



Rare Diseases and Big Data – Hopes, Opportunities and Challenges

In the past years, considerable amounts of data have been generated creating new opportunities for evidence collection in the area of health to find better and more effective therapies for severe and life-threatening diseases. There is a growing international trend to support data-sharing initiatives in order to improve science and transparency.

Rare diseases pose a major challenge to patients and families, to physicians and researchers, and to society as a whole. A disease is considered rare when not more than one person per 2,000 people is affected. Data sharing presents a new and promising opportunity for research in the field of rare diseases.

This workshop is dedicated to present the opportunities and potential pitfalls of rare diseases research and big data with a keynote speech and a panel discussion in German on the first day, followed by individual presentations on the second day.

This meeting is a cooperation between the Medical University of Vienna, the UNESCO Chair on Bioethics at the Medical University of Vienna and the Ludwig Boltzmann Institute for Rare and Undiagnosed Diseases (LBI-RUD) in Vienna/Austria (www.rare-diseases.at).

Please register until 20th February 2018 via josephinum@meduniwien.ac.at



Josephinum, Collections of the Medical University of Vienna
Währinger Straße 25, 1090 Vienna



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22nd – 23rd February 2018

Josephinum, Collections of the Medical University of Vienna

www.meduniwien.ac.at

In collaboration with



Ludwig Boltzmann Institute
Rare and Undiagnosed Diseases

Ce-M-M- Research Center for Molecular Medicine of the Austrian Academy of Sciences



St. Anna Kinderkrebsforschung
CHILDREN'S CANCER RESEARCH INSTITUTE



United Nations
Educational, Scientific and
Cultural Organization



UNESCO Chair
on Bioethics,
Medical University of Vienna



MEDICAL UNIVERSITY
OF VIENNA

22.02.2018

Venue: Josephinum Reading Room

5.00 – 5.05 pm

Welcome

Christiane Druml, Medical University of Vienna,
UNESCO Chair on Bioethics, LBI-RUD

5.05 – 5.50 pm

Keynote lecture

Meine Daten, Deine Daten - Daten sind für alle da? *)

Christiane Woopen, University of Cologne, European Group on Ethics

5.50 – 6.40 pm

Panel discussion

'Big Data' im Kontext von biomedizinischer Forschung: Herausforderungen, Chancen und mögliche Gefahren *)

Christiane Druml, Medical University of Vienna,
UNESCO Chair on Bioethics, LBI-RUD

Kaan Boztug, LBI-RUD

Boris Marte, Father of two children with the rare blood disease Diamond-Blackfan anemia/DBA

Peter Bauer, Medical University of Vienna

Host: Ulrike Weiser, Austrian daily newspaper
"Die Presse"

*) in German

23.02.2018

Venue: Josephinum Sala Terrena

8.30 – 9.30 am

Registration

9.00 – 10.30 am

Session 1

Scientific/Technical background and opportunities

Chair: Vanja Nagy, LBI-RUD

What if ... We Will All Know Each Other's Genomes?

Christoph Bock, Medical University of Vienna, CeMM,
LBI-RUD

RareConnect - How a Social Network for People Living with Rare Diseases is Moving Towards Patient Self-phenotyping in Collaboration with the Research and Clinical Community

Denis Costello, EURORDIS, The Voice of Rare Disease Patients in Europe

Drawing Insights of Big Data: Electronic Health Records, Sharing Data and Methods for Predicting Rare Diseases

Nikolas Popper, TU Wien

The Politics of Knowledge and Ignorance in the Era of Big Data: A Social Science Perspective

Katharina Paul, University of Vienna

10.30 – 11.00 am

Coffee break

11.00 am – 12.30 pm

Session 2

Mapping different contexts and landscapes

Chair: Christiane Druml, Medical University of Vienna,
UNESCO Chair on Bioethics, LBI-RUD

Governing Big Data and Rare Diseases

Paul Just, LBI-RUD

Who Owns My Data? About Biobanks, Researchers and Patients/Citizens

Michaela T. Mayrhofer, Medical University of Graz

Rare Diseases, Countless Vulnerabilities

Maria do Céu Patrão Neves, Universidade dos Açores

Genetics and the Culture of Deafness: Are They Compatible?

