RARE X PROGRAMME

DAY 1 Wednesday, 14 February 2024

08:30 - 10:00	SESSION 1 - Venue: Kgotla 2 Policy: Looking towards a brighter future	
08h30 - 08h45	Welcome and introduction Ms Kelly du Plessis Rare Diseases South Africa	
08h45 - 09h10	Address by National Department of Health	
09h10 - 09h30	Commitment to the rare disease and congenital disorder community Dr Praveena Sukhraj-Ely National Department of Women, Youth and Persons with Disabilities (DWYPD)	
09h30 - 10h00	Living with SMA while managing accessibility and the understanding of what that means Ms Kerry Walsh Kerry Walsh Trust	
10h00 - 10h20	Tea/Coffee Break	
10h20 - 12h15	SESSION 2 - Venue: Kgotla 2 Global perspectives on policies relating to access	
10h20 - 10h40	Defining rare: What does it mean to be "rare"? Associate Prof Helen Malherbe Rare Diseases South Africa	
10h40 - 11h00	World Health Organization (WHO) commitment to rare diseases WHO Representative	
11h00 - 11h20	African Medicines Agency (AMA) AMA Representative	
11h20 - 11h40	The National Health Laboratory Service (NHLS) and rare diseases Associate Prof George van der Watt NHLS	
11h40 - 12h00	Developing innovative and sustainable models for access to rare disease medicines in Low and middle-income countries (LMIC) - The example of South Africa Prof Fatima Suleman WHO/University of Kwazulu-Natal	
12h00 - 12h15	Q&A	
12h15 - 13h30	Lunch	
13h30 - 15h00	SESSION 3a - Venue: Kgotla 2 Data & Research	SESSION 3b - Venue: Lecture Room 10 Community Engagement
13h30 - 13h42	An overview of data and research issues in rare diseases Prof Michele Ramsay Sydney Brenner Institute for Molecular Bioscience	A family centric approach Mr Hedley Lewis CHOC
13h42 - 13h54	Overview of the International Rare Diseases Research Consortium (IRDiRC) Dr David Pearce IRDiRC	Community advisory boards for rare diseases Mr François Houÿez EURORDIS Rare Diseases Europe
13h54 - 14h06	Recruitment strategies for a non-hospital based academic biobank in South Africa Dr Elne Conradie North-West University	Addressing challenges of inclusion, equity, and diversity for rare disease patients Ms Malebo Malope Stellenbosch University
14h06 - 14h18	Patient-Initiated Rare Disease Registry for South Africa Ms Kelly du Plessis Rare Diseases South Africa	Patient perspective (interview format) Ms Miracle May & Ms Nomsa Mtshali Moebius Support South Africa
14h18 - 14h30	Patient perspectives of Sickle Cell Disease care in emergency centre's of South African hospitals Ms Nabeelah Peerbhai University of Cape Town	The negative impact of medical pricing and policy issues on patients living with rare diseases in South Africa Ms Belinda Nell Caregiver
14h30 - 14h42	The burden of hereditary angioedema in South Africa in 2023: A patient survey Ms Janice Strydom Hereditary Angioedema Foundation of South Africa	Transplant Conversations: Transplant education for medical staff, patients and the public Mrs Stella de Kock Transplant Education for Living Legacies (TELL)
14h42 - 15h00	Q&A	Q&A

15h00 - 15h15	Tea/Coffee Break
15h15 - 17h00	SESSION 4 - Venue: Kgotla 2 Clinical Trials (Panel Discussion)
15h15 - 17h00	Clinical trials: what, where, why, how & who - Mr Peter Bach, BioPharmaLogic Understanding clinical trial possibilities and limitations - South African Health Products Regulatory Authority (SAHPRA) Representative Practicality and efficiency in clinical trials for rare diseases - Ms Catherine Lund, OnQ South Africa Patient engagement in clinical trials - Mrs Tasnim & Iman Casoojee, Cure SMA When your clinical trial becomes permanent - Mr Luc McIntosh, Patient Real World Evidence (RWE) and rare diseases - Mr Arshad Hassim, Aurum Institute
17h00	Cocktail Reception

