WEDNESDAY, 19 OCTOBER

RDSA

Venue	Time		
Foyer	07:30 - 17:00	Registration	
Foyer	07:30 - 17:00	Exhibition and posters	
Auditorium	08:30 - 09:00	Opening Session	
PARALLEL ONE AUDITORIUM AND SIMONSBERG	09:00 - 10:00	VENUE: AUDITORIUM **Mitochondrial Disease** - Diagnosis of Mitochondrial Disease in the South African context - *Francois van der Westhuizen, North-West University, South Africa* - Mitochondrial Disease: an overview - *Izelle Smuts, University of Pretoria, South Africa*	VENUE: SIMONSBERG **Genetic Landscape in South Africa** - Training of health care professionals in medical genetics in South Africa - *Dave Woods, Perinatal Education Trust, South Africa* - Surveillance and monitoring of congenital disorders and patient registries in South Africa - *Vuyiswa Lebese, National Department of Health, South Africa* - The relevance of genetic counselling for rare genetic conditions - *Merlyn Glass, University of Witwatersrand, South Africa* - Zika Virus embryopathy: the world's second modern plague after HIV? - *Arnold Christianson, University of Witwatersrand, South Africa*
Foyer	10:00 - 10:30	Refreshment break	
PARALLEL TWO AUDITORIUM AND SIMONSBERG	10:30 - 12:30	VENUE: AUDITORIUM **Best practices in Transitional Medicine** - Rare disease transition and your best chance of getting it right - *Chris Hendriksz, Salford Royal NHS Foundation Trust, UK* **Paediatric rheumatology** - What is Rare in Paediatric Rheumatology? - *Chris Scott, University of Cape Town, South Africa* **Hyperammonemia and inborn errors of metabolism (IEMS): known and novel application for differential diagnosis** - *Marli Dercksen, North-West University, South Africa* VENUE: AUDITORIUM **Rare Tumours and Malignancies** - Executive function deficits in adults with Tuberous Sclerosis Complex: implications for clinical practice and clinical trials - *Loren Leclezio, University of Cape Town, South Africa* - Malignant Peripheral Nerve Sheath Tumours (MPNST) associated with Neurofibromatosis Type I: a case series and discussion of the implications for screening, referral and management in the Western Cape - *Henriette Burger, Stellenbosch University, South Africa*	VENUE: SIMONSBERG **Feeding support: a multidisciplinary approach** - Feeding matters: creating a seamless timeline from birth - *Claudia McGregor, The Children's Therapy Centre, South Africa* - Powerful implications of sensory development - *Tracey Venter, The Children's Therapy Centre, South Africa* - The impact of gastro-intestinal discomfort on feeding difficulties - *Inge Retief, Inge Retief & Associates, South Africa* **Supportive therapy - what is accessible in South Africa** - Early intervention and neuromusculoskeletal health - *Jacqui Bunge, The Children's Therapy Centre, South Africa* - How we move matters - *Andrea Fraser-Aldridge, The Children's Therapy Centre, South Africa* - Knowledge and understanding of parents of children affected with Sickle Cell Anaemia in Cape Town - *Katryn Fourie, Stellenbosch University, South Africa*
Foyer	12:30 - 13:30	Lunch	
PARALLEL THREE AUDITORIUM AND SIMONSBERG	13:30 - 15:00	VENUE: AUDITORIUM **Inborn Errors of Metabolism** - Rare diseases diagnosis: the importance of a diagnosis in genetic counselling and management - *Engela Honey, University of Pretoria, South Africa* - An overview of Lysosomal Storage Disease - *Karen Fieggen, University of Cape Town, South Africa* - A genetic approach for rare inherited metabolic diseases - *Louisa Bhengu, University of Witwatersrand, South Africa* - Psychiatric and neurological presentation of Inborn Errors - *Chris Hendriksz, Salford Royal NHS Foundation Trust, UK* - The search for biomarkers in Mitochondrial Disease: lessons learned from urine metabolomics investigation - *Roan Louw, North-West University, South Africa*	VENUE: SIMONSBERG **Palliative Care in South Africa** - A case based palliative care approach to children with rare diseases - *Julia Ambler, Umduduzi Hospice Care for Children, South Africa* - The role of palliative care in rare diseases - *Michelle Meiring, Paedspal Cape Town, South Africa* - Palliative care: the "magic wand" to quality of life for persons with disabilities - *Petra Burger, HPCA, South Africa*
Foyer	15:00 - 15:30	Refreshment break	

WEDNESDAY, 19 OCTOBER (CONTINUED)

	Venue	Time	
RDSA	Auditorium	15:30 - 17:30	**Ethics discussion** - Developing an essential package of health care for children in South Africa: what should be in the box? - *Anthony Westwood, Western Cape Department of Health, South Africa* - National Health Insurance (NHI) developments and rare diseases - *Rajesh Patel, Board of Healthcare Funders of Southern Africa, South Africa* - Patient reimbursement rights - *Peta-Anne Durrant, Elsabé Klinck Consulting, South Africa* - Panel discussion: newborn screening - **Moderator:** *Gareth Baynam, Western Australian Department of Health* **Panellists:** *Benjamin Djoudalbaye from the African Union Commission, Gary Kantor from Discovery Health, and Chris Vorster from PLIEM*
	Tasting Centre	17:30	Welcome Networking Reception *(sponsored by iConnect)*