Karen B. Avraham, Tel Aviv University

12.30 – 1.15 pm

Looking beyond: Experiences and visions of the future

Big Data and Rare Diseases: Why and How Bioethics Might Drive Policy Change

Hugh Whittall, Nuffield Council on Bioethics

Rare Diseases, Big Data and Solidarity – Considerations for the Future

Alena Buyx, Kiel University

Wrap-up and discussion

Speakers and panellists (in alphabetic order)

Karen B. Avraham is a faculty member of the Department of Human Molecular Genetics, holds the Drs. Sarah and Felix Dumont Chair for Research of Hearing Disorders and is Vice Dean at the Faculty of Medicine of Tel Aviv University. Karen Avraham was awarded the Bruno Memorial Prize, the TEVA Prize for Groundbreaking Research in the Field of Rare Diseases, and the Teva Founders Prize on Breakthroughs. She is President of the Scientific Committee of the Foundation for Hearing (Paris) and is a board member of EMBO and the Human Genome Organization. Prof Avraham has supervised 52 students toward Masters, PhD and MD-PhD degrees and has published 101 manuscripts, 37 reviews, and 17 book chapters.

Peter Bauer earned his PhD in Applied Mathematics from the Technical University of Vienna in 1970 and joined the Department of Medical Statistics/University of Vienna. In 1986, he was appointed as the Director of the Department of Medical Documentation and Statistics/University of Cologne, and from 1995 onwards he was head of the Department of Medical Statistics/University of Vienna until his retirement in 2010. Peter Bauer has published numerous original research papers on statistical methodology and cooperative research with medical scientists. His main research interest is in sequential and flexible (adaptive) designs, multiple inference, quality control and clinical epidemiology. He has been chief editor of Biometrical Journal and member of various scientific societies, editorial boards of scientific journals, in scientific advisory boards, in data safety monitoring boards and in committees of governmental regulatory institutions.

Christoph Bock is a principal investigator at the CeMM Research Center for Molecular Medicine of the Austrian Academy of Sciences, and group leader of the Bioinformatics group at LBI-RUD. His research focuses on dissecting the role of epigenetics in cancer and on developing high-throughput technologies for precision medicine. He is also a guest professor at the Medical University of Vienna, scientific coordinator of the Biomedical Sequencing Facility at CeMM, and adjunct group leader for bioinformatics at the Max Planck Institute for Informatics. He has received several research awards, including the Max Planck Society's Otto Hahn Medal (2009), an ERC Starting Grant (2016-2021), and the Overton Prize of the International Society of Computational Biology (2017).

Kaan Boztug joined the CeMM Research Center for Molecular Medicine of the Austrian Academy of Sciences as a Principal Investigator in 2011, and became director of the Ludwig Boltzmann Institute for Rare and Undiagnosed Diseases in April 2016. He is the head of the CeRUD Vienna Center for Rare and Undiagnosed diseases and the Jeffrey Modell Foundation Diagnostic and Research Center Vienna. Kaan Boztug holds a dual appointment as Associate Professor at the Department for Pediatrics and Adolescent Medicine at the Medical University of Vienna, and is a Consultant in Pediatric Hematology/Oncology and Head of Pediatric Immunology at St. Anna Children's Hospital Vienna. With his research team, he has made seminal contributions to the field of rare diseases and immunology including discovery and mechanistic dissection of a number of previously unknown disease entities, for which he has received numerous awards and prizes including, amongst others, an FWF START Prize (2012), an ERC Starting Grant (2013-), the Eva Luise and Horst Köhler Research Award for Rare Diseases and a Science Award of the Austrian Society for Pediatrics and Adolescent Medicine.

Alena Buyx, MD, PhD is Professor of Biomedical Ethics and Head of the Department of Biomedical Ethics at the Institute of Experimental Medicine, Christian-Albrechts-University of Kiel, Germany. She has previously held appointments at the University of Münster, Harvard University; and was Assistant Director of the Nuffield Council on Bioethics, London. Alena Buyx is a medical doctor with postgraduate degrees in philosophy and sociology. Her research spans the whole field of biomedical ethics, with a particular focus on questions of solidarity and justice in applied contexts such as public health, resource allocation, and Big Data and precision medicine. She is particularly keen on interdisciplinary approaches and collaborates regularly with clinical colleagues as well as with public health professionals, political and social scientists, philosophers, lawyers or health economists.

Denis Costello has worked with EURORDIS since 2008 as the organisation's Online Communications Manager. During that time he has led the launch and operation of RareConnect, a multilingual social network for patients and families living with rare diseases (www.rareconnect.org), and was part of the team that oversees the online awareness campaign around Rare Disease Day (last day of February; www.rarediseaseday.org). He is a regular speaker at international conferences, including international rare disease conferences (European Hematology Association, ECCO, Skeletal Care Academy). His responsibilities also include working with rare disease patient organisations internationally to build effective networks, raise awareness of rare diseases, and support patient-led initiatives that engage with the broader public. He enjoys putting a specific focus on the opportunities provided by online communication tools and social media. Prior career experiences include running his own technology firm while also presenting his favorite classical music on Irish national radio.

Christiane Druml was born in Vienna and studied law at the University of Vienna. She is currently holding the UNESCO Chair on Bioethics at the Medical University of Vienna and also serves as Director of Ethics, Collections and History of Medicine of the Medical University of Vienna. She is also key researcher of the Bioethics Research Group and Deputy Director of the LBI-RUD. 2007 she has been appointed President of the Austrian National Bioethics Commission by the Austrian Federal Chancellor. Furthermore she is a member of the Medical Council of the Republic of Austria and serves in many international functions.

Paul Just studied Political Science, Spanish and Macroeconomics at the University of Vienna, University of Essex and Georgetown University. Paul Just earned his MA degree in Political Science in 2009 and is PhD candidate at the Department of Science and Technology Studies at the University of Vienna (Ulrike Felt). As member of the Bioethics team led by Christiane Druml at the LBI-RUD he currently researches and explores the ethical, legal, and political aspects of rare and undiagnosed diseases in Austria and beyond.

Michaela Th. Mayrhofer is a political scientist and historian by training. She was educated in Vienna, Louvain-la-Neuve, Essex and Paris, earning her PhD from both the Ecole des Hautes Etudes en Sciences Sociales and the University of Vienna. She is a Research Fellow at the IFZ Graz and was a Member of the Editorial Board of the Austrian Journal of Political Science (ÖZP). Currently, she serves as Coordination and Policy Officer for the research infrastructure BBMRI-ERIC.

Vanja Nagy obtained her PhD with George W. Huntley at the Icahn School of Medicine at Mount Sinai, USA. For her postdoctoral training, she worked with Ivan Dikic at the Mediterranean Institute for Life Sciences, Croatia, and Josef Penninger at the Institute

for Molecular Biotechnology, Austria. She joined the Ludwig Boltzmann Institute for Rare and Undiagnosed Diseases in 2016, where she has established her own independent research group. Her group focuses on the identification and characterization of genetic underpinnings of rare neuronal disorders affecting both the central and peripheral nervous systems.

Maria do Céu Patrão Neves is Full Professor of Ethics, at the University of the Azores (Portugal), where she is also responsible for teaching and research on applied ethics (mainly bioethics). She was the consultant on Ethics of Life for the President of the Portuguese Republic, a Member of the National Ethics Committee, and of the Board of Directors of the International Association of Bioethics). She authored about 180 papers, 10 books, and edited 12 others, and is currently leading a 12 volume collection on Applied Ethics, and a research project on (bio)Ethics, Science and Society: Challenges for BioPolitics. She was a member of the European Parliament, from 2009 to 2014.

Katharina T. Paul obtained her PhD in social sciences at the University of Amsterdam and has since specialized in comparative policy analysis and interpretive methods. She holds an Elise Richter Fellowship (FWF - Austrian Science Fund) at the University of Vienna (Department of Political Science) and acts as work package leader in an H2020 research project on science and health diplomacy. Her current research focuses on health policy, specifically vaccination, in Austria and beyond.

Nikolas “Niki” Popper studied Mathematics, Philosophy and Jazz Theory in Vienna, Barcelona (Spain) and Moscow, Idaho (US) and received his ScD (Dr.techn.) from the Technical University of Vienna (TU Wien). He worked as science editor and journalist and co-founded two successful companies. Niki Popper published and presented about 150 articles and talks in journals and at international conferences.

He is coordinator of COCOS “Centre for Computational Complex Systems” at TU Wien as well as chairman of DEXHELPP, the COMET K-Project (Decision Support for Health Policy and Planning: Methods, Models and Technologies based on Existing Health Care Data).

Hugh Whittall is the Director of the Nuffield Council on Bioethics, whose task is to identify and report on ethical questions raised by new developments in biological and medical research. Recent reports have covered areas including neurotechnology, emerging biotechnologies, children and clinical research, biodata and genome editing. Hugh was previously at the UK Department of Health, where he was involved with the preparation of the Human Tissue Act 2004 and the setting up of the Human Tissue Authority. He was also involved in end-of-life issues and transplantation policy. Prior to that Hugh Whittall spent three years at the European Commission in Brussels, involved in the funding and promotion of bioethics research, and he was for several years Deputy Chief Executive of the Human Fertilisation and Embryology Authority.

Christiane Woopen is Professor for Ethics and Theory of Medicine at the University of Cologne. There she is Executive Director of the Cologne Center for Ethics, Rights, Economics, and Social Sciences of Health (ceres). She is as well Head of Research Unit Ethics and vice-dean at the medical faculty. From 2012 to 2016 she was chair of the German Ethics Council and 2016 President of the Global Summit of National Ethics Committees. Furthermore, she was member of the International Bioethics Committee of UNESCO. Recently she was appointed Chair of the European Group on Ethics of Science and New Technologies, which advises the European Commission.