Sickle cell anemia patient 'cured' through gene therapy

Researchers have reported that the first sickle cell disease patient in the world has achieved complete remission after an experimental gene therapy. This took place at Necker Children's Hospital in Paris. The patient was treated with lentiviral vector-mediated addition of an antisickling β-globin gene into autologous hematopoietic stem cells. The patient started treatment from age 13, and after 15 months of treatment, the level of anti-sickling β-globin remained high and the patient has reported no symptoms and has not required any further treatment. Researchers believe that the patient is cured but long term follow-up is required. This is a proof of concept for human patients. Future plans involve proceeding though clinical trials and if results are promising, treatment will be made available to patients. The case report was published in The New England Journal of Medicine and can be found at http://www.nejm.org/doi/10.1056/NEJMoa1609677#t=articleTop

International rare diseases day

The 28th of February 2017 marked the 10th annual international Rare Diseases Day. The staff and students of Human Genetics at the University of Cape Town were once again involved in supporting and assisting Rare Diseases SA in their efforts to raise awareness about rare diseases and research. In order to assist their fundraising campaign, Rare Disease Day ribbons were sold and everyone was encouraged to wear jeans in support of 'Jeans for Genes'. On the day, a stand was set up at the Red Cross War Memorial Children's hospital. The team interacted with many curious visitors and engaged in the wide range of interests. Donations were made from the following organisations, namely the Heart and Stroke Foundation, the Groote Schuur Hospital Haematology Clinic, the Muscular Dystrophy Foundation and Genetic Alliance. Money was raised for this fantastic cause and the day proved to be a great success.

Discovery of new gene that causes sudden death in adolescents

Medical researchers, through a global collaboration, have identified a new gene that is a major cause of sudden death among young people and athletes. The gene, called CDH2 (Cadherin 2), causes Arrhythmogenic Right Ventricle Cardiomyopathy (ARVC), a genetic disorder that predisposes young people to cardiac arrest. In ARVC, the heart muscle tissue is replaced by fatty and fibrous tissue. This process encourages the development of an abnormal heart rhythm (cardiac arrhythmias) such as rapid heart rhythm or rapid and erratic heart rhythm (ventricular fibrillation). This causes loss of consciousness and cardiac arrest. In the case of ventricular fibrillation, without a ready device to shock the heart, it causes sudden death in a few minutes. The discovery has both scientific and clinical impact. It helps to clarify the genetic mechanisms and contributes toward a more complete identification of the disease genes involved in cardiomyopathy. It also makes possible the early detection of many unsuspecting people who are affected by ARVC. More information at: http://www.health.uct.ac.za/news/press-release-discovery-new-gene-causes-sudden-death-adolescents#sthash.JZSk8Fo2.dpuf; http://circgenetics.ahajournals.org/content/10/2/e00160

PROFILE

Tarryn Shaw

MSc (Med) Genetic Counselling

Division of Human Genetics, UCT

1. **What is your current area(s) of research?**

   I completed my master's research on Fetal Alcohol Spectrum Disorders (FASD) in the Western Cape. I wanted to understand the reasons behind why women drink during their pregnancy, to understand their education and awareness about the effects of alcohol consumption during pregnancy, and to determine what prevention and intervention work is required to attempt to reduce the alarming rates of FASD in South Africa.

2. **What is your background and how did you become interested in your field of research?**

   I am fascinated by both the medical and molecular (genetic) aspects of diseases. I have a passion for working with and helping people and therefore my career as a genetic counsellor is the perfect fit for combining these aspects and working with people. I obtained my BSc Honours degree at Stellenbosch University in 2012. I completed my MSc (Med) Genetic Counselling degree in 2015 at UCT and I recently completed my HPCSA internship in 2016. I am now a newly qualified genetic counsellor.

3. **What piece of advice would you give to somebody beginning their career in genetics?**

   "Choose a job you love and you will never have to work a day in your life." Following your dreams is challenging but your passion will drive you. Don't ever let anyone stand in the way of your dreams or make you doubt your own capability. Where there is a way, find it. Where there is no way, make it.

4. **What does your typical working day look like?**

   I work at different clinics at both Groote Schuur Hospital and Red Cross Children's Hospital. I am also involved in facilitating medical students with cases presented to them and participate in the clinical and academic activities at UCT.

5. **Do you know any science jokes you can share?**

PROFILE

Nicholas Ekow Thomford

Postdoctoral Research Fellow

University of Cape Town

What is your current area(s) of research?

I am currently looking at Pharmacogenomics of commonly used medicinal herbs in Africa and their drug interaction potential. I am also exploring their HIV-latency reactivation potential. I have sequenced RNA from HIV models and analyzing the data to look at how these herbs modulate latency activation genes. I am also looking at the genetics of congenital heart malformations.

What is your background and how did you become interested in your field of research?

I have Medical Biochemistry background (BSc and MPhil). My interest in herbal medicine research stem from observations whiles working at a Teaching Hospital where patients were coming in with conditions related to taking herbals. My interest in genetics of congenital heart malformations was influenced by my son being born with a congenital heart disease.

What piece of advice would you give to somebody beginning their career in genetics?

Hard work and determination pays.

What does your typical working day look like?

I normally get to the lab by 7:30am every day. Read my mails and start organizing for the day. I do my planned research and assignments and leave the lab earliest at 5pm. I come in on weekends if I am not able to complete my planned tasks for the week.

Has anyone in particular inspired you in your field or science in general?

I am inspired by my PI, Prof. Collet Dandara. He builds me up and encourages me to be the best I can be. He has achieved a lot in his field and I look forward to following in his footsteps.