DAY 2 Thursday, 15th February 2024

08h30 - 10h00	SESSION 5 - Venue: Kgotla 2 Rare Diseases in Africa - The Continental Impact
08h30 - 08h50	The rare disease landscape in Sub-Saharan Africa Associate Prof Helen Malherbe Rare Diseases South Africa
08h50 - 09h10	Universal Health Coverage (UHC) for rare diseases Ms Alexandra Heumber Perry Rare Diseases International
09h10 - 09h30	Establishment of Global Centre of Excellence for Rare Diseases Ms Nadia Bodkins & Prof Mapeseka Seheri Sefako Makgatho Health Sciences University
09h30 - 09h50	Africa Rare Disease Alliance (ARDA): Continental Overview ARDA Members
09h50 - 10h00	Q&A
10h00 - 10h20	Tea/Coffee Break
10h20 - 12h15	SESSION 6 - Venue: Kgotla 2 Health Technology Assessment (HTA)
10h20 - 10h40	What is health economic assessment and why do we need it for rare diseases? Prof Alex van den Heever University of the Witwatersrand
10h40 - 11h00	Public sector view Mr Andy Gray University of KwaZulu-Natal
11h00 - 11h20	Private sector view Ms Niri Bhimsan Discovery Health
11h20 - 11h40	The Professional Society for Health Economics and Outcomes Research (ISPOR) & improving HTA in South Africa – the role of patients Ms Laurenne James ISPOR South Africa
11h40 - 12h00	Patient perspective: Patient involvement in HTA Ms Lauren Pretorius Campaigning for Cancer
12h00 - 12h15	Q&A
12h15 - 13h30	Lunch

13h30 - 15h00	SESSION 7a - Venue: Kgotla 2 Data & Research	SESSION 7b - Venue: Lecture Room 10 Coordinated & Multidisciplinary Care
13h30 - 13h42	The sub-Saharan African Congenital Anomalies Network (sSCAN): a regional network to optimize care and research in congenital anomalies and rare diseases Associate Prof Helen Malherbe Rare Diseases South Africa	Multi-disciplinary care – what is it, who does it and how does it work? Prof Christian Hendriksz A Rare Cause
13h42 - 13h54	The relevance & impact of H3Africa for rare disease patients in Africa Associate Prof Zané Lombard University of Witwatersrand/NHLS	Role of genetic counselling in the rare disease community Ms Monica Araujo University of Witwatersrand/NHLS

13h54 - 14h06	Transitioning to the new REDCap SA Haemophilia Registry – first steps at Red Cross War Memorial Children's Hospital Dr Yasmin Goga Red Cross War Memorial Children's Hospital	Traditional medicine and rare diseases in Africa: culture, religion, stigma and community Mr Elliot Mqansa Makhathini University of KwaZulu-Natal
14h06 - 14h18	The genetics of Neuromuscular Diseases in South Africa: current outcomes of the ICGNMD study Prof Francois Van Der Westhuizen North-West University	Developing consensus statements for the care of patients with Epidermolysis Bullosa in South Africa: A Multidisciplinary Approach Dr Antoinette Chateau University of KwaZulu-Natal
14h18 - 14h30	Utilizing Multi-OMICS Technologies to unravel molecular mechanisms of orphan diseases in Low-and-Middle-Income -Countries: A focus on Konzo Dr Matthew Bramble Children's National, Washington DC, USA	The utilisation of genetic counselling services amongst prenatal healthcare providers in Gauteng, South Africa Ms Megan Duvenhage University of Witwatersrand/National Health Laboratory Services
14h30 - 14h42	A Southern African multiple acyl-CoA dehydrogenase deficiency cohort: clinical, biochemical, and genetic spectrum Ms Michelle Bisschoff North-West University	RDSA Project diagnosis and Lab Hub Ms Ankia Greyling Rare Diseases South Africa
14h42 - 15h00	Q&A	Q&A
15h00 - 15h15	Tea/Coffee Break	
15h15 - 17h00	SESSION 8a - Venue: Kgotla 2 Omics in Africa	SESSION 8b - Venue: Lecture Room 10 The Diagnostic Odyssey
15h15 - 15h30	The Nngwe initiative – Ending the diagnostic odyssey using Omics Prof Chris Vorster North-West University/Nngwe Initiative	Navigating the diagnosis journey: A patient perspective Mrs Stacey Leslie Andrea's Gift Foundation
15h30 - 15h45	Sharing is caring: Towards democratizing genomic data in South Africa Mr Jaco Oosthuizen University of the Free State	Ending the diagnostic odyssey for South African patients with rare developmental disorders – the DDD-Africa study Ms Zandisiwe Goliath University of Witwatersrand
15h45 - 16h00	Study design and getting the right story out of your data: making proteomics data for profiling and diagnosis possible at scale Dr Liam Bell Centre for Proteomic & Genomic Research	Untold Stories: South African Dysautonomia patients share their experiences of diagnostic delay Ms Jana Fokkens Dysautonomia Support South Africa
16h00 - 16h15	Sharing small molecules on a big scale: A proposal for collaborative metabolomics for rare disease characterisation and diagnosis Dr Ilse du Preez North-West University	Navigating Whole Genome Sequencing (WGS) frontiers: A parent's journey with online consumer DNA kits in rare disease diagnosis Ms Beth O'Conner-Cilliers Parent
16h15 - 16h30	Functional studies: a key outcome tool for a multi-omics approach to rare disease diagnostics Prof Francois van der Westhuizen North-West University	Bulletproof: A mother's uphill battle for her children's survival Mrs Chantal Mackenzie Caregiver
16h30 - 17h00	Q&A	Q&A
19h00	Conference dinner	

DAY 3 Friday, 16th February 2024

08h30 - 10h00	SESSION 9 - Venue: Kgotla 2 Access to Innovation
08h30 - 08h50	Medical education programmes in Low- and Middle-Income Countries are not a lost cause! Prof Christian Hendriksz A Rare Cause
08h50 - 09h10	Risk equalization models Mr Charlton Murove Board of Healthcare Funders
09h10 - 09h30	National Department of Health's (NDOH) view on access to innovation limitations Ms Khadija Jamaloodien NDOH
09h30 - 10h00	Challenges of treating a rare disease in Rwanda Prof Christian Hendriksz A Rare Cause
10h00 - 10h20	Tea/Coffee Break
10h20 - 12h15	SESSION 10 - Venue: Kgotla 2 The local rare disease landscape and its challenges
10h20 - 10h40	Update on implementation of South Africa's 2021 Clinical Guidelines for Genetic Services Dr Manala Makua National Department of Health
10h40 - 11h00	Council for Medical Schemes (CMS): PMB legislation CMS Representative
11h00 - 11h20	Legal overview on where rare diseases fit into South African law and legislation Ms Elsabé Klink Elsabé Klink & Associates

11h20 - 11h40	Rare Disease Access Initiative (RDAI) Ms Meliska Volschenk Sanofi	
11h40 - 12h15	Q&A	
12h15 - 13h30	Lunch	
13h30-15h00	SESSION 11a - Venue: Kgotla 2 Policy	SESSION 11b - Venue: Lecture Room 10 Supportive Care
13h30 - 13h45	Influencing health and education policy in Tanzania: The role of Ali Kimara Rare Disease Foundation (AKRDF) Dr Mohamedraza Ebrahim AKRDF/Aga Khan Hospital Tanzania	SA Palliative care guidelines and implications for rare disease patients Dr Michelle Meiring Paedspal
13h45 -14h00	Newborn screening policy: from global to local Prof Chris Vorster North-West University/Nngwe Initiative	Respite care for rare diseases in South Africa – is there any? Ms Tersia Burger Association of Palliative Care Centres/Stepping Stones Hospice
14h00 - 14h15	Transitional care in Africa: living beyond expectancy Prof Mignon McCulloch Red Cross War Memorial Children's Hospital/University of Cape Town	What is psychosocial support and who provides it? Ms Nomsa Mtshali Rare Diseases South Africa
14h15 - 14h30	The impact of Covid-19 on rare disease patients in South Africa Ms Marianne Gomes PathCare	Healthcare professional compassion fatigue and burnout Ms Kim Ballantine, Camber Coaching
14h30 - 14h45	Africa roadmap project Ms Roselyn Odero International Gaucher Alliance	
14h45 - 15h00	Q&A	Q&A
15h00 - 15h15	Tea/Coffee Break	
15h15 - 17h00	SESSION 12a - Venue: Kgotla 2 Diagnostics	SESSION 12b - Venue: Lecture Room 10 Innovative Patient Care
15h15 - 15h30	Expect the unexpected: Inborn errors of metabolism diagnosed in early/late adulthood Dr Marli Dercksen North-West University	Challenges and health system strengthening opportunities identified to support the detection and assessment of congenital disorders in the UBOMI BUHLE Pregnancy Exposure Registry Cohort Ms Halalisani (Lisa) Mahlaba Ubomi Buhle Pregnancy Exposure Registry
15h30 - 15h45	An update on advocacy efforts for rare diseases in South Africa Associate Prof Helen Malherbe Rare Diseases South Africa	Mucopolysaccharidosis VI (Maroteaux-Lamy Syndrome): Rarity of diagnosis and difficulty accessing treatment: Dr Anele Dube Pule Kwazulu-Natal Provincial Department of Health
15h45 - 16h00	Exploring gene mutations and secondary mitochondrial disease in a patient with neuromuscular diseases. A diagnostic odyssey. Dr Maryke Schoonen North-West University	Nova, Teaching with Alternative Communication Mrs Dionne Snyders Parent
16h00 - 16h15	STAC3 variants cause a congenital myopathy with distinctive dysmorphic features in African paediatric patients Dr Tendai Makwikwi North-West University	Moving beyond verbiage Mr Andrew Miller Freelance Journalist & Patient
16h15 - 16h30	Proposal for selection criteria for newborn screening disorders Ms Brenda Klopper North-West University	A brighter and more inclusive future for those affected by achondroplasia Mrs Chantelle Hall Parent
16h30 - 16h45	Q&A	Q&A
16h45 - 17h00	Plenary: Call to action & conference closure: Ms Kelly du Plessis Conference Chair	
19h00	Speaker dinner (by invitation only)	

DAY 4 Saturday, 17 February 2024 (Open to support groups/patients and caregivers/patient advocates)

08h30 - 17h00	Venue: Kgotla 2 Advocacy in Action - Capacity Building (Sponsored by Rare Diseases International, Novartis & Boehringer Ingelheim)
08h30 - 12h30	Fundraising Workshop Facilitated by Ms Michelle Stein - Handmade Fundraising Capacity Building - Strategic development of Vision, Mission and Strategic overview Faciliated by Ms Kelly du Plessis - Rare Diseases South Africa
12h30 - 13h30	Lunch & talk - Compassion fatigue and burnout for caregivers: Dr Rob & Ms Kim Ballantine, Camber Coaching
13h30 - 17h00	Closed Meeting for National Alliances Understanding policy and engaging policy makers Facilitated by Veronica Lopes Gousset – VLG Consulting
19h00	Dinner for Patient Organisations/Patient Advocates and Support Group Representatives

RARE

POSTER PROGRAMME & board allocation

Presenting Author Names	Paper Title	Theme	Poster Board No.
Mrs Tania Grobler	Enhancing diagnostic efficiency of Amino Acid-related disorders through automated sample preparation and liquid chromatography Tandem Mass Spectrometry Analysis	Access to innovation	P1
Dr Engela Conradie	Rare Disease Capacity building in Africa	Community engagement	P2
Ms Tina Angelos	Making Multiple Sclerosis real and relevant - The reality of living a full and inspired life through the eyes of an MS warrior	Co-ordinated care	P3
Ms Thembelihle Ngcai	Highlighting the gap in co-ordinated care in mitigating risks of convulsive syncope in patients with spinal muscular atrophy (SMA) as a result of epinephrine-combined anaesthesia during dental extractions	Co-ordinated care	P4
Ms Derylize Beukes	Feasibility study: Effect of various sample pre-treatment procedures on stability of creatinine results and overall implication on downstream interpretation and diagnosis of inherited metabolic disorders	Diagnosis	P5
Miss Elmarie Davoren	Proton nuclear magnetic resonance (1H-NMR) spectroscopy as a diagnostic tool for purine/pyrimidine disorders in South Africa	Diagnosis	P6
Mr Zachary Cline Mr Sidney Muchemwa	Redefining the rare disease diagnostic journey: Leveraging ICD-10 Code R69 for improved patient outcomes	Diagnosis	P7
Dr Chinmaya Singh	Neuronal ceroid lipofuscinosis type-11 in early childhood	Diagnosis	P8
Prof Marco Zampoli	Cystic Fibrosis in South Africa: who are we missing?	Diagnosis	P9
Ms Wanyue Wang	Use of NGS-based carrier screening in prevention of severe recessive monogenic disease	Diagnosis	P10
Dr Monique Opperman	Non-Enzymatic Formation of N-acetylated Amino Acid Conjugates in Urine	Research	P11
Mr D'andre Spencer	AI-guided newborn screening and rapid genetic testing in low-resource settings	Research	P12
Mr Armand Vorster	Functional comparison of in vitro models for RYR1 and STAC3 gene knockdown	Research	P13
Mr Liknaw Bewket Zeleke	Successful surgical repair outcomes and determinants of obstetric fistula in Low- and Middle-Income Countries: A systematic review and meta-analysis	Research	P14
Dr Evashin Pillay	Gaucher disease in the adult patient: a diagnostic approach from the perspective of a haematopathologist	Diagnosis	P15