programs such as the Molecular Libraries and the NIH Epigenomics programs.



**Cristina Csimma** has held drug development roles in biopharmaceutical, venture capital and academic settings. She is currently Chief Executive Officer of Cydan Development, Inc., a privately-held company focused on advancing the translation of therapeutics for orphan diseases.



Her former roles include: Vice President, Drug Development at Virdante Pharmaceuticals Inc., Principal at Clarus Ventures LLC, a life sciences venture capital firm, and several senior roles in Clinical Research and Development and in Translational Research at Wyeth Research and Genetics Institute. Her extensive drug development experience encompasses global development in multiple therapeutic areas including neuromuscular diseases, exploratory through registration clinical studies, and integration of biomarkers and novel technologies into clinical development. Before joining the biopharmaceutical industry, she was a clinical pharmacist at the Dana Farber Cancer Institute.

She is a member of: the MDA Venture Philanthropy Scientific Development Advisory Committee; the TREAT-NMD Advisory Committee for Therapeutics (TACT) (former chair), the DoD CDMRP Duchenne Muscular Dystrophy Research Program Integration Panel, and Northeastern University's Health Sciences Entrepreneurs program steering committee. She serves on the board of directors of Columbia Labs Inc.

Dr. Csimma received her Bachelor of Science and Doctor of Pharmacy degrees from Massachusetts College of Pharmacy, and a Master of Health Professions from Northeastern University.

Dr. **Garry Cutting** is Professor of Pediatrics and Medicine and the Aetna/U.S. Healthcare Professor of Medical Genetics at Johns Hopkins University School of Medicine. Dr. Cutting received his undergraduate and medical degrees at the University of Connecticut. He completed both residency training in Pediatrics (1986) and a fellowship in Medical Genetics (1989) at Johns Hopkins and has remained at Hopkins for his entire professional career. Dr. Cutting's primary research interest is in the molecular genetics of cystic fibrosis and more recently elucidating modifier genes underlying variation in the severity of cystic fibrosis. He is also the Director of the CFTR2 project, a worldwide collection of genotype and phenotype data on over 40,000 patients with cystic fibrosis.



Previous research achievements in other areas include the cloning and characterization of a new class of GABA receptor subunits (GABA<sub>c</sub>) and structure/function studies of voltage gated chloride channels.

Dr. Cutting's clinical and educational activities are focused on laboratory diagnostics. He is the Director of the DNA Diagnostic Laboratory and Director of the Clinical Genetics Laboratory Training Program at Johns Hopkins University School of Medicine. Dr. Cutting facilitates translation of bench top discoveries at Hopkins to clinical application as the Director of the Genetic Translational Technology Core at Hopkins. Dr. Cutting is the Co-Editor of the journal Human Mutation and Board member of the Federation of American Societies for Experimental Biology. He is the recipient of the Paul di Sant'Agnese Distinguished Scientific Achievement Award from the Cystic Fibrosis Foundation and a MERIT award from the NIH and has published over 150 peer-reviewed articles.

**Avril Daly** has worked with Fighting Blindness since June 2000. During her time as Head of Public Affairs, she advocated for mechanisms to be put in place to support research into retinal disease in particular and rare diseases in general. She was also responsible for raising awareness of rare retinal degenerative diseases, including retinitis pigmentosa (RP), LCA, Stargardt's disease and LHON as well as more common chronic conditions such as age-related macular degeneration and diabetic retinopathy. Avril has been CEO of Fighting Blindness since 2009 and is responsible for the continuation of the charity's significant research portfolio and the creation and implementation of the Fighting Blindness Strategic Development Plan.



Avril represents Fighting Blindness on the board of the Irish Platform for Patients' Organisations, Science and Industry (IPPOSI) and in the Medical Research Charities Group (MRCG). She is the current chair of the National Rare Disease Alliance - Genetic and Rare Disorders Organisation (GRDO). She was the first Irish person to be elected to the Board of the European Organisation for Rare Diseases (EURORDIS) in 2009 and was appointed Vice President in May 2012. She was advisor to EUROPLAN I for Ireland and for the UK and is now working on Europlan 2, an EU Commission initiative to establish national plans for rare diseases throughout the EU's Member States.

Throughout her education, Avril worked as a volunteer in the disability sector. Having studied Communications and Business, she wanted to apply her skills in this area to the promotion of patient rights, which she continued to do on a voluntary basis while working in the media sector. Avril is a person living with the rare disease retinitis pigmentosa (RP), which causes the degeneration of the retina, and has 15% vision. Although she has been living with the symptoms since pre teens, she was diagnosed at age 23.

Prior to working with Fighting Blindness, Avril worked in publishing and film production.

### Rafael De Andres – Medina

- Chief of the Docs. and Technical Studies Department, Fund for Health Research (FIS), National Institute of Health Carlos III (**Instituto de Salud Carlos III / ISCIII**), Madrid (Spain), since 2004.
- Deputy Center Director and Head of the Docs. and Technical Studies Department at the Institute for Research on Rare Diseases (**IIER**), Madrid, Spain, 2003-2004. National Institute of Health Carlos III / ISCIII.

Delegate representing Spain (ISCIII) in:

- The Executive Committee of **IRDiRC** (International Rare Diseases Research Consortium).
- Art. 185 TFEU: **AAL** (Ambient Assisted Living, also serving as the Treasurer at its Executive Board) and **EDCTP** (European and Developing Countries Clinical Trials Partnership, also served as Vice-chair of its ENNP, 2009).
- European Joint Programming Initiatives: **JP ND** (Neurodegeneration) and **JP MYBL** (More Years Better Life: demographic change).
- ESFRI H&FSWG (Health and Food Strategic Working Group, also serving as its ad hoc Vice-chair).
- ESFRI / ERICs: **ELIXIR** (European Life-science Infrastructure for Biological Information). **BBMRI** (Biobanking and Biomolecular Resources Research Infrastructure, also serving as the Chair of its Search Committee for the recruitment of the Director General of BBMRI-ERIC). **EATRIS** (European Advanced Translational Research Infrastructure). **ECRIN** (European Clinical Research Infrastructures Network and Biotherapy, also serving as the Chair of its PDB - member states' ministries and funding agencies -, in the feasibility phase 2008-2012). **ERINHA** (European Research Infrastructure on Highly Pathogenic Agents).
- He is also active in ERA-Nets: E-RARE, NEURON, Transcan, EuroNanoMed, ERASysAPP, ...



**Chris Denning** is a Professor in Stem Cell Biology, with particular interests in cardiomyocyte differentiation of human embryonic and induced stem cells for use in drug screening and in production of new in vitro models of genetic-based cardiovascular disease. Associated with this, he has also focussed on optimisation of culture conditions, in robotic culture to allow automated scale-up and high throughput screening, and in genetic modification.



PhD in Cancer Gene Therapy, Glasgow, 1997;

Postdoc: Institute for Stem Cell Research, Edinburgh 1997-98;

Postdoc: Roslin Institute 1998-2001;

Fellow (2001), Lecturer (2006), Reader (2008), Professor (2011) in Stem Cell Biology, University of Nottingham

**Luc Dochez**, Chief Business Officer and Senior Vice-President Business Development joined Prosensa in November 2008 and is responsible for all business and corporate development activities at the company. He has over 15 years experience in the biotech industry and was directly involved in multiple financing rounds and partnering deals with various biotech companies. Before joining Prosensa, he was a consultant within Arthur D. Little's biotechnology practice, Director of Business Development at Methexis Genomics NV, VP Business Development at TiGenix NV and President of TiGenix Inc. Luc holds a PharmD degree from the University of Leuven (Belgium), a postgraduate degree in Business Economics from the same university and an MBA from Vlerick Management School. He is also a Supervisory Board member of Ovizio SA.



Dr **Ruxandra Draghia-Akli** (MD, PhD) is Director of the Health Directorate at the Research and Innovation DG of the European Commission.

Dr Draghia-Akli served as Vice-President of Research at VGX Pharmaceuticals (now Inovio) and VGX Animal Health. Her research activities focused on molecular biology, gene therapy and vaccination. She is a global leader in the field of nucleic acid delivery for therapeutic and vaccination applications. She is an inventor on more than a hundred patents and patent applications.

