RARE Rare Diseases Conference 2018

13 - 16 September 2018 | Johannesburg

Provisional Programme as at 1 July 2018

Date	Venue	Time	
THURSDAY, 13 SEPTEMBER	Foyer	07:30 - 17:00	Registration
	Foyer	08:30 - 17:00	Exhibition and Posters
	Venue A	08:30 - 09:30	Opening Session - *Dr Yogan Pillay, Deputy Director-General| Department of Health* Plenary Session - Chasing the rainbows through the storms: a courageous journey with MoyaMoya disease | *Timothy Stones*
		Time	Venue A
		09:30 - 10:30	***Session 1: Genetic Landscape in Africa - Thirona Naicker, Chairperson***
	Session 1		Overview of CDs in South Africa - Epidimiology, legal framework, services and the way forward / *Dr Helen Malherbe, Helen Malherbe Consult* From bright idea to health gain. The contribution of the patients and families to innovation and healthcare in research and service delivery in rare diseases | *Dr Alastair Kent OBE* Genomic analyses in African populations identify novel risk loci near ctnna2 and in sult2a1 for cleft palate | *Professor Azeez Butali , University of Iowa*
	Foyer	10:30 - 11:00	*Refreshment break*
		11:00 - 12:30	***Session 2: Diagnostics abd Research - Dr Marli Dercksen, Chairperson***
	Session 2		Genetic diagnosis of disorders/differences of sex development (dsd): the dsd-translational research network experience | *Dr Eric Vilain, Children's National Health System* The development of a therapeutic approach for the treatment of individuals with Prader-Willi syndrome and their primary caregivers | *Dr. Molelekeng Sethuntsa , Dr George Mukhari Academic Hospital* An evaluation of preimplantation genetic diagnosis outcomes in Johannesburg, South Africa | *Ms Bianca Carzis, National Health Laboratory Service/ University of Witwatersrand* Exome sequencing of the first reported south african x-linked primary immunodeficiency case identifies a hemizygous mutation in msn | *Dr Brigitte Glanzmann, Stellenbosch University* Utilizing facial recognition software for the detection of genetic syndromes: a focus on the Democratic Republic of the Congo | *Dr Matthew Bramble, Children's National Medical Center* Mitochondrial disease in South Africa: clinical, biochemical and genetic outcomes in paediatric patients | *Ms Maryke Schoonen, North West University*
	Foyer	12:30 - 13:30	*Lunch*
		13:30 - 15:00	***Session 3: Current and Emerging Treatments - Professor Chris Hendrickz, Chairperson***
	Session 3		Breakthrough for Huntingtons Disease |*Mr Michael Howard, Huntington's Association of South Africa* The impossible is now possible - treatments for rare retinal conditions have begun | *Ms Claudette Medefindt, Retina South Africa*
		15:00 - 15:30	*Refreshment break*
		15:30 - 17:00	***Session 4: Rare Disease Policy - Neil McKerrow, Chairperson***
	Session 4		
	Foyer	17:00	*Welcome Networking Reception*

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FRIDAY, 14 SEPTEMBER	Foyer	07:30 - 17:00	Registration
	Foyer	08:30 - 17:00	Exhibition and Posters
		08:30 - 09:15	*Keynote -Zanofi Genzyme sponsored session*
		09:15 - 10:45	***Session 5: Rare Diseases in Public Health - Neil McKerrow, Chairperson***
	Session 5		Topic TBA | *Dr Neil McKerrow, Department of Health KZN* Patient-centered care model for orphan drugs | *Rizwan Arshad Khan, Health Services* Barriers to diagnosis of rare genetic diseases in the South African state healthcare system |*Dr Michael Urban, Tygerberg Hospital, Stellenbosch University* Effective utilisation of available laboratory investigations for rare inherited disorders in South Africa| *Ms Surita Meldau, University of Cape Town*
	Foyer	10:45 - 11:15	*Refreshment break*
		11:15 - 12:45	***Session 6: The Multidisciplinary Approach - Professor Bertram Henderson, Chairperson***
	Session 6		Topic TBA | *Dr Karen Fieggen, University of Cape Town* A pilot study evaluating depression in mothers with children diagnosed with Down Syndrome in state healthcare | *Ms Melissa Swanepoel, University of Witwatersrand, Hleath Laboratory Service Braamfontein* Palliative care for children with rare diseases | *Mrs Busi Nkosi, International Children's Palliative Care Network* Topic TBA | *Ms Sarah Walters, AMPATH*
	Foyer	12:45- 13:45	*Lunch*
		13:45 - 15:15	***Session 7: Adaptive approaches to Care - Professor Chris Hendrickz, Chairperson***
	Session 7		
	Foyer	15:15 - 15:45	*Refreshment break*
	Session 8	15:45 - 17:15	***Session 8: Panel discussion: Patient Centricity and what It actually means***

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SATURDAY, 15 SEPTEMBER	Foyer	07:30 - 17:00	Registration
	Foyer	08:30 - 18:00	Exhibition and Posters
		08:30 - 09:15	*Plenary Session*
		09:15 - 10:45	***Session 9: Patient Registries / Surveillance - Dr Helen Malherbe, Chairperson***
	Session 9		Public health surveillance of birth defects. The experience of the National Network of Congenital Anomalies of Argentina | *Dr Boris Groisman, National Network Of Congenital Anomalies Of Argentina* Development of a register to optimize care of children in South Africa with Duchenne Muscular Dystrophy | *Dr Alhaji Alusine Jalloh, Paediatric Neurology, University Of Cape Town* Diamond-blackfan anaemia: an African registry makes strides for research & diagnosis | *Dr Colin Noel, Olive Children's Foundation* Topic TBA | *Dr Manala Makua, National Department of Health* Topic TBA | *Dr Portia Mutavedzi, National Institute for Communicable Diseases*
	Foyer	10:45 - 11:15	*Refreshment break*
		11:15 - 12:45	***Session 10: Support Groups - Kelly du Plessis***
	Session 10		Undiagnosed? Have you considered testing for 22q11.2 Deletion Syndrome? | *Mrs Benita Penfold, 22q11.2 Deletion Syndrome Foundation South Africa* Olive Children's Foundation| *Dr Colin Noel, Olive Children's Foundation*
	Foyer	12:45 - 13:45	*Lunch*
		13:45 - 15:15	***Session 11: Patient Huddle Session 1***
	Session 11		
	Foyer	15:15 - 15:45	*Refreshment break*
		15:45 - 17:15	***Session 12: Patient Huddle Session 2***
	Session 12		Driving policy change through advocacy - how to, what is needed, what opportunities are available | *Kelly Du Plessis, Rare Diseases South Africa*
	Indaba Hotel	19:00	*Rare Diamond Awards Dinner*

Date	Venue	Time	
SUNDAY, 16 SEPTEMBER	Foyer	07:30 - 13:00	Registration
	Foyer	08:30 - 17:00	Exhibition and Posters
		09:00 - 10:30	***Session 13: Patient Huddle***
	Session 13		Driving policy change through advocacy - how to, what is needed, what opportunities are available |*Kelly Du Plessis, Rare Disease South Africa*
	Foyer	11:00 - 11:30	*Refreshment break*
		11:30 - 12:00	Resourcing and sustaining an advocacy - what is needed, how to secure resources | *Care Beyond Diagnosis*
		12:00 - 12:30	*Closing Session*