RARE Rare Diseases Conference 2018

Programme

as at 11 September 2018

THURSDAY, 13 SEPTEMBER

	07:30 - 17:00	Registration
	08:30 - 17:00	Exhibition
Opening Session	08:30 - 08:40	Welcome | ***Kelly du Plessis, Rare Diseases South Africa***
	08:40 - 09:10	Opening | ***Nonhlanhla Dlamini, Chief Director: Child and Youth Health, National Department of Health***
	09:10 - 09:30	***Session 1: Keynote | Kelly du Plessis, Chairperson***
Keynote	09:10 - 09:30	Chasing the rainbows through the storms: A courageous journey with Moyamoya disease | ***Timothy Stones***
	09:30 - 10:30	***Session 1: Genetic Landscape in Africa | Thirona Naicker, Chairperson***
Session 1	09:30 - 10:00	Role and structure of human genetics policy | ***Manala Makua, National Department of Health***
	10:00 - 10:30	Overview of congenital disorders and rare diseases in South Africa | ***Helen Malherbe, University of KwaZulu-Natal***
	10:30 - 11:00	*Refreshment break*
	11:00 - 12:30	***Session 2: Diagnostics and Research | Marli Dercksen, Chairperson***
Session 2	11:00 - 11:30	Metabolic diseases and the continued importance of biochemical studies in the new -omics era | ***Ronald JA Wanders, University of Amsterdam***
	11:30 - 11:45	Genetic diagnosis of disorders/differences of sex development (DSD): The DSD-translational research network experience | ***Eric Vilain, Children's National Medical Center, Washington, DC***
	11:45 - 12:00	Impact and cost effectiveness of newborn screening | ***Chris Vorster, North-West University***
	12:00 - 12:15	Utilising facial recognition software for the detection of genetic syndromes: A focus on the Democratic Republic of the Congo | ***Matthew Bramble, Children's National Medical Center, Washington, DC***
	12:15 - 12:30	An evaluation of preimplantation genetic diagnosis outcomes in Johannesburg, South Africa | ***Bianca Carzis, National Health Laboratory Service, University of the Witwatersrand***
	12:30 - 13:30	*Lunch*
	13:30 - 15:00	***Session 3: Current and Emerging Treatments | Chris Hendriksz, Chairperson***
Session 3	13:30 - 14:00	Limb Girdle Muscular Dystrophy (LGMD): An expanding family of rare genetic myopathies | ***Mark Roberts, Greater Manchester Neurosciences Centre***
	14:00 - 14:15	Exome sequencing of the first reported South African x-linked primary immunodeficiency case identifies a hemizygous mutation in MSN | ***Brigitte Glanzmann, Stellenbosch University***
	14:15 - 14:30	The development of a therapeutic approach for the treatment of individuals with Prader-Willi Syndrome and their primary caregivers | ***Molelekeng Sethuntsa , Dr George Mukhari Academic Hospital***
	14:30 - 14:45	Mitochondrial disease in South Africa: Clinical, biochemical and genetic outcomes in paediatric patients | ***Maryke Schoonen, North-West University***
	14:45 - 15:00	Neuroendocrine tumours (NETs0 | ***John Devar, Wits Donald Gordon Medical Centre supported by Novartis***
	15:00 - 15:30	*Refreshment break*
	15:30 - 17:15	***Session 4: Rare Disease Access Frameworks: Challenges and Solutions | Kelly du Plessis, Chairperson***
Session 4	15:30 - 16:15	Prescribed Minimum Benefits and National Health Insurance updates | ***Peta-Anne Durrant, Elsab é Klinck & Associates***
	16:15 - 17:00	Diagnosis, treatment, and management of rare diseases in Africa |***Chris Hendriksz, FYMCA Medical Ltd***
	17:00 - 17:15	Poster presentations | ***Flavia Zita Francies and Elzette Nienaber***
	17:15	**Welcome Networking Reception (Riversands Cafe)**

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FRIDAY, 14 SEPTEMBER

	07:30 - 17:00	Registration
	08:30 - 17:00	Exhibition
Keynote	08:30 - 09:15	The Fabry masquerade | ***Bertram Henderson, University of the Free State, supported by Sanofi Genzyme***
	09:15 - 10:45	***Session 5: Rare Diseases in Public Health | Chris Hendriksz, Chairperson***
Session 5	09:15 - 09:45	From common to rare long-term conditions | ***Neil McKerrow, KwaZulu-Natal Department of Health***
	09:45 - 10:00	Neonatal mortality remains unacceptably high: No stone should be left unturned | ***Manala Makua, National Department of Health***
	10:00 - 10:15	Barriers to diagnosis of rare genetic diseases in the South African state healthcare system | ***Michael Urban, Tygerberg Hospital, Stellenbosch University***
	10:15 - 10:30	Public health approach to birth defects: The Argentine experience | ***Boris Groisman, National Network of Congenital Anomalies of Argentina (RENAC)***
	10:30 - 11:15	*Refreshment break*
	11:15 - 12:45	***Session 6: The Multidisciplinary Approach | Helen Malherbe, Chairperson***
Session 6	11:15 - 11:30	Introduction to the multidisciplinary approach | ***Kelly du Plessis, Rare Diseases South Africa***
	11:30 - 11:45	Genetics services and role in diseases affecting multiple systems| ***Bertram Henderson, University of the Free State***
	11:45 - 12:00	A pilot study evaluating depression in mothers with children diagnosed with Down Syndrome in state healthcare | ***Melissa Swanepoel, University of the Witwatersrand, National Health Laboratory Service Braamfontein***
	12:00 - 12:15	Palliative care for children with rare diseases | ***Busi Nkosi, International Children's Palliative Care Network***
	12:15 - 12:30	Pulling it all together: The role of genetic counsellors | ***Sarah Walters, AMPATH Trust***
	12:30 - 12:45	From the head to the heart | ***Julia Ambler, Umduduzi - Hospice Care for Children***
	12:45 - 13:45	*Lunch*
	13:45 - 15:30	***Session 7: Adaptive Approaches to Care | Chris Hendriksz, Chairperson***
Session 7	13:45 - 14:15	Guillain-Barre syndrome (GBS): A rare potentially treatable disorder | ***Mark Roberts, Greater Manchester Neurosciences Centre***
	14:15 - 14:30	The Kidney Beanz Trust | ***Stacey Hanekom, The Kidney Beanz Trust***
	14:30 - 14:45	Olive Children's Foundation| ***Colin Noel, Olive Children's Foundation***
	14:45 - 15:00	Differences/disorders of sex development: A world of uncertainty | ***Eric Vilain, Children's National Medical Center, Washington DC***
	15:00 - 15:15	Breakthrough for Huntington's Disease | ***Michael Howard, Huntington's Association of South Africa***
	15:15 - 15:30	Huntington's Disease Youth Organisation | ***Catherine Martin, Huntington's Disease Youth Organisation***
	15:30 - 16:00	*Refreshment break*
Session	16:00 - 17:15	***Session 8: Panel discussion: "Patient-centricity" and what it actually means (Kelly du Plessis, Bertram Henderson, Julia Ambler, Lauren Pretorius, Michael Urban)***
	17:15	Genetic Alliance South Africa General Meeting