Dr Draghia published numerous scientific papers and served as ad-hoc reviewer for granting agencies, meetings for gene therapy and endocrinology societies, and scientific journals in Europe and the USA.

Dr Draghia received an MD from Carol Davilla Medical School and a PhD in human genetics from the Romanian Academy of Medical Sciences. She also completed a doctoral fellowship at the University of Rene Descartes in Paris and a post-doctoral training at Baylor College of Medicine (BCM), Houston, Texas, USA, and served as faculty at BCM. In 2012, she became an honorary member of the Romanian Academy of Medical Sciences.



**Stuart Elborn** is Director of the Centre for Infection and Immunity in Queens University Belfast and Professor of Respiratory Medicine. He runs an Adult CF programme with 280 patients and collaborates closely with his paediatric colleagues who provide care for 180 patients. His scientific interests are in a number of areas:

- **Clinical trials.** He has been involved in a range of clinical trials involving antibiotics, anti-inflammatory agents and CFTR modulating drugs. He also has a clinical trials programme in Bronchiectasis. He leads a team of PI's who have recently been selected as part of a Translational Research Partnership in the UK which focuses on academics in the Pharma industry in preclinical and early clinical trials. His group are also part of the European CF Society Clinical Trials Network. He is PI on a number of early stage anti-inflammatory and potentially disease modifying therapies in CF and other lung diseases.
- **Clinical and laboratory research.** He has a major programme funded jointly by NIH, MRC and SFI investigating the clinical implications of bacterial diversity and in particular anaerobes in the CF airway. This involves clinical research and also a range of projects investigating the interaction between bacteria and the host innate immunity.
- **Research and care management.** He is President of the European Cystic Fibrosis Society and a Chair of the Research Advisory Committee of the UK CF Trust.



Dr. **Christine Eng** is Senior Director of the Medical Genetics Laboratories and Medical Director of the Whole Genome Laboratory at Baylor College of Medicine. She is Board Certified in pediatrics and medical genetics. Her interests are in translation of molecular techniques to the diagnosis of genetic disorders most recently utilizing whole exome sequencing. She also has a long-standing clinical interest in the lysosomal storage disorders.



**Alessandra Ferlini** (MD, PhD)

Associate Professor at the University of Ferrara (Italy) and Director of the Medical Genetics Unit, born in 1958 in Bologna (I), married, one son. She graduated in medicine and surgery at the University of Bologna in 1983. She specialized in Neurology (1988) and Medical Genetics (1993) at the Universities of Bologna and Ferrara. In 2002 she achieved a PhD in Genetics at the Imperial College School of Medicine in London (UK). She is Director of both Unit (Hospital) and Section (University) of Medical Genetics. As part of her professional career she has accomplished profound research experience at the Universities of Bologna, Padova, Milan and Modena. In 1995 she moved to London where she worked as Senior Research Officer at the Hammersmith Hospital, Neuromuscular Unit, Department of Paediatrics, under the supervision of Prof. F. Muntoni. In June 1999 she returned to Italy.



She is member of the American Society of Human Genetics, the American Society of Cell and Gene Therapy, the European Society of Human Genetics, the World Muscle Society, the Italian Society of Human Genetics and the Italian Association of Myology. She is also Associate Editor of the journal *Neuromuscular Disorders*.

She published more than 150 scientific articles in international magazines (reaching a total impact factor of 470).

**Helen V. Firth**, D.M., FRCP is a Consultant Clinical Geneticist at Cambridge University Hospitals, Cambridge, UK. In 2004, Dr. Firth initiated the DECIPHER project <http://decipher.sanger.ac.uk> that enables clinical geneticists around the world to share information about rare genomic variants to facilitate diagnosis and help to elucidate the role of genes whose function is not yet known.



Since 2010 Dr. Firth has been Clinical Lead for the Deciphering Developmental Disorders study (DDD study) [www.ddduk.org](http://www.ddduk.org). DDD is a partnership project between all 23 NHS Regional Genetics services in the UK and the Wellcome Trust Sanger Institute to undertake detailed genomic analysis of 12,000 children with severe developmental disorders. The DDD study aims to improve the diagnosis of developmental disorders and to further understanding of the genomic architecture and biology of these conditions. A further aim of the DDD study is to translate whole genome technologies such as exome sequencing from a research environment into clinical practice.

Dr. Firth is an Honorary Faculty Member of the Wellcome Trust Sanger Institute, Hinxton, UK and a Bye-Fellow of Newnham College, Cambridge. She has published more than 50 peer-reviewed manuscripts and co-authored two books: 'Oxford Desk Reference: Clinical Genetics' (OUP 2005) and 'Oxford Handbook of Genetics' (OUP 2009).

**Anne Galy** PhD

Director of Program "Blood and Immune Diseases" at Genethon  
Director of Inserm Unit U951

Anne Galy has received a Doctorate in Pharmacy in 1985 and a PhD in Immunology at the University of Lyon France in 1989. Following completion of hospital residency in biology, she trained as a post-doctoral fellow at DNAX research institute in Palo Alto, California to study T cell development, then worked as a scientist at SySTEMix Inc. also in Palo Alto to further study lymphopoiesis and human hematopoietic stem cells. In 1995 she joined the faculty at the school of medicine of Wayne State University and Barbara Ann Karmanos Cancer Institute in Detroit, Michigan where she became a tenured Associate Professor. In 2001 Dr. Galy was recruited to Inserm, the French National Medical Research Institute where she holds a permanent position as Director of Research.



Dr. Galy's laboratory is situated within Genethon, a non-for profit institute dedicated to the treatment of rare diseases by cell and gene therapy.

Dr. Galy is expert in immunology and gene therapy. She was awarded the "Thermo Fischer Scientific" Biotherapy prize in 2009. She sits on the scientific editorial board of "Human Gene Therapy", on the gene therapy committee of the International Society for Cell Therapy and on the board of the French Society for Cell and Gene Therapy.

Clinical Professor **Jack Goldblatt** AM, MB ChB, MD, FCP, FRACP (Clin. Geneticist, HGSA) is the Director of Genetic Services and the Familial Cancer Programme of Western Australia. He has been president of the Human Genetics Society of Australasia and a foundation board member of the International Federation of Human Genetics Societies. He has co-authored 223 papers in international, peer reviewed journals on aspects of clinical, biochemical and molecular genetics. Prof. Goldblatt is a specialist physician and medical geneticist who has worked in the academic field of human genetics since 1975. In 2011 he was made a Member of the Order of Australia in the General Division (AM) in the Queen's Birthday Honours List for his service to medicine in the area of human genetics as a clinician and researcher.



Dr. **Ivo Gut**, Director, Centro Nacional de Análisis Genómico (CNAG)

Ivo Gut is qualified in Chemistry at the University of Basel and obtained a PhD in Physical Chemistry from the same University with Prof. Jakob Wirz in 1990. He was a Research Fellow in the group of Prof. Irene Kochevar at Harvard Medical School (1990-1993), and in the laboratory of Dr. Stephan Beck Imperial Cancer Research Foundation of London (1993 to 1996).

He led a group in the Department for Vertebrate Genomics of Prof. Hans Lehrach. Later at the Max-Planck-Institute for Molecular Genetics (1996-1999), and during 11 years before joining the CNAG (2010), he was at the Centre National de Génotypage (CEA) first as a Head of Technology Development and later as an Associate Director.

His research interests are genomics, genetics, high-throughput nucleic acid analysis methods, proteomics, implementation of -omics methods, automation and data analysis.

Ivo Gut is author of more than 160 research papers, inventor of 25 patents or patent applications, founder of 4 biotech companies (Genom Analytik GmbH, Biopsytec GmbH, Epigenomics AG and Integragen SA), and Coordinator of the 12M€ EU FP7-funded Integrated Project READNA on DNA sequencing technology.



