RARE DISEASES
REGISTRIES AND NATIONAL PLANS
AN INNOVATIVE SPECIALIZED WORKSHOP

7-8 April 2017, Kingdom of Bahrain
To mark the occasion of International Rare Disease Day 2017 and continuing the activities dedicated to increase the awareness for rare diseases, Arabian Gulf University/ Al Jawhara Center for Molecular Medicine invite you to join an innovative workshop on registries and national plans for rare disorders as a step forward towards international integration of GCC efforts and achievements in the field of rare diseases.

The event is the first of its kind to be organized in Bahrain and will gather international experts in the fields of genomics, scientific research, public health, and patient advocacy to discuss why and how rare diseases should be included in the health agendas as priority. Learning from European experiences and using as references their registries and national plans, the workshop goal is to contribute to initiate and grow in GCC the rare disorders health and research policies and to offer a better care for the affected patients and their families.

The workshop aims to provide stakeholders from GCC Ministries of Health, academia researchers, medical experts and doctors dealing in their practice with rare disorders, with a platform able to allow them to initiate registries and national plans for rare diseases, to develop and expand their research in order to contribute to a Rare Disease GCC network.

DR CRISTINA SKRYPNYK
Chair of the organizing committee
سجل الأمراض النادرة والخطط الوطنية
ورشة عمل تخصصية إبداعية

احتفالاً بمناسبة اليوم العالمي للأمراض النادرة لعام 2017، ومواصلة قيام الأنشطة التي تزيد من
توحية المجتمع بخصوص الأمراض النادرة، يدعوكم مركز الأميرته الجوهرة/ جامعة الخليج العربي
للإنضمام إلى ورشة العمل المبتكرة لتأسيس سجل دولي وعمل خطة وطنية خاصة للأمراض
النادرة كخطوة نحو التكامل الدولي لجهود دول مجلس التعاون الخليجي والإنجازات المرتبطة
بتاريخ الأمراض النادرة.

تعتبر هذه الورشة هي الأولى من نوعها في مملكة البحرين حيث أنها تجمع بين التجارب
العالمية في مجال الجينوم، البحوث العلمية، الصحة العامة وحماية المرضى عن طريق مناقشة
سبب وكيفية تضمين الأمراض النادرة كأولويّة هامة في جدول أعمال الصحة من خلال التعلم
من التجربة الأوروبية في إنشاء السجل الخاص بالأمراض النادرة وتضمينها في الخطة الوطنية.
وهذا ما تسعى إليه الورشة لتحقيق النمو في تطوير السياسات التابعة لبحوث الأمراض النادرة و
تقديم الرعاية الأفضل للمرضى المصابين بالأمراض النادرة وذويهم.

تهدف الورشة أيضًا إلى تثمين الأقسام المطلوبة لمنظمة الصحة العالمية في دول مجلس التعاون
الخليجي، والباحثين الأكاديميين، و التقدماء الخبراء في هذا المجال أو العاملين فيه لبدأ وضع
سجلات وخطط وطنية خاصة بالأمراض النادرة وتوفير وزيادة عدد البحوث للمساهمة البيئية.

الدكتورة كريستينا سكريبنيك
رئيسة اللجنة المنظمة
RARE DISEASES REGISTRIES AND NATIONAL PLANS
AN INNOVATIVE SPECIALIZED WORKSHOP

RARE DISEASES IN GENOMIC ERA - A HEALTH PRIORITY

TOPICS COVERED:

