

Awareness leads people out of the darkness of neurodegenerative disease

Patients diagnosed with Spinocerebellar ataxia type 7 (SCA7) suffer from degeneration of the cerebellum, the part of the brain that controls movement. They also suffer from degeneration of retinal photoreceptors, the cells at the back of the eye that are responsible for vision.



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Danielle Smith, a PhD graduate in Human Genetics at the University of Cape Town, believes it is vital for the medical fraternity and the South African public to know more about the disease, which often leads to people being wheelchair-bound and unable to see.

"The Spinocerebellar ataxias are not well known, even amongst the medical community, because they are relatively rare. However, we have shown that South Africa has a considerable number of patients and families with SCA and I believe they deserve to be helped."

The disease manifests mainly when patients are in their 40s and 50s.

"Younger patients usually develop eye problems at first. They often have problems with walking, swallowing and eating. The disease usually progresses to death within 10 years."

Smith says it is important to raise awareness of the condition so that people can be referred for molecular testing.

"Overall, we hope that this body of work will have a significant impact on the lives of many African patients with ataxias, since they will receive a molecular diagnosis for their condition, allowing for better healthcare management, planning and future treatment."

"It's a tragic disease and it's important for patients and their families to be counselled and warned of the risks for current and future offspring."

Her research also focused on the optimisation and implementation of an improved molecular diagnostic test for SCA7. Because of Smith's research, this method is now used routinely in the National Health Laboratory Services diagnostic laboratory, so that a greater number of patients can receive a confirmed molecular diagnosis through DNA testing. There is currently no cure for SCA7, although clinical trials are on the horizon.

Smith also researched using a new technology to turn patient skin cells into a type of stem cell, called induced pluripotent stem cells (iPSCs). These cells can be cultured in the laboratory, and turned into any cell type in the body.

"I used SCA7 patient iPSCs to grow photoreceptors (eye cells) which could be used in the laboratory to study the disease," said Smith.

Smith, who hopes her research will contribute to a greater understanding and study of the SCAs in Southern Africa, said :

was inspired to study genetics at high school.

"I find it fascinating that DNA, something which is invisible to the human eye, can control the development, functioning and health of the human body."

Smith graduated with a PhD in Human Genetics at UCT in December 2014.

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