Division of Human Genetics
University of Cape Town

Schizophrenia

What is Schizophrenia?

Schizophrenia (SCZ) is a multifactorial, long-term, complex psychiatric disorder involving a breakdown in the relation between thought, emotion and behaviour, often characterised by inconsistent or contradictory elements. The disease affects approximately 24 million individuals worldwide, with an age at onset ranging from 15 to 45 years. The prevalence of SCZ is approximately 0.5-1%, with an incidence rate of 8-40 new cases per 100 000 individuals reported annually. Whilst the prevalence of SCZ is more or less equal between male and female individuals, males tend to have an earlier onset of the disorder as well as a more debilitating course of the illness. In an African context, it is estimated that between 4 and 5 million African individuals suffer from a psychiatric disorder. SCZ accounts for approximately 10% of the health burden in sub-Saharan Africa. In particular, neuropsychiatric disorders are ranked third in terms of South African burden of disease, with an estimated one third of South African individuals suffering from some form of psychiatric illness.

What causes Schizophrenia?

To date, there is no established cause of SCZ, although it is thought to be caused by a variety of factors. Environmental factors such as family history, obstetric and prenatal conditions, infection, and place/time of birth have been shown to increase risk to the disease. In particular, unique African cultural factors may also contribute to the onset of SCZ, an example of which may be the initiation rites of males in the African Xhosa culture.

Genetics plays a large role in disease susceptibility, and SCZ has been shown to have a high rate of heritability (60-80%), which suggests a major role for transmitted genetic variants in disease risk. Linkage analysis, genome-wide association studies (GWAS) and whole exome sequencing (WES) are current techniques used for the identification of genetic markers for SCZ, and to date have allowed for the discovery of unique disease characteristics.

SCZ is described as a polygenic disorder, which suggests that a number of genes with variants/mutations of small effect interact to produce the observed symptoms.

Symptoms

SCZ is characterized by many severe and varying symptoms which can be classified as either positive, negative, cognitive or mood symptoms. These symptoms are measured on various scales, including the Positive and Negative Syndrome Scale (PANSS), the Scales for the Assessment of Negative and Positive Symptoms (SANS, SAPS) and the Brief Psychiatric Rating Scale (BPRS). Positive symptoms are defined as being absent in the general population but present in the SCZ individual, and include impaired reality testing, delusions, hallucinations, and disorganized speech and behaviour. Negative symptoms (present in the general population but absent in the SCZ individual) include Lack of motivation and speech, reduced social drive and emotional blunting. Mood symptoms such as depression, increased emotional arousal and impairment of the affected experiences and expression often precede the onset of the disorder. Other symptoms can be cognitive, and include impairment in attention and working memory.

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Treatment

To date, there