THURSDAY, 20 OCTOBER

	Venue	Time	
ICORD	Foyer	07:30 - 17:00	Registration
	Foyer	07:30 - 17:00	Exhibition and posters
	Auditorium	08:30 - 09:00	Opening Session
	Auditorium	09:00 - 09:45	**Session 1. Introductory presentations** ***Chair:*** *John Forman, ICORD, New Zealand* A) Global overview of the rare disease field - *Stephen Groft, National Institutes of Health, USA* B) African overview of the rare disease field - *João L Carapinha, Carapinha & Company, South Africa*
	Foyer	09:45 - 10:15	Refreshment break
	Auditorium, Stellenberg and Simonsberg	10:15 - 12:15	**Session 2. Open discussion groups (break-out sessions)** - Group A: Diagnostics (*Facilitators: Ann Nordgren and James Chipeta*) - Group B: Congenital malformation *(Facilitators: Manuel Posada and Mike Urban)* - Group C: Global Rare Disease Policies and Programmes *(Facilitators: Stephen Groft and Domenica Taruscio)* - Group D: Access to treatment (*Facilitators: Emilio Roldan and Vinciane Pirard*) - Group E: Research (*Facilitators: Gareth Baynam and Petra Kaufmann*) - Group F: Improved quality of life (*Facilitators: Monika Esser and Kelly du Plessis*) - Group G: Patient organisations (*Facilitators: Durhane Wong-Rieger*)
	Foyer	12:15 - 13:15	Lunch
	Auditorium	13:15 - 15:00	**Session 3. Obtaining a diagnosis through available diagnostic procedures** **Introduction: report from Group A** ***Chairs:*** *Ann Nordgren, Karolinska University Hospital, Sweden and James Chipeta, University of Zambia School of Medicine, Zambia* - The NIH Undiagnosed Network: hope for more families and links to the International Rare Diseases Community - *Cynthia Tifft, National Institutes of Health, USA* - Behaviour and diagnostics in rare diseases - *Petrus de Vries, University of Cape Town, South Africa* - Diagnosis for Primary Immunodeficiency (PID) by accessing available resources in South Africa - *Monika Esser, Stellenbosch University, South Africa* - Newborn screening and status in South Africa - *Chris Vorster, The Potchefstroom Laboratory for Inborn Errors of Metabolism (PLIEM), South Africa* - Common inherited metabolic conditions in South Africa: diagnosing "rare" disease in genetically unique and understudied population groups - *Surita Meldau, University of Cape Town, South Africa*
	Foyer	15:00 - 15:30	Refreshment break
	Auditorium	15:30 - 17:15	**Session 4. Congenital malformations** **Introduction: report from Group B** ***Chairs:*** *Manuel Posada, Instituto de Salud Carlos III, Spain and Mike Urban, Stellenbosch University, South Africa* - Overview of congenital malformations and genetic mechanisms behind congenital malformations and intellectual disabilities - *Ann Nordgren, Karolinska University Hospital, Sweden* - The Western Australian Register of Developmental Anomalies and Australian Aboriginal Genomics and Phenomics - *Gareth Baynam, Western Australian Department of Health, Australia* - Congenital disorders and medical genetic services in South Africa - *Helen Malherbe, Genetic Alliance South Africa, South Africa* - Spectrum of congenital anomalies among newborns from selected Sub-Saharan African tertiary hospitals: focus on Zambia - *James Chipeta, University of Zambia School of Medicine, Zambia*
	Simonsberg	17:15 - 19:15	**Genetic Alliance South Africa Forum: building unity in the human genetics community** *(For more information: chair@geneticalliance.org.za)*
	Auditorium	17:30 - 19:30	**ICORD General Assembly** - *ICORD members only*