- Rare Disorders in Genomic Era and impact into health care
- EU strategies on rare diseases
- National Plans for rare diseases
- Registries for rare diseases - Italy experience
- Best-practices for rare diseases and external quality assurance of genetic testing
- Pharmacoeconomics and orphan drug therapies
- CFTR2 a model for assignment of variant pathogenicity in rare diseases
- Orphanet: overview of its activities
- IRDC consortium and international collaborative research in rare diseases
- Integrated services for patients with rare diseases NoRo
- Rare diseases patient registry – Romanian experience
- National and international networking for patients with rare diseases
- Living together and learning together in rare diseases
- Precision medicine in Rare Diseases
- RD-connect.eu task-force
- Implementing the electronic pharmacogenomics assistant (ePGA) in rare diseases
- Cost-effectiveness in Rare Diseases
سجل الأمراض النادرة والخطط الوطنية
ورشة عمل تخصصية إبداعية
الأمراض النادرة في عصر الجينوم - أولوية صحية
المواضيع المطروحة:
الأمراض النادرة في عصر الجينوم وتأثيره في العناية الطبية
استراتيجيات الاتحاد الأوروبي في مجال الأمراض النادرة
الخطط الوطنية للأمراض النادرة
السجل الخاص بالأمراض النادرة- التجربة البيليطالية
أفضل الممارسات في الأمراض النادرة وضمان الجودة الخارجية في الاختبارات الجينية.
الاقتصاديات الصيدلانية وعلاجات الدوائية البيتينة
نموذج CFTR2 بديل لتعيين المرض النادر
شركة اورفانت: نظرة عامة عن أنشطتها و التعاونات الدولية في بحوث الأمراض النادرة.
جمعية IRDC و التعاونات الدولية في بحوث الأمراض النادرة.
خدمة متكاملة للمريض الذين يعانون من الأمراض النادرة-تجربة نورو
السجل الخاص بالأمراض النادرة- التجربة الرومانية
الشبكة العالمية والوطنية لمريض المصابين بالأمراض النادرة
نعشي وتتعلم معًا عن الأمراض النادرة
الطب الدقيق بالأمراض النادرة
فرق عمل جمعية الأمراض النادرة
تطبيق الاقتصاديات الصيدلانية المساعدة في الأمراض النادرة (ePGA)
تكلفة الفعالة في مجال الأمراض النادرة
RARE DISEASES REGISTRIES AND NATIONAL PLANS
AN INNOVATIVE SPECIALIZED WORKSHOP

PROGRAM

DATE: 7 APRIL 2017

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<tr>
<th>Time</th>
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<tr>
<td>8:30-9:00</td>
<td>Registration</td>
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<tr>
<td>9:00-9:15</td>
<td>Welcoming speech</td>
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<tr>
<td>9:15-9:45</td>
<td>Rare Disorders in Genomic Era and impact into health care</td>
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<td>9:45-10:30</td>
<td>EU strategies on rare diseases</td>
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<td>Coffee Break</td>
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<td>10:45-11:30</td>
<td>IRDC consortium and international collaborative research in rare diseases</td>
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<td>11:30-12:15</td>
<td>Integrated services for patients with rare diseases NoRo</td>
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<td>12:15-13:00</td>
<td>Precision medicine in Rare Diseases</td>
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<td>13:00-14:00</td>
<td>Lunch and prayer time</td>
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<td>14:00-14:45</td>
<td>National Plans for rare diseases</td>
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<td>14:45-15:30</td>
<td>CFTR2 a model for assignment of variant pathogenicity in rare diseases</td>
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<td>15:30-16:15</td>
<td>National and international networking for patients with rare diseases</td>
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<td>16:15-17:00</td>
<td>RD-connect.eu task-force</td>
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<td>17:00-17:30</td>
<td>Discussion</td>
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<td>1:15-14:00</td>
<td>Best-practices for rare diseases and external quality assurance of genetic testing</td>
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<td>16:30-17:00</td>
<td>Conclusion</td>
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SPEAKERS:

→ PROFESSOR MILAN MACEK JR., MD, PHD
  Head of National Coordination Centre for Rare Diseases Prague, Czech Republic
  Former President of European Society of Human Genetics
  European Commission Expert Group on Rare Diseases
  RD-connect.eu

→ DOCTOR DOMENICA TARUSCIO, MD, PHD
  Director of National Centre for Rare Diseases, Italy
  Former president of ICORD
  European Commission Expert Group on Rare Diseases
  RD-connect.eu

→ PSYCHOLOGIST DORICA DAN
  President of Romanian National Alliance for Rare Diseases
  National Council for Rare Disorders, Romania
  EURORDIS board officer

→ DOCTOR THEODORA KATSILA, PHD
  Greek National Genetic Database Consortium
  Pharmacogenomics and Personalized Medicine Group, Greece
  RD-connect.eu