Dr. **Ada Hamosh**, the Dr. Frank V. Sutland Professor of Pediatric Genetics, is the Clinical Director of the McKusick-Nathans Institute of Genetic Medicine at Johns Hopkins University School of Medicine, the Scientific Director of Online Mendelian Genetics in Man (OMIM®), and the Co-Chair of the Phenotype Review Committee of the combined and Baylor-Hopkins Centers for Mendelian Genomics (CMG), a National Human Genome Research Institute-funded project to identify the genes responsible for known and novel Mendelian disorders. Over the past year, she and colleagues have developed PhenoDB (<http://phenodb.net>), a web-based tool for the collection, storage, and analysis of standardized phenotype data for use in the CMG project that is freely available to all for use in clinical and research use.



**Katherine A. High, M.D.**

William H. Bennett Professor of Pediatrics, Univ. PA School of Medicine  
Investigator, Howard Hughes Medical Institute  
Director, Center for Cellular and Molecular Therapeutics, CHOP

Katherine High graduated from Harvard with a degree in Chemistry in 1972, and from the University of North Carolina (UNC) School of Medicine, where she received her M.D. degree. After completing her training in internal medicine, she trained as a Fellow in the Hematology Section at Yale University School of Medicine. Presently, Dr. High is the William H. Bennett Professor of Pediatrics at the Perelman School of Medicine at the University of Pennsylvania, Investigator, Howard Hughes Medical Institute, and Director, Center for Cellular and Molecular Therapeutics at The Children's Hospital of Philadelphia. Dr. High has been elected to Institute of Medicine (IOM) and the American Academy of Arts & Sciences (AAAS).



Dr. High's research interests focused initially on the molecular basis of blood coagulation, and the use of novel genetic therapies to treat hemophilia. More recently she has pioneered safe and effective clinical translation of genetic therapies for inherited disorders. These clinical trials have led to correction of disease in hemophilia B and in Leber's congenital amaurosis, a hereditary cause of blindness.

**Wills Hughes-Wilson** is **Vice President External Affairs & Chief Patient Access Officer at Sobi** and a **member of the global Executive Management Team**. She joined Sobi in February 2012.

Hughes-Wilson is responsible for the company's external liaison with all stakeholders working to support the availability of treatments for rare diseases on behalf of Sobi; and for supporting the development of sustainable healthcare systems for rare disease diagnosis, treatment and care.

Hughes-Wilson is a **member of the European Commission's EU Committee of Experts on Rare Diseases (EUCERD)**, the EU-level committee officially responsible for advising the European Commission on all aspects of implementing effective rare disease policies across Europe. This includes key aspects around reimbursed access to orphan drugs. She is the spokesperson for the industry representation on this Committee.

She is an **industry representative on the working group of European pricing & reimbursement authorities looking at access to orphan drugs ("MOCA")**.

Hughes-Wilson is also **Chair of the European Industry Task Force on Orphan Drugs & Rare Diseases**, a joint working group that brings together at a European level the companies involved in the research and development of orphan drugs and treatments for rare diseases.

Prior to joining Sobi, Hughes-Wilson was the head of Health- & Market Access Policy at Genzyme EMEA for more than 6 years. She has led the Emerging Biopharmaceutical Enterprises (EBE), a specialised group of the European Federation of Pharmaceutical Industries & Associations (EFPIA), representing the interests of biotechnology companies in Europe, as well as holding key positions in the animal health / veterinary medicines industry and Ernst & Young Consulting.

Hughes-Wilson is an Honours graduate in Law.



**Emil D. Kakkis**, M.D., Ph.D., Chief Executive Officer and President

Dr. Kakkis is best known for his work over the last 18 years to develop novel treatments for rare disorders. He began his work developing an enzyme replacement therapy (Aldurazyme®) for the rare disorder MPS I, with minimal funding and support. The struggle to get the therapy translated from a successful canine model to patients succeeded due to the critical financial support of the Ryan Foundation, a patient organization formed by Mark and Jeanne Dant for their son Ryan.

Aldurazyme development was later supported by BioMarin Pharmaceutical and eventually their partner Genzyme leading to FDA approval in 2003. During his tenure at BioMarin, Dr. Kakkis guided the development and approval of two more treatments for rare disorders, MPS VI and PKU, and has contributed to the initiation of 7 other treatment programs for rare disorders, three of which are now in clinical development.

After 11 years at BioMarin, Dr. Kakkis left industry to initiate an effort to improve the regulatory and clinical development process for rare diseases. In early 2009, Dr. Kakkis launched and funded the Kakkis EveryLife Foundation to accelerate biotech innovation for rare diseases. The Foundation initiated a campaign to improve the regulatory and clinical development process for rare diseases. In just over a year, 160 patient organizations and physician society partners have endorsed the Campaign.

Dr. Kakkis has founded Ultragenyx™ to return to development of drugs for rare diseases. For many rare diseases, reasonable science exists that needs to get translated to patients. He will build on his previous experiences and will assemble an experienced team to efficiently develop treatments for rare diseases.

Dr. Kakkis is board certified in both Pediatrics and Medical Genetics. He graduated from Pomona College, magna cum laude and received combined M.D. and Ph.D. degrees from the UCLA Medical Scientist Program and received the Bogen prize for his research. He completed a Pediatrics residency and Medical Genetics Training Fellowship at Harbor- UCLA Medical Center. He became an assistant professor of Pediatrics at Harbor-UCLA Medical Center from 1993 to 1998 where he initiated the enzyme therapy program for MPS I. In 1998, he joined BioMarin where he remained for 11 years in various titles eventually as Chief Medical Officer, before leaving in 2009.



Dr. **Nicholas Katsanis** obtained his first degree in Genetics from UCL in London in 1993 and his doctorate from Imperial College, University of London in 1997. He then joined the laboratory of Dr. Lupski at Baylor College of Medicine, where he initiated his studies on Bardet-Biedl syndrome. In 2002, he relocated to the Institute of Genetic Medicine, Johns Hopkins University where he led studies that contributed to the unification of several allied conditions under the ciliopathy umbrella. In 2009, he moved to Duke University to establish the Center for Human Disease Modeling, where he is the Director; this new structure aims to facilitate collaboration across disciplines and to develop physiologically relevant tools to study variation found in human patient genomes. In parallel, the Katsanis lab pursues questions centered on the signaling roles of vertebrate cilia, the translation of signaling pathway defects on the causality of ciliary disorders, and the dissection of second-site modification phenomena as a consequence of genetic load in a functional system. In recognition of his work, Dr. Katsanis was awarded the Young Investigator Award from the American Society of Nephrology in 2009 and the E. Mead Johnson Award from the Society for Pediatric Research in 2012. Dr Katsanis is a Professor in the Departments of Cell Biology and Pediatrics and holds the Brumley Distinguished Professorship. He has published over 170 research papers, reviews, and book chapters, serves on several advisory, editorial and organizational boards and has delivered over 120 lectures in 17 countries.



**Alastair Kent** OBE is the Director of Genetic Alliance UK – the national charity of over 150 patient organisations, supporting all those affected by genetic conditions. Genetic Alliance UK's mission is to promote the development of the scientific understanding of genetics and the part that genetic factors play in health and disease, and to see the speedy transfer of this new knowledge into improved services and support for patients.

Alastair is also the Chair of Rare Disease UK (RDUK) the national alliance for people with rare diseases and all who support them. RDUK has over 1,200 members including over 220 patient organisations, health professionals, researchers, the pharmaceutical industry and individual patients and families.

Alastair has worked in the field of genetic and rare disease healthcare for over 15 years. Alastair represents the interests of patients on numerous platforms; he is the president of the European Genetic Alliances Network (EGAN), Director of the European Platform for Patient Organisations, Science and Industry (EPPOSI) and the EU Committee of Experts on Rare Diseases amongst others.



**Christoph Klein** is chair of the department of pediatrics at the Ludwig-Maximilians-University Munich, Germany. He has a longstanding clinical and scientific interest in rare disorders of the blood and immune system. His team has discovered the genetic basis of several primary immunodeficiency disorders and is actively involved in the development of innovative therapeutic strategies. Christoph Klein is coordinator of national and international networks on rare diseases of the immune system and serves as spokesman for the German networks on rare diseases. He has received numerous scientific awards such as the William-Dameshek Award by the American Society of Hematology and the Gottfried-Wilhelm-Leibniz Award by the German Research Foundation.