FRIDAY, 21 OCTOBER

	Venue	Time	
ICORD	Foyer	07:30 - 17:00	Registration
	Foyer	07:30 - 18:00	Exhibition and posters
	Auditorium	08:30 - 10:30	**Session 5. Global rare disease policies and programmes** **Introduction: report from Group C** ***Chairs:*** *Domenica Taruscio, Instituto Superiori di Sanitá, Italy and Stephen Groft, National Institutes of Health, USA* - UN Committee for Rare Diseases - *Anders Olauson, Ågrenska Foundation, Sweden* - International Rare Diseases Research Consortium (IRDiRC) - *Christopher Austin, National Institutes of Health, USA* - RARE-Bestpractices: a platform for sharing best practices for the management of rare diseases - *Domenica Taruscio, Istituto Superiori di Sanità, Italy* - A global approach to patient registries - *Manuel Posada, Instituto de Salud Carlos III, Spain* - Rare Diseases International - *Durhane Wong-Rieger, Canadian Organization for Rare Disorders, Canada* - Analysis of rare disease global policies and programmes to advance access to care and treatment - *Safiyya Dharssi, Pfizer, USA*
	Foyer	10:30 - 11:00	Refreshment break
	Auditorium	11:00 - 12:45	**Session 6. Access to treatment** **Introduction: report from Group D** ***Chairs:*** *Emilio Roldan, SLADIMER, Argentina and Vinciane Pirard, Genzyme, Belgium* - Closing the gap: the World Federation of Hemophilia's experience in responding to the challenges of a global rare disease - *Alain Weill, World Federation of Hemophilia, Canada* - Logistical aspects of access to treatment - *Jason Blackman, Marken, South Africa* - The need for innovative thinking to treat rare diseases in Africa - *Kelly du Plessis, Rare Diseases South Africa, South Africa* - GSK's graduated approach to patents and intellectual property to widen access to medicines in the world's poorest countries - *Rudy Onia, GSK, South Africa* - The difficulties in receiving access to high priced and orphan drugs: some thoughts - *Marc Blockman, University of Cape Town, South Africa*
	Foyer	12:45 - 13:45	Lunch
	Auditorium	13:45 - 15:45	**Session 7. Research and research funding** **Introduction: report from Group E** ***Chairs:*** *Petra Kaufmann, National Institutes of Health, USA and Gareth Baynam, Western Australian Department of Health, Australia* - Gaining access to available NIH rare diseases and translational research resources - *Christopher Austin, National Institutes of Health, USA* - Catalysing progress from discovery to health benefits through research collaboration - *Petra Kaufmann, National Institutes of Health, USA* - Applications to rare diseases from the H3 Africa Research Project - *Raj Ramesar, University of Cape Town, South Africa* - H3Africa BioNet and opportunities for rare diseases research - *Judit Kumuthini, University of Cape Town, South Africa* - In vitro assays for characterisation of novel rare genetic variants - *Maja Stojiljkovic, University of Belgrade, Serbia* - The NCATS Rare Diseases Clinical Research Network - *Rashmi Gopal-Srivastava, National Institutes of Health, USA*
	Auditorium	15:45 - 16:45	**Session 8. Panel discussion - "Problem Board"** - Panel to be confirmed
	Foyer	16:45 - 18:00	**Session 9.** Poster Session with refreshments
	Banqueting Courtyard	19:00	Conference Dinner

SATURDAY, 22 OCTOBER

	Venue	Time	
ICORD	Foyer	07:30 - 13:00	Registration
	Foyer	07:30 - 13:00	Exhibition and posters
	Auditorium	08:30 - 10:30	**Session 10. Improved quality of life** **Introduction: report from Group F** ***Chairs:*** *Monika Esser, Stellenbosch University, South Africa and Kelly du Plessis, Rare Diseases South Africa, South Africa* - Overcoming unmet social and daily life needs of people living with a rare disease - *Raquel Castro, EURORDIS, France* - Albinism: the stigma and what can be done - *Narcisse Kimbassa, Western Cape Albinism Hypo-Pigment Foundation, South Africa* - Measuring the quality of life in patients with rare diseases and setting pragmatic treatment goals - *Chris Hendriksz, Salford Royal NHS Foundation Trust, UK* - Quality of life from a parent's perspective: Sam's story - *Melissa Platt, South Africa* - A program of the National Center for Advancing Translational Sciences: the Genetic and Rare Diseases Information Center (GARD): 13 years of providing access to genetic and rare diseases information - *Janine Lewis, GARD, USA*
	Foyer	10:30 - 11:00	Refreshment break
	Auditorium	11:00 - 12:30	**Session 11. Patient organisations: a panel discussion** **Introduction: report from Group G** ***Chair:*** *Durhane Wong-Rieger, Canadian Organization for Rare Disorders, Canada* - Patient organisations in Kenya - *Christine Mutena, Stepping Stones, Kenya* - Botswana Organisation for Rare Diseases (BORDIS) - what it's all about - *Eda Selebatso, BORDIS, Botswana* - Overview of Genetic Alliance South Africa - *Helen Malherbe, Genetic Alliance South Africa, South Africa* - Fondation Internationale Tierno et Mariam: who they are and what they do - *Hawa Dramé, FITIMA, Burkina Faso* - Overview of Rare Diseases South Africa - *Kelly du Plessis, Rare Diseases South Africa, South Africa*
	Auditorium	12:30 - 13:00	**Conference closing** - Benjamin Djoudalbaye, *African Union Commission, Ethiopia* - Kelly du Plessis, *RDSA, South Africa* - John Forman, *ICORD, New Zealand* and Manuel Posada, *ICORD, Spain* - Passing the baton: ICORD 2017, Beijing, *Rachel Yong and Kevin Huang, CORD, China*
	Simonsberg	14:00 - 17:00	**Capacity Buiding Workshop for Health care Based NPOs - a RDI / RDSA initiative** - Pre-registration required, please enquire at the REGISTRATION AND INFORMATION desk - Email admin@rarediseases.co.za for more information