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SATURDAY, 15 SEPTEMBER

	07:30 - 17:00	Registration
	08:30 - 18:00	Exhibition
Keynote	08:30 - 09:00	The Jenna Lowe Trust | ***Gabi Lowe, The Jenna Lowe Trust***
	09:00 - 10:45	***Session 9: Patient Registries / Surveillance | Helen Malherbe, Chairperson***
Session 9	09:00 - 09:15	Surveillance methods and the proposed way forward | ***Manala Makua, National Department of Health***
	09:15 - 09:30	Harnessing technology for innovative and efficient integrated disease surveillance and control | ***Portia Mutevedzi, National Institute for Communicable Diseases***
	09:30 - 09:45	Public health surveillance of birth defects: The experience of the National Network of Congenital Anomalies of Argentina | ***Boris Groisman, National Network of Congenital Anomalies of Argentina (RENAC)***
	09:45 - 10:00	National cancer surveillance registry: Lessons learnt from mandatory reporting for cancer | ***Elvira Singh, National Health Laboratory Service***
	10:00 - 10:15	Development of a register to optimise care of children in South Africa with Duchenne Muscular Dystrophy | ***Alhaji Alusine Jalloh, Paediatric Neurology, University of Cape Town***
	10:15 - 10:30	Diamond-Blackfan Anaemia: An African registry makes strides for research and diagnosis | ***Colin Noel, Olive Children's Foundation***
	10:30 - 10:45	Undiagnosed? Have you considered testing for 22q11.2 Deletion Syndrome? | ***Benita Penfold, The 22q11.2 Deletion Syndrome Foundation South Africa***
	10:45 - 11:15	*Refreshment break*
	11:15 - 12:45	***Session 10: Support Groups | Lauren Pretorius, Chairperson***
Session 10	11:15 - 11:30	Global rare disease advocacy: Where are we, future plans, where does Africa fit in? | ***Yann Le Cam, Rare Diseases International***
	11:30 - 11:45	Driving policy change through advocacy: How to, what is needed, what opportunities are available ? | ***Kelly du Plessis, Rare Diseases South Africa***
	11:45 - 12:00	Advocacy 101 | ***Lauren Pretorius, Prime Consulting***
	12:00 - 12:45	STEP 1 - Defining the current state of the problem: Developing a list of issues | ***Group exercise***
	12:45 - 13:45	*Lunch*
	13:45 - 15:15	***Session 1: Patient Huddle | Helen Malherbe, Chairperson***
Session 1: Patient Huddle	13:45 - 14:15	STEP 2 - Finding solutions: Charitable access programmes | ***Tanya Collin-Histed, European Gaucher Alliance***
	14:15 - 14:35	Problem + Solution = Advocacy Issues | ***Lauren Pretorius, Prime Consulting***
	14:35 - 14:55	Developing a tailored issue tree | ***Group exercise***
	14:55 - 15:15	STEP 3 - The role of data and research in advocacy | ***Helen Malherbe, Genetic Alliance South Africa***
	15:15 - 15:45	*Refreshment break*
	15:45 - 17:00	***Session 2: Patient Huddle | Kelly du Plessis, Chairperson***
Session 2: Patient Huddle	15:45 - 16:00	Resourcing and sustaining an advocacy: What is needed, how to secure resources? | ***Weronika Wojtowicz, Care Beyond Diagnosis***
	16:00 - 17:00	Narrating problem statements | ***Group exercise***
	18:30	**Rare Diamond Awards (Indaba Hotel)**

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SUNDAY, 16 SEPTEMBER

	08:30 - 12:00	Registration
	08:30 - 17:00	Exhibition
	09:00 - 10:30	***Session 3: Patient Huddle | Helen Malherbe, Chairperson***
Session 3: Patient Huddle	09:00 - 10:00	STEP 4 - Who do we speak to? What do we say? | ***Lauren Pretorius, Prime Consulting***
	10:00 - 10:30	STEP 5 - Building support for your plan | ***Lauren Pretorius, Prime Consulting***
	10:30 - 11:00	*Refreshment break*
	11:00 - 11:30	Solutions: Plans for the way forward | ***Collective report***
	11:30 - 12:00	Closing session | ***Kelly du Plessis, Rare Diseases South Africa***