**Bartha Maria Knoppers**, PhD (Comparative Medical Law), holds the Canada Research Chair in Law and Medicine (Tier 1: 2001 - ). She is Director of the Centre of Genomics and Policy, Faculty of Medicine, Department of Human Genetics, McGill University.

Former Chair of the International Ethics Committee of the Human Genome Project (HUGO), (1996-2004), she was a member of the International Bioethics Committee of the United Nations, Educational, Scientific and Cultural Organization (UNESCO) which drafted the *Universal Declaration on the Human Genome and Human Rights* (1993-1997). In 2007, she founded the international Population Project in Genomics and Society (P3G) and CARTaGENE Québec's population biobank (20,000 indiv.). Former holder of the Chair d'excellence Pierre Fermat (France: 2006-2008), she was named Distinguished Visiting Scientist (Netherlands Genomics Initiative) (2009- 2012) and received the ACFAS prize for multidisciplinary (2011). She is Chair of the Ethics Working Party of the International Stem Cell Forum (2006 - ), Co-Chair of the Sampling/ELSI Committee of the 1000 Genomes Project (2008 - ) and a member of the Scientific Steering Committee of the International Cancer Genome Consortium (ICGC) (2009- ).



She holds four Doctorates Honoris Causa, is Fellow of the American Association for the Advancement of Science, of The Hastings Center (Bioethics) and of the Canadian Academy of Health Sciences (CAHS) and Officer of the Order of Canada and of Québec. She also received an award "Prix Montreal In Vivo: Secteur des sciences de la vie et des technologies de la santé".

Prof. **Jeffrey Krischer** (USA): Professor and Chief of the Division of Bioinformatics and Statistics, and Director of the Pediatric Epidemiology Center, Department of Pediatrics, University of South Florida College of Medicine in Tampa, Florida. He has a long-standing interest in the design, implementation and analysis of clinical trials in rare diseases. In 2003 and again in 2009, Dr. Krischer's office was selected to be the Data Technology and Coordinating Center for the Rare Disease Clinical Research Network funded by the Office of Rare Diseases Research, NIH. The data management for this international study group is entirely web-based and supports a network of 19 clinical consortia that extends to more than 200 clinical sites worldwide. Dr. Krischer is expert in the design and conduct of multi-institutional clinical trials and is also Co-Principal Investigator, with Dr. Peter Merkel of the University of Pennsylvania, of a study of novel methods in the conduct of clinical trials for vasculitis.



**Paul Lasko**, Ph. D.

James McGill Professor, Department of Biology, McGill University  
Scientific Director, CIHR Institute of Genetics

Prof. Lasko received his PhD from the Massachusetts Institute of Technology in 1986 and joined McGill in 1990 after a postdoctoral period at the University of Cambridge. Using the *Drosophila* system, Dr. Lasko's research concerns regulatory processes that control gene expression at the levels of mRNA stability or translation, and that underlie germ cell or early embryonic development. He received the Award of Excellence from the Canadian Society of Genetics in 2004. At McGill he served as Chair of the Department of Biology from 2000-2011. He assumed his position at CIHR in May 2010 but maintains his research lab at McGill.



Dr. Lasko has been highly active in research grant adjudication and served on CIHR or Canadian Cancer Society grant panels continuously since 1995. He has also worked extensively for the Human Frontiers of Science Program Organization (HFSP) over the past ten years, serving on its program grant panel from 2001-2005, and then as one of two Canadian representatives on the Council of Scientists. He chaired the HFSP Council of Scientists from 2007-2010. Dr. Lasko also served as President of the Genetics Society of Canada from 2007-2010.

As Scientific Director of the CIHR Institute of Genetics, Dr. Lasko oversees the Institute's strategic research funding initiatives, many of which involve fostering international partnerships. He is the incoming chair of the Executive Committee of the International Rare Diseases Research Consortium.

**Yann Le Cam**, CEO of EURORDIS

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

Yann was one of the founding members of the European Organisation for Rare Diseases in 1996-1997 and joined EURORDIS as Chief Executive Officer in 2001.

Yann contributed to the adoption of the European Regulation on Orphan Drugs in December 1999. He was one of three patient representatives appointed to the Committee for Orphan Medicinal Products (COMP) at the European Medicines Agency (EMA) from the first mandate in 2000-2003 to the third mandate 2006-2009, and served as its Vice Chairman from 2000 to 2006. In addition, He was Co-Chair of the COMP Working Group of Interested Parties at the EMA. He was a member of the DG Sanco Task Force on Rare Diseases. Yann contributed to the adoption of the Commission Communication « Rare Diseases: Europe's challenges » and of the Council Recommendation for an Action in the field of Rare Diseases promoting national plans or strategies on rare diseases across Europe.

Currently, Yann is a member of the EU Committee of Experts on Rare Diseases (EUCERD) in Luxembourg since its creation in 2010 and serves as Vice-Chairperson of EUCERD since 2011. Yann has advocated in several other European Commission working groups such as the EU High Level Group on Healthcare Organisation, EU Pharmaceutical Forum, EU Mechanism of Coordinated Access to Orphan Drugs.

Yann served as Director General of the AIDES Fédération Nationale from 1992 to 1998. He was Special Advisor to the French Neuromuscular Association (AFM-Téléthon) from 1998 to 2000 and founded the Alliance Maladies Rares, a national umbrella organisation of over 200 patient associations in France. He co-founded and served as Vice Chair of the Board of Directors of the International Alliance of Patients Organisations (IAPO) in London until 2000. In 2001, Yann was appointed to the Management Board and Executive Board of the French National Agency for Health Accreditation and Evaluation (ANAES) until 2005 and served until 2008 on the Commission for Quality of Medical Information at the French High Health Authority (HAS) in Paris.

Yann holds an Executive MBA from the Hautes Etudes de Commerces – HEC – Jouy-en-Josas France (2000) and an MBA from the Institut Superior de Gestion (1984), Paris, France.

Yann has three daughters, the eldest of whom has cystic fibrosis. He lives in France, Paris, and in Belgium, Brussels.

**David K. Lee** is the Director of the Office of Legislative and Regulatory Modernization, Health Canada.

The mandate of the Office of Legislative and Regulatory Modernization is to modernize the Canadian *Food and Drugs Act* and corresponding regulatory frameworks. The modernization of these regulatory tools will focus on food safety frameworks and the implementation of a life-cycle approach to the regulation of health products, including pharmaceuticals, biologics, and medical devices.

Formerly, Mr. Lee was the Director of the Office of Patented Medicines and Liaison which administers the *Patented Medicines (Notice of Compliance) Regulations* and provides liaison for Therapeutic Products Directorate litigation under the *Food and Drugs Act* and Regulations along with the Department of Justice. The scope of proceedings managed under the *Food and Drug Regulations* has included judicial reviews in the Federal Court of Canada, actions and class actions relating to drug injury and alleged regulatory negligence and coroner's inquiries.

Mr. Lee received his law degree at Queen's University at Kingston Ontario, and articulated at the Federal Court of Canada. He worked in private practice until joining Health Canada in 1999. He has lectured extensively on intellectual property law and pharmaceuticals law both nationally and internationally.



**Kevin Lee** is CSO and Head of the Rare Disease Research Unit at Pfizer.

Prior to joining Pfizer, Kevin conceptualized and led epigenetics research at GSK and was responsible for the creation of the EpiNova DPU as well as leading the formation of multiple strategic commercial and academic partnerships for the company. Kevin studied pharmaceutical sciences at Nottingham University followed by a PhD in pharmacology at Cambridge. He undertook postdoctoral training as a Wellcome Trust International Prize Fellow before joining the Parke Davis Research unit in Cambridge.

Prior to joining GSK, Kevin lectured at Warwick Medical School, founded Cambridge Biotechnology and Neurosolutions. Kevin is an author on over 100 peer reviewed scientific publications, has an MBA from Warwick Business School and has been awarded an honorary Chair in Molecular Pharmacology from the University of Warwick



**Nicolas Lévy** is Professor of Human and Molecular Genetics. He is the head of the Medical Genetics department and of the Inserm research center “ Medical Genetics and Functional Genomics” in Marseille, France. In 2009, he was appointed as the director of the French national institute for rare diseases and, in 2012, he is in charge of creating and heading the “rare diseases foundation”, a unique alliance of academic partners and private entities to promote, organise, federate and facilitate research in rare diseases.

