New study sheds light on genetics of schizophrenia in South Africa

Schizophrenia affects approximately 1% of people in all parts of the world and is one of the leading causes of disability worldwide. A study revealed that amaXhosa individuals with schizophrenia are significantly more likely to carry rare damaging genetic mutations compared to amaXhosa individuals without severe mental illness.

Image source: Getty/Gallo

Co-author of the paper, Professor Dan Stein, head of the Department of Psychiatry at UCT, said: “The World Mental Health Survey consortium and the Global Burden of Disease studies have emphasised how prevalent and burdensome mental disorders are. Schizophrenia is an important component of that.”

Furthermore, the genes disrupted by the rare damaging mutations of these patients are likely to be involved in the organisation and function of brain synapses. Synapses coordinate the communication between neuronal cells, and the firing and organisation of neuronal synapses are ultimately responsible for learning, memory and brain function.

The study was undertaken not because the amaXhosa have an unusual prevalence of schizophrenia, but because there is need for genetics work in African populations, and because these populations have the greatest wealth of human genetic diversity. Importantly, the depth of genetic variation in Africa allowed findings on schizophrenia to emerge using a moderate sample size, and this helps inform our understanding of the genetics of schizophrenia worldwide.

Co-author Professor Raj Ramesar, head of the Department of Pathology and Division of Human Genetics at UCT, continues on this point: “We all share the vast majority of our genome. As a tool when looking at the relationship between populations we look at the tiny bits that we know we vary by, and these can provide insights. We’re keen on findings that will lead to better management of the illness locally. To some extent it’s still very early. Schizophrenia is a combination of mutations and various other factors.”

Human biology is universal. The genes and pathways identified by this research inform the understanding of schizophrenia for all human populations and provide potential mechanisms to design new more effective treatments.

Source: University of Cape Town