→ DOCTOR CRISTINA SKRYPNYK, MD, PHD
  Consultant Medical Geneticist
  AlJawhara Center, Bahrain
  EURORDIS member
  Chair of Rare Disease Day campaign, Bahrain
Professor Milan Macek Jr. MD, DSc did his postdoctoral studies at Humboldt University Berlin then 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 is the chairman of the largest academic medical and molecular genetics institution in the Czech Republic and cochair of the National Cystic Fibrosis Centre. He is also the past President of the European Society of Human Genetics and past-board member of the European Cystic Fibrosis Society. He was member of the EUCERD.eu committee, and currently serves as an EC-appointed expert at the European Commission Expert Group on Rare Diseases. His department was designated by the Czech Ministry of Health as a National Coordination centre for rare diseases (www.nkcvo.cz) and serves as a “clearing centre” (Min Health Bulletin 4/2012) for dissemination of knowledge in rare disease-related genetics gathered within various international European projects related to cystic fibrosis, such as CF Thematic Network, EuroGentest, EuroCareCF, RD-connect.eu, Orphanet or Norway Grants to Central / Eastern Europe and the Middle Eastern diagnostic- research groups. Prof. Macek is also the Czech National coordinator of Orphanet and member of the Diagnostic Committee of the International Rare Disease Consortium (www.irdirc.org), his work has been cited more than 3000 times with H-index 35. His main research and clinical interest is molecular genetics/genomics in rare diseases, and how to bring genetics knowledge to the bedside.
DOCTOR DOMENICA TARUSCIO

DIRECTOR OF NATIONAL CENTRE FOR RARE DISEASES, ITALY
FORMER PRESIDENT OF ICORD
EUROPEAN COMMISSION EXPERT GROUP ON RARE DISEASES
RD-CONNECT.EU

Domenica Taruscio is the Director of the Italian National Centre for Rare Diseases at the Italian National Institute of Health (Istituto Superiore di Sanità, Rome, Italy). She holds a M.D., a specialization in Histopathology from the University of Bologna and a master in Bioethics, University Sapienza, Rome. She has carried out post-doctoral studies in Human Genetics at Yale University, CT-USA. She is the past-president of ICORD -International Conference for Rare Diseases and Orphan Drugs- Society, the member of the Italian Committee for Orphan Medicinal Products from European Medicines Agency EMA, European Rare Diseases Task Force, EUCERD-European Union Committee of Experts on Rare Diseases and of the European Expert Group and of the Health Research Advisory Group. Dr Taruscio is a member of the Interdisciplinary Committee of IRDiRC, the International Rare Diseases Research Consortium and is in the management board of Advisory European Molecular Genetics Quality Network, European Molecular Genetics Quality Network (EMQN) and the Advisory Board of Eurogentest (NoE). Dr Taruscio is the Scientific Coordinator of several EU projects on rare diseases and Work Package leader in other EU projects. Since 2001 she is the Scientific Coordinator of the bilateral Agreement ITALY/ Italian National Institute of Health- USA/NIH, Office for Rare Diseases Research and of the bilateral project “Undiagnosed rare diseases”. She has published more than 130 scientific publications on peer reviewed leading scientific journals. For decades, her efforts have been directed to complex challenges posed by rare diseases and has addressed them from various facets- science research, public health, training health professionals and empowerment of patients and their families.
PSYCHOLOGIST DORICA DAN

PRESIDENT OF ROMANIAN NATIONAL ALLIANCE FOR RARE DISEASES
NATIONAL COUNCIL FOR RARE DISORDERS, ROMANIA
EURORDIS BOARD OFFICER