After having identified the LMNA mutation causing Progeria, Nicolas Lévy's efforts have been dedicated to gene's identification in related progeroid syndromes, understanding of pathophysiological mechanisms involved in Progeria and development of therapeutic strategies. In collaboration with Carlos Lopez-Otin, his laboratory has developed and pre-clinically tested a combination of global prenylation inhibitors in human cells and living mouse model. Based on the demonstrated efficacy of this combination, Nicolas Lévy is the PI of the first phase II trial using amino-bisphosphonates and Statins for Progeria, at La Timone Hospital in Marseille, France. Based on a newly generated mouse model of Progeria, Carlos Lopez-Otin and Nicolas Lévy's teams recently developed an antisense based approach as a potential treatment for Progeria. In the last years, his long term pathophysiological and pre-clinical studies towards the development of therapeutic strategies in rare diseases, led his team to demonstrate proofs of principle of both exon skipping and AAV-minigene transfer as potential therapies in rare neuromuscular disorders.



**Jordi Llinares-Garcia**, M.D., MSc  
Orphan Medicines Section Head  
European Medicines Agency (EMA), UK

Dr. Jordi Llinares is the head of orphan medicines at the European Medicines Agency (EMA). He is responsible for the group assessing applications for orphan designation and coordinates the activities necessary for the adoption of opinions on orphan designation, including the drafting of summary reports and supports the secretariat of the Committee for Orphan Medicinal Products (COMP).



Dr. Llinares joined the EMEA in 2002. Prior to joining the EMEA, Dr Llinares worked as clinical pharmacologist at Hospital de Sant Pau (Barcelona) and collaborated with the Spanish Drugs Agency as clinical assessor. Dr Llinares was a member of the Ethics Committee of the Institut d'Assistència Sanitària (Salt).

Dr. Llinares graduated as an MD from the University of Barcelona and specialised (MIR) as clinical pharmacologist at Hospital Vall d'Hebron (Barcelona). Dr Llinares holds a Masters in Science in Epidemiology from the London School of Hygiene and Tropical Medicine (London).

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### **Hanns Lochmüller**

Professor, University of Newcastle upon Tyne, UK

Professor Hanns Lochmüller trained as a neurologist in Munich (Germany) and Montreal

(Canada). He was appointed chair of experimental myology in the neuromuscular research group at the Institute of Genetic Medicine of Newcastle University in 2007. Hanns has a longstanding interest in the molecular genetics of the inherited myopathies and neuromuscular junction disorders, and is interested in the further study of animal models of these disorders as a means to understand their pathophysiology as well as to develop methods of monitoring disease progression and therapeutic interventions. His ongoing work in these areas in cell and animal models of muscular dystrophy is concentrating on gene transfer, pharmacological interventions and cell therapy. Hanns is actively involved in rare disease networking activities at a European level and is co-founder and former coordinator of the German muscular dystrophy network (MD-NET) and member of the Executive Committee of the TREAT-NMD Alliance. He plays a leading role in European biobanking and patient registries activities as the scientific coordinator of EuroBioBank, a European network of biobanks for rare disorders, and leader of the TREAT-NMD Network of Excellence activity on patient registries and



biobanking. He chairs the TREAT-NMD Global Database Oversight Committee which unites the neuromuscular patient registries in more than 40 countries worldwide.

Professor **Milan Macek** Jr. MD, DSc is the chairman of the largest academic medical / molecular genetics institution in the Czech Republic, which also comprises a research / diagnostics reproductive genetics centre /ublg.lf2.cuni.cz/. He is also the Vice President of the European Society of Human Genetics ([www.eshg.org](http://www.eshg.org)), board member of the European Society for Human Reproduction and Embryology ([ESHRE.com](http://ESHRE.com)) and of the European Cystic Fibrosis Society ([ECFS.eu](http://ECFS.eu)). His institute is a “clearing centre” for dissemination of knowledge in genetics gathered within various international European projects, such as CF Thematic Network, EuroGentest, EuroCareCF or Techgene, to Central and Eastern Europe. Prof. Macek did his first postdoc at the Institut of Human Genetics in Berlin, continued as a postdoctoral fellow at the McKusick-Nathans Centre for Genetic Medicine, Johns Hopkins University in Baltimore and during that time he was also a fellow at Harvard School of Medicine in Boston. He was the local host of the 1995 HUGO Mutation Detection Course in Brno, the 2005 European Society of Human Genetics conference and of the 2008 European Cystic Fibrosis Conference, both held in Prague. Prof. Macek is national coordinator of Orphanet ([www.orpha.net](http://www.orpha.net)), active member of Eurogentest ([www.eurogentest.org](http://www.eurogentest.org)), has been the chief advisor of the Czech EU Council Presidency under which the “EU Council recommendation on an action in the field of rare diseases” was adopted in June 2009. He also serves at the EUCERD.eu committee on rare diseases.



**Gert Matthijs** is a molecular geneticist, involved in the diagnostics of inherited diseases. He is the head of the Laboratory for Molecular Diagnostics at the Center for Human Genetics, at the University Hospital in Leuven, Belgium. The Center for Human Genetics is the largest genetic department in Belgium.

He is the coordinator of EuroGentest, a network for development, harmonization and standardization of genetic testing in Europe, funded by the European Commission. It also deals with clinical, legal and ethical aspects of genetic testing, and with the introduction of new technologies for diagnostics. EuroGentest aims at setting new standards for genetic testing in Europe.

His research interest is in Congenital Disorders of Glycosylation (CDG), a group of rare inborn errors of metabolism. His group is focusing on the systematic search for novel types of CDG. The success is partly due to the fact that the Leuven group has committed itself, since 1999, to the coordination of EUROGLYCANET, a European network and database on CDG.

His (translational) research activities deal with the development and validation of novel technologies for diagnostic use. Currently, the focus is on the implementation of the newest massive parallel sequencing platforms for mutation scanning in a diagnostic setting.

Gert has been a board member of the European Society of Human Genetics (ESHG). He was very actively involved in the European opposition against the BRCA patents. At the national level, he has been a thriving force for a revision of the reimbursement system for genetic tests.

He is an alternate member of EUCERD and a member of the Diagnostics Committee of IRDiRC.



**Maria Mavris** PhD

EURORDIS Therapeutic Development Director

Maria joined the Research and Therapeutic team of EURORDIS as Drug Development Programme Manager in January 2008 and in January 2012 she became the Therapeutic Development Director. She is responsible for following the development of orphan drugs as an observer on the Committee for Orphan Medicinal Products (COMP) at the European Medicines Agency (EMA). She is also responsible for coordinating the group of high-level EURORDIS representatives who sit on the various scientific committees at EMA. Maria is also implicated in activities of the working parties at the EMA, namely the Scientific Advice Working Party (SAWP) where she is responsible for the identification of patients' representatives to participate in Protocol Assistance and she has a supportive role for EURORDIS representatives in the Patients' and Consumers' Working Party.



In addition, Maria is co-leader on the EURORDIS Round Table of Companies (ERTC), a bi-annual workshop for companies involved in the development of orphan therapies for rare diseases.

In order to train and support patients' representatives in regulatory activities, she is also the organiser of the EURORDIS Summer School in clinical trials and drug development, a capacity-building project for patients' advocates in Europe.

\* EURORDIS- European Organisation for Rare Diseases

**Colin McKerlie** DVM, DVSc, MRCVS

Dr. McKerlie is a Senior Associate Scientist at The Hospital for Sick Children and Professor in the Department of Laboratory Medicine & Pathobiology at the University of Toronto. A Veterinary Pathologist and Phenogenomic Scientist, he is also Director of Research Partnerships at the Toronto Centre for Phenogenomics (TCP) and Staff Scientist with the Samuel Lunenfeld Research Institute of Mount Sinai Hospital.



His research focuses on developing and using mouse models to discover and understand the function of genes that cause disease in children and adults; particularly the comparative pathology and tissue changes related to genetic manipulation. Dr. McKerlie is project leader of the NorCOMM2 project, Canada's contribution to the International Mouse Phenotyping Consortium (IMPC) that aims to produce and phenotype up to 20,000 mutant mouse lines and make the repository of models and data publically available as a hypothesis-generating resource. The IMPC's mission is to get its data and resources into the hands of the research community to support their hypothesis-driven research, and generate collaborations where the mouse biology, production, phenotyping, and pre-clinical application knowledge and expertise it has can help. The NorCOMM2 project within the IMPC has established expertise in imaging and pathology-based phenotyping, particularly suited for characterization of embryo & neonatal lethal (congenital) phenotypes for comparative analysis to human disease.

**Pierre Meulien, Ph.D.**

President and CEO  
Genome Canada

Dr. Pierre Meulien was appointed President and CEO of Genome Canada in October 2010. Prior to this appointment, he served as Chief Scientific Officer for Genome British Columbia from 2007 to 2010. From 2002 to 2007, Dr. Meulien served as the founding CEO of the Dublin Molecular Medicine Centre (now Molecular Medicine Ireland) which linked the three medical schools and six teaching hospitals in Dublin to build a critical mass in molecular medicine and translational research. The Centre managed the Euro 45 Million "Program for Human Genomics" financed by the Irish government and was responsible for coordinating the successful application for the first Wellcome Trust funded Clinical Research Centre to be set up in Ireland. For over 20 years, Dr. Meulien has managed expert research teams with a number of organizations, including Aventis Pasteur in Toronto (Senior Vice President of R&D), and in Lyon, France (Director of Research). He also spent seven years with the French biotechnology company Transgene in Strasbourg, France as a research scientist and part of the management team. Dr. Meulien's academic credentials include a PhD from the University of Edinburgh and a post-doctoral appointment at the Institut Pasteur in Paris.



**Lucia Monaco** is the chief scientific officer of Fondazione Telethon since 2008. She joined Telethon in 2001 as research program manager and head of the Telethon Science Watch Office. Her responsibilities include management of the Telethon research portfolio and peer review process. In 2008, she promoted and managed the creation of the Telethon Network of Genetic Biobanks. She represents Fondazione Telethon in the IRDiRC Executive Committee.

She previously worked as group leader at the San Raffaele Scientific Institute in Milan and earlier she was senior researcher in the Molecular Biology Laboratory of Farmitalia Carlo Erba in Milan. Her main research interests were non-viral gene therapeutic approaches for renal genetic diseases and glycosylation engineering for the production of therapeutic proteins in mammalian cells. Her work was supported by FP4, FP5 and FP6 EU funds, by the Italian Telethon Foundation and by the Italian Cancer Association and resulted in several publications in international journals.

She graduated in chemistry in 1979 at the University of Pavia and received her training in biochemistry at the University of Iowa in Iowa City and in molecular biology at the European Molecular Biology Laboratory in Heidelberg.



### **Lesley Murphy** Rare Voices Australia

Board of Directors/Secretary Lesley Murphy has over 25 years of experience working on various community projects ranging from Child Care Centre management, to school parents and citizens committees. She was the Community Support manager at Muscular Dystrophy WA for 7 seven years. She is the community representative on the Neuro-Muscular disease registry working group, and is a community representative on the National Health and Medical Research Council, Human Genetics Advisory Committee.



She represents Rare Voices Australia on the National Rare Diseases coordinating committee. She has also been appointed as a visiting research associate to the Centre of Comparative Genomics at Murdoch University.

In her professional capacity Lesley has worked as a Registered Nurse, Midwife and Primary School teacher. In 2012 she became a co-founder of Rare Voices Australia. A national company formed to become the unified voice for all Australians who live with a rare disease.

She has three adult sons, the youngest of whom has Duchenne Muscular Dystrophy. She lives in Fremantle, WA

### **Luigi Naldini**

*Professor of Cell and Tissue Biology and Professor of Gene and Cell Therapy, "Vita Salute San Raffaele" University School of Medicine, Milan, Italy*

*Scientific Director, San Raffaele Telethon Institute for Gene Therapy (HSR-TIGET), Milan, Italy*

*Director, Division of Regenerative Medicine, Stem Cell and Gene Therapy, San Raffaele Scientific Institute, Milan, Italy*

*Member of Strategic Committee for Research, San Raffaele Institute, Milan, Italy*



Inventor of 7 granted international patents and 9 pending. These include a cornerstone patent on lentiviral vector technology owned by the Salk Institute and a family originator patent owned by Cell Genesys. Intellectual property recently generated at the San Raffaele Institute covers bidirectional vectors for coordinate gene expression, micro-RNA regulated vectors, angiogenic monocytes and macrophage miRNA, tolerogenic and cell reprogramming vectors, site-specific integration.

Has published 183 papers in international scientific journals (Total Impact Factor 1,895.385 based on Journals IF 2011, with average I.F.= 10.53 per paper). Overall, his papers have been cited >19,797 times since 1996 (as of February 2013). Scopus "h" index: 66.

**Katherine Needleman** is a Health Science Administrator in the Office of Orphan Products Development (OOPD) where she serves as the Director for the Orphan Products Grants Program with responsibility for management of the \$14 million OOPD extramural research budget. She works closely with project officers, researchers, and organizations to advance promising medical products to market approval, to increase the publication of significant findings in the scientific literature, and to oversee the responsible use of federal funds for clinical research. She participated in developing the Common EMA/FDA Application for Orphan Medicinal Product Designation and currently chairs the Standard Operating Procedures and Policies committee in OOPD.



Ms. Needleman joined FDA, CBER, Office of Therapeutics Research and Review in 2002 as a Consumer Safety Officer/Regulatory Project Manager in the area of therapeutic proteins. She also served as a Regulatory Project Manager in the Division of Neurology Products, Office of Drug Evaluation I, Office of New Drugs, CDER. As a Regulatory Project Manager, Ms. Needleman served as an administrative and regulatory expert for the review process from pre-IND/discovery through post-marketing approval, labeling chair/reviewer, and mentor for newly hired staff. She was the electronic submissions coordinator to the division and participated in user testing for several electronic databases FDA uses. She has also served as a member of many workings groups such as the Good Review Management Practices Process Improvement Team and the team for the eCTD viewer tool evaluation initiative.

Ms. Needleman received her BA degree with a double major in Biochemistry and Mathematics from Bowdoin College, Brunswick, Maine in 2000, graduating Summa Cum Laude and Phi Beta Kappa, and she received her Masters degree in Pharmacology and Molecular Sciences from the Johns Hopkins University School of Medicine in Baltimore, MD in 2002. In 2005, she obtained the Regulatory Affairs Certification (RAC) from the Regulatory Affairs Professional Society (RAPS). In 2012, Dr. Needleman was awarded a Ph.D. in Experimental and Clinical Pharmacology from the University of Minnesota.

### **David A Pearce**

Vice President, Research Sioux Falls Region, Sanford Health  
Director, Sanford Childrens Health Research Center, Sanford Research USD  
Professor, Department of Pediatrics  
Sanford School of Medicine of the University of South Dakota



Dr. Pearce oversees a national registry for rare diseases known as the Coordination of Rare Diseases at Sanford (CoRDS). He is Vice President for Research and Director of the Sanford Childrens Health and Research Center in Sioux Falls South Dakota. He completed his undergraduate Bachelors of Science Degree with Honors in Biological Sciences at Wolverhampton Polytechnic in 1986. He gained his Ph.D in 1990 at the University of Bath, UK, and did postdoctoral training at the University of Rochester, USA and Oxford University, UK.

Dr. Pearce heads the leading lab in juvenile Batten disease research. He has been researching Juvenile Neuronal Ceroid Lipofuscinosis (Batten Disease) since 1997. He is responsible for the establishment of the Batten Disease Diagnostic and Clinical research Center at the University of Rochester School of Medicine (<http://dbb.urmc.rochester.edu/labs/pearce/bddcrc/index.htm>). His research has lead to the first clinical trial for juvenile Batten Disease. He has published over 70 research papers on Batten disease.

**Patricia Reilly** qualified as a veterinary surgeon from University College Dublin in 1996, and worked in mixed clinical practice until 2001, when she joined the Irish Department of Agriculture, Fisheries and Food. In 2004 she joined the Irish Embassy in Warsaw as Ireland's first Agricultural Attaché to Poland. On return to the Department of Agriculture in 2008, she re-joined the National Disease Control Centre, where her work involved veterinary international trade policy and contingency planning.



Patricia is a graduate of the King's Inns, Dublin, and other academic qualifications include an MSc in European Food Regulation and a Diploma in European Law from the Law Society of Ireland.

Patricia joined the Cabinet of Commissioner Máire Geoghegan-Quinn in February 2010, and is responsible for the health and consumer protection and bioeconomy policy areas, as well as communication and Joint Research Centre coordination.

Prof. Dr. **Olaf Rieß**

**Position** : Head, Institute of Human Genetics, Full Professor, Medical doctor, Tübingen, Germany

**Research fields** : familial tumours ▪ neurological diseases ▪ mental retardations respectively syndromic diseases of children ▪ myopathys

#### **Scientific Career History**

1985 - 1987	Molecular Human Genetics, Research Assistant, Academy of Sciences, Berlin, GDR
1987 - 1990	Department of Human Genetics, non-tenured employee, Humboldt-University, Berlin, GDR
1990 - 1992	Department of Medical Genetics, Research Associate, UBC, Canada
1992 - 1999	Molecular Human Genetics, Senior Scientist, Ruhr-University Bochum, Germany
1999 - 2001	Associate Professor (C3), Head of the Department of Medical Genetics,
2001 - Present	Full Professor (C4) of Medical Genetics, Head of the Department Human Genetics, University of Tübingen



#### **Awards, Scholarships, Faculty Positions**

1990 - 1992 Fellowship of the MRC Canada  
Prize of the "Deutsche Heredoatixie Gesellschaft"  
Poster prize at the Movement Society Meeting, New York, USA

### Peter Robinson

Peter was trained as a physician and obtained certification in paediatrics ("Fachart") in 2000. He has additional training in mathematics and computer science, and has led the bioinformatics group at the Charité Institute for Medical Genetics since 2004, and is currently Professor for Medical Genomics at the Charité and also associated faculty at the Free University Berlin (Bioinformatics) and the International Max Planck Research School for Computational Biology and Scientific Computing. His research involves molecular biology work, including a number of projects on Marfan syndrome which identified GxxPG-containing fragments as a factor contributing to disease progression and the demonstration that treatment with an antibody against these fragments ameliorates disease manifestations in mice; genomics and next-generation sequencing, including the identification of a number of novel disease genes including PIGV, PIGO, and PGAP2; and bioinformatics, including 20 papers on bio-ontologies and the development of the Human Phenotype Ontology (HPO), which is now being used by many groups internationally for genotype-phenotype databases in human genetics and beyond.



**Franz Schaefer**, M.D. is Professor of Pediatrics and Chief of the Pediatric Nephrology Division at Heidelberg University Hospital. Dr. Schaefer received his M.D. in 1986 at Würzburg University Medical School. He performed research scholarships at the Institute of Child Health, London, the University of Virginia and Stanford University.

His research interests include the genetic basis of congenital and hereditary kidney diseases, the physiopathology of kidney disease progression, and the cardiovascular, metabolic and endocrine consequences of pediatric chronic kidney disease.

In clinical research, he conducted numerous collaborative clinical trials and established several international consortia including the European Study Group for Progressive Chronic Kidney Disease in Children (ESCAPE), the International Pediatric Dialysis Network (IPDN) and the E-Rare-funded PodoNet Project for Research into Steroid Resistant Nephrotic Syndrome.

Dr. Schaefer currently serves as Secretary of the ERA-EDTA Workgroup for Inherited Kidney Diseases and as Coordinator of the European pediatric renal replacement therapy registry. His most recent accomplishment is the coordination of the EU FP7 research project EURenOmics, in which high-throughput research approaches will be applied to rare kidney diseases.

Dr. Schaefer has published more than 350 articles and book chapters, including publications in prestigious journals such as the New England Journal of Medicine, Nature Medicine, Nature Genetics, the Lancet, the Journal of Clinical Investigation and Circulation. He co-edited the standard textbooks "Comprehensive Pediatric Nephrology" and "Pediatric Dialysis".

Dr. Schaefer received several prestigious awards for innovative research including the Recklinghausen Prize and the IBM Faculty Award. Dr Schaefer is a current council member of the International Pediatric Nephrology Association (IPNA), member of several editorial boards and associate editor of two leading nephrology journals.



**Hans GCP Schikan**, PharmD

Hans Schikan is CEO of Prosensa, an innovative Dutch biopharmaceutical company focusing on the discovery, development and commercialization of novel treatments for rare diseases like Duchenne muscular dystrophy, myotonic dystrophy and Huntington's disease, using its RNA modulation platform. In 2009 Prosensa announced a key agreement with GlaxoSmithKline for part of its Duchenne compounds at a value of nearly USD 700 million. Before joining Prosensa, Hans worked at Genzyme for five years in various executive roles, including as Vice President for Global Marketing and Strategic Development of Genzyme's portfolio of products for rare genetic diseases. Prior to Genzyme, he spent 17 years at Organon, both at corporate level and in country operations which included assignments in Asia and Europe. Next to his role at Prosensa, Hans is currently Executive Board Member of the Dutch Top Institute Pharma, Non-executive Director of Sobi (Swedish Orphan Biovitrum) and Member of the Core Team of the Dutch Top Sector Life Sciences & Health. He is also past Chairman of Nefarma, the Dutch Association of Research Based Pharmaceutical Industry. He has a PharmD from Utrecht University. Hans has given numerous presentations on orphan drugs, rare diseases and innovation.

**Tsveta Schyns-Liharska**, PhD

Tsveta is molecular geneticist and consultant in research management and communication.

Tsveta is the founder of the European Network for Research in Alternating Hemiplegia, ENRAH (since 2003) and of the European Network for Rare Paediatric Neurological Disorders, nEUroped (since 2007). Since 2008 she serves as member of the Paediatric Committee representing rare disease patient community and Eurordis at the European Medicines Agency, London.

Before her current role, Tsveta pursued an academic career at the Free University of Amsterdam, The Netherlands. She was trained in Molecular Biology at the University of Sofia, Bulgaria. She specialized and conducted her post-graduate studies at the University of Wageningen, The Netherlands where she received her doctoral degree in Genetics in 1998.



Dr **Michael Shy** is Professor of Neurology, Pediatrics and Neuroscience at the Carver College of Medicine at the University of Iowa. He is PI of the Inherited Neuropathy Consortium (INC) Rare Disease Clinical Research Consortium (RDCRC) of the Rare Disease Clinical Research Network (RDCRN). He is also Chair of the Medical Advisory Board of the Charcot Marie Tooth Association (CMTA). He directs one of the largest clinics for patients with inherited neuropathies. He has served on the Medical Advisory Committee (MAC) of the Muscular Dystrophy Association since 2002 and is President Elect of the Peripheral Nerve Society.



Dr **Nicolas Sireau** is Chairman and CEO of the AKU Society, a medical charity that works to find a cure for and support patients with AKU, which affects his two sons. AKU (short for Alkaptonuria) is a monogenic disease caused by a missing enzyme, leading to the accumulation of a substance called homogentisic acid at 2,000 times the normal rate. This acid binds to cartilage and bone, turning them black – hence it is also called Black Bone Disease.

The AKU Society is a fast growing international patient movement. We work with AKU research teams, clinical centres, biotech and pharma partners across the world. We launched in November 2012 a major five-year programme of international clinical trials for a promising new treatment called Nitisinone as part of an EC-funded consortium. The website of the AKU Society is [www.akusociety.org](http://www.akusociety.org).



Dr Sireau is also the Co-founder and Chairman of Findacure ([www.findacure.org.uk](http://www.findacure.org.uk)), a foundation that seeks to change conventional thinking about rare diseases by funding scientific research into fundamental diseases: rare diseases that are gateways to understanding common diseases.

**Sharon F. Terry** is President and CEO of Genetic Alliance, a network of more than 10,000 organizations, of which 1,200 are disease advocacy organizations. Genetic Alliance enables individuals, families and communities to reclaim their health and become full participants in translational research and services.

She is the founding CEO of PXE International, a research advocacy organization for the genetic condition pseudoxanthoma elasticum (PXE). As co-discoverer of the gene associated with PXE, she holds the patent for ABCC6 to act as its steward and has assigned her rights to the foundation. She developed a diagnostic test and conducts clinical trials.



Terry is also a co-founder of the Genetic Alliance Registry and Biobank. She is the author of more than 100 peer-reviewed articles. In her focus at the forefront of consumer participation in genetics research, services and policy, she serves in a leadership role on many of the major international and national organizations, including the Institute of Medicine Science and Policy Board, the IOM Roundtable on Translating Genomic-Based Research for Health, the National Coalition for Health Professional Education in Genetics Board, the International Rare Disease Research Consortium Interim Executive Committee and as a member of the Board of eSpRare Foundation. She is on the editorial boards of several journals. She was instrumental in the passage of the Genetic Information Nondiscrimination Act. In 2005, she received an honorary doctorate from Iona College for her work in community engagement; the first Patient Service Award from the UNC Institute for Pharmacogenomics and Individualized Therapy in 2007; the Research!America Distinguished Organization Advocacy Award in 2009; and the Clinical Research Forum and Foundation's Annual Award for Leadership in Public Advocacy in 2011. In 2012, she became an honorary professor of Hebei United University in Tangshan, China, and also received the Facing Our Risk of Cancer Empowered (FORCE) Spirit of Empowerment Advocacy Award. She was named one of FDA's "30 Heroes for the Thirtieth Anniversary of the Orphan Drug Act" in 2013. She is an Ashoka Fellow.

**Chris Van Geet** graduated at the University of Leuven as a candidate in psychology in 1984 and doctor in medicine in 1986. She started her training in pediatrics in 1986 (till 1993). From 1986 -1991, she joined the "Platelet Unit" of Prof Vermylen in the Center for Thrombosis and Haemostasis (head: Prof. Verstraete/D. Collen) as a junior scientist of the National Fund for Scientific Research. In 1992, she received the degree of "Higher Education in Medicine" with the PhD thesis on Signal transduction in human endothelial cells and prostacyclin production in vivo in men. After finishing her training in paediatrics and in paediatric haematology-oncology, she worked as a clinician in the Department of Paediatrics (General Paediatrics and Paediatric Hematology) of the University Hospital Leuven. Since 1999, she became a Senior Clinical Investigator of the Fund for Scientific Research (FWO, Flanders) and returned part-time to the research team of Prof. Vermylen, where she became involved in the (patho)physiological study of blood platelet function and formation. Platelet genesis with regulation of megakaryocyte maturation and differentiation is studied both at the fundamental level and in patients with hereditary thrombocytopenias. She has co-authored over 90 research papers. She is also PI of the Bridge consortium (Cambridge). In 2005 she was appointed Head of the Paediatric Department of the University hospital. Her clinical specializations are non-oncological haematology and haemostasis in paediatrics. Since 2005, she is full professor at the faculty of Medicine of the University of Leuven, with teaching responsibilities in haemostasis, paediatrics and biomedical ethics.



**Philip J. Vickers, Ph.D.**, is Senior Vice President and Global Head of Research and Development at Shire Human Genetic Therapies (HGT), a business unit of Shire plc. Dr. Vickers has over 22 years of global experience in the pharmaceutical industry, encompassing both big pharma and biotechnology companies. Dr. Vickers holds a Ph.D. in Biochemistry from the University of Toronto, and a Bachelor of Science degree in Applied Biochemistry from the University of Salford, Manchester. Dr. Vickers was also a Visiting Fellow at the National Cancer Institute in Bethesda, Maryland, where he studied multi-drug resistance in breast cancer. His first industry experience was at Merck Frosst in Montreal, where his teams developed cox-2 and leukotriene synthesis inhibitors. Dr Vickers then spent 13 years at Pfizer in a number of R&D leadership positions of increasing responsibility in the UK and US. He was then appointed Senior Vice President and US Head of Research at Boehringer-Ingelheim Pharmaceuticals, where he had responsibility for the Ridgefield CT research site, which focused on the development of small molecules and biologics in the areas of cardiovascular and immunology & inflammation. Immediately prior to joining Shire, Phil was Chief Scientific Officer and President of Resolvix Pharmaceuticals.



**Dr. Lu Wang** graduated from Sichuan University, China, in 1984, with the highest honor and a degree of Bachelor in Genetics. She received her degree of Masters in Genetics from Chinese Academy of Science in 1987, and her Ph.D. in microbiology from Cornell University in 1995. She performed her postdoctoral research at The Rockefeller University studying transcriptional regulation of B-lymphocyte development. From 1999 to 2006, Dr. Wang worked for the Monsanto Company, Third Wave, and then Pel-Freez /Dynal, focusing on molecular diagnostics. During her tenure at Pel-Freez at head of Research and Development and eventually Chief Operations Officer, she led the development and commercialization of a number of FDA 510(k)-cleared molecular products serving tissue/organ transplant diagnostics and immunogenetics research. Dr. Wang joined National Human Genome Research Institute (NHGRI), National Institutes of Health (NIH) in 2006. As Program Director in the Division of Genome Sciences, her main responsibility is managing genome sequencing programs, including the Centers for Mendelian Genomics Program (<http://www.mendelian.org/>). Dr. Wang is the author of a number of research and review articles on transcriptional regulation, as well as on molecular diagnostic assay development.



Professor **Tarun Weeramanthri** is Executive Director, Public Health and Clinical Services Division, and Chief Health Officer in Western Australian Health Department. He is a specialist physician in both public health and internal medicine. His interests are in the contribution public health can make to Aboriginal health improvement, health services research, use of spatial technologies in health, promoting a public health approach to rare diseases, use of new social networking technologies in public health, and the implementation of health policy.



**Terry-Lynn Young**, BSc (Hons), MSc, PhD  
Discipline of Genetics, Faculty of Medicine  
Memorial University, St. John's, Newfoundland and Labrador, Canada

Terry-Lynn Young is a human molecular geneticist. She completed a Ph.D. in Medicine at Memorial University (St. John's NL, Canada) in the laboratory of Dr. William Davidson, where she carried out positional cloning studies on families with a rare form of kidney disease known as Bardet-Biedl syndrome. She then moved to the laboratory of Dr. Mary-Claire King in the Department of Genome Sciences at the University of Washington (Seattle, USA) where she identified several novel genes underlying hereditary hearing loss. Dr. Young is currently an Associate Professor with expertise in gene discovery of rare diseases in Newfoundland's genetically isolated population and currently leads an interdisciplinary team in translational research for lethal cardiomyopathies. In 2009, Dr. Young won the President's Award for Outstanding Research for her contribution to identifying novel genes of medical importance to Newfoundland and Labrador and received a community recognition award from the Canadian Hard of Hearing Association-NL the same year. Dr. Young currently serves as director on several local, regional and national scientific bodies including the Research and Development Corporation of Newfoundland (RDC), the Beatrice Hunter Cancer Research Institute (BHCRI) and the Institute of Genetics Advisory Board for the Canadian Institutes of Health Research (CIHR).



**Massimo Zeviani** graduated in Medicine at the University of Padua. He specialised in Endocrinology and Neurology, and obtained a PhD in Genetics at the University René Descartes in Paris. In 1984, he moved to Columbia University, New York, where he worked as a post-doc for five years with Billi Di Mauro on the biochemical and molecular definition of respiratory chain disorders.

In 1990, he became Assistant in Neurology at the Department of Biochemistry and Genetics of the Istituto Neurologico “C. Besta” in Milan where he organized a laboratory of molecular biology on mitochondrial disorders. In 2001 he became the director of the Unit of Molecular Neurogenetics and, in 2011 he became the director of the Department of Molecular Medicine of the same Institute. In January, 2013, he was appointed Director of the MRC Mitochondrial Biology Unit in Cambridge, UK.

Author of  $\approx 300$  scientific publications in peer-reviewed journals (H index 63), he has identified and characterized numerous disease genes associated with OXPHOS defects, contributing to the elucidation of the molecular pathogenesis of mitochondrial disorders. More recently he focused on the development of therapeutic approaches to treat these conditions in experimental models and, eventually, patients.





