Newsflash!

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SMU Commemorates Rare Disease Day

Sefako Makgatho Health Sciences University (SMU) and the Rare Advocacy Movement (RAM) in partnership with Rare360 observed Rare Disease Day (RDD) on the 29th of February 2024 under the theme 'developing a global rare disease centre of excellence'. The day was commemorated to raise awareness about rare diseases and improve access to treatment and medical representation for individuals living with unusual diseases.

A rare disease is a medical condition that affects a small number of people compared to the general population. Types of rare diseases include Gaucher disease caused by enzyme deficiency that leads to the storage of complex lipids in certain types of blood cells, Haemophilia a blood disorder that stops blood from clotting properly and Cystic fibrosis is a genetic (inherited) disease that causes sticky, thick mucus to build up in organs, including the lungs and the pancreas amongst others.

SMU's Vice-Chancellor Prof Peter Mbati welcomed all esteemed guests at the event and indicated that this day was held in honour and celebration of the incredible strength and resilience of those who are affected by rare diseases. He informed the audience that the Health Equity Diagnostic Infrastructure (HEDI) Initiative hosted by the RAM will partner with the university to establish a medical infrastructure as a global rare disease centre of excellence within SMU that will offer diagnostic and clinical services and support therapeutic research and development programmes. “My humble request to you is to help champion this course and for the Department of Health to work closely with us in the establishment of this global rare disease centre of excellence,” said Prof Mbati.

Gauteng MEC for Health and Wellness Nomantu Nkomo-Ralehoko said in her address that, it was an honour to stand before the audience on this occasion of Rare Disease Day. She explained that this day serves as an important reminder of the ongoing struggles faced in our country by individuals with these diseases and the importance of collective action taken in addressing these challenges. “In South Africa, many individuals with rare diseases face not only health challenges but also social and economic barriers. it is our duty as a community to ensure that no one is left behind in our pursuit of health equity. I would also like to extend my gratitude to the university and its leadership for hosting and organizing this event,” shares the MEC.

Founder and Chief Executive Office of Rare Disease South Africa Kelly du Plessis stressed issues around the Rare Disease Ecosystem. She showed that the ecosystem of rare diseases is overlooked within the borders of South Africa. She explained that numerous rare diseases silently affect lives by challenging individuals and families in ways that many of us cannot fully comprehend. “The ecosystem of rare diseases encompasses not only the patients, but also the healthcare providers, researchers, policymakers and carriers, and caregivers that navigate the intricate landscape of diagnosis, treatment, and support on a day-to-day basis. It is crucial that we recognize the importance of collaborative efforts by fostering a network of medical professionals, researchers, and advocates,” said Du Plessis.

Master of Science in Biochemistry student Siphesihle Mkhwanazi said his highlight of the event was meeting the exhibitors. He mentioned that he was able to exchange contacts and foster relationships for future collaborations, which could lead to publishing together with one of the exhibitors.

About 72% of rare diseases are estimated to have a genetic component. Starting in childhood: Around two-thirds of people living with a rare disease are children. Having a broad range of symptoms: Symptoms can vary greatly from disease to disease, and even from person to person with the same disease. Being difficult to diagnose.

In addressing the initiatives to prioritise the needs of rare disease patients and their families, RDD brought together a vibrant confluence of partners including advocacy groups, academia, industry professionals and allies, as well as regulatory authorities, public policy professionals, and political leaders. All of whom supported the rare disease community by advocating for improved access to treatment and allied services, as well as by engaging in activities that discuss novel solutions, the latest clinical cases and the future of rare disease therapeutics.