Dorica Dan got her psychologist specialization from Spiru Haret University, Bucharest Romania, initiated RPWA-Romanian Prader Willi Syndrome Association in 2003, established RONARD - Romanian National Alliance for Rare Diseases through a project funded by CEE Trust in 2007 and Romanian Rare Cancers Association in 2011. She is the initiator of the National Plan for Rare Diseases in Romania and her efforts dedicated to people suffering by rare disorders lead in the opening of the first Centre for Information about Rare Genetic Diseases in Romania and in 2011, of the Pilot Reference Center for Rare Diseases “NoRo” through a project implemented in partnership with Frambu Norway and funded by Norway Grants. Today she is the President of the Romanian National Alliance for Rare Diseases-RONARD, President of the Romanian Association for Rare Cancers, the chair of the Romanian Prader Willi Association, Romania–RPWA and board member of International Prader Willi Syndrome Organisation IPWSO. She is coordinator of the Centre for Information about Rare Genetic Diseases and NoRo Center Romania. In recognition of her tremendous work and advocacy for people with rare disorders she was appointed as a member of National Council for Rare Disorders from Romanian Ministry of Health and member of European Commission, Experts Committee for Rare Disorders. She is elected board officer of EURORDIS and one of the EURORDIS EUROPLAN advisors. Dorica represents EURORDIS at International Conferences throughout Europe and beyond, and is part of the European Commission Expert Group on Rare Diseases the Work Package leader of the EUCERD Joint Action Working for Rare Diseases (EJA) Work Package 6 focusing on Specialized Social Services, and she is also a member of the European Council of National Alliances. Dorica’s daughter of 31 years old is suffering from PWS.
DOCTOR THEODORA KATSILA

GREEK NATIONAL GENETIC DATABASE CONSORTIUM
PHARMACOGENOMICS AND PERSONALIZED MEDICINE GROUP, GREECE
RD-CONNECT.EU

Theodora Katsila obtained her PhD in Chemistry from the University of Patras, Greece and the Biomedical Research Foundation of the Academy of Athens, Greece and continued her postdoctoral studies at Vall d'Hebron Institute of Oncology, Spain. She holds a MSc in Clinical Biochemistry and Molecular Diagnostics, University of Athens, Greece and a BSc in Biochemistry with a year in industry/research from Imperial College London, UK. Dr. Katsila has acquired a multidisciplinary expertise, investigating the underlying molecular mechanisms of diseases, focusing on drug research and development (Merck Sharp & Dohme Research Laboratories, UK) and biomarkers -cancer secretome. She has developed substantial pan-omics in vivo and in vitro skills and her research interest involved pharmacometabolomics-aided pharmacogenomics and personalized medicine. She currently serves as a senior research fellow and academic scholar in the Pharmacogenomics and Personalized Medicine group in the University of Patras, Greece. Dr. Katsila is an active member of the Greek National Genetic Database Consortium and RD-Connect. She has given numerous keynote and plenary lectures in international conferences as invited speaker in the field of genomics, personalized medicine and rare disorders. She serves Associate Editor and member of the editorial board of several scientific journals, she has been an external evaluator member of different research agencies.
Cristina Skrypnyk obtained her M.D. title from the University of Medicine and Pharmacy Iasi, Romania and was awarded later with a PhD degree in Medical Genetics. She got her Consultant title in Medical Genetics from the Romanian Board of Health. She had postgraduate international training in cytogenetics, molecular cytogenetics and genetic counselling and was fellow of prestigious genetics institutes in Germany (Institute of Clinical Genetics Dresden, Institute of Human Genetics Wuerzburg). With an experience of 20 years in diagnosis and management of genetic disorders she is a member of reputed international genetics societies (ESHG, ECA, ASHG, APSHG), as well as a volunteer of genetic disorders patients organizations (EURORDIS, Unique, Information Center for Rare Disorders Romania, Romanian National Association of Rare Disorders) and former member of EMEA and of the Romanian National Council for Rare Disorders. In 2010 she joined Arabian Gulf University Bahrain as Assistant Professor at the College of Medicine and Medical Sciences and Al Jawhara Center of Molecular Medicine as Consultant Medical Geneticist. She is actively involved in medical genetics university education, genetic research and public awareness of genetic diseases and genetic testing and is the initiator of the rare disease move in Bahrain with public campaigns organized in the month of February each year.
Workshop details:

Time: 7-8 April 2017, 9am-5 pm.
Venue: Arabian Gulf University/ Al Jawhara Center
Registration fee: BD 300
Places are limited – you will receive confirmation in order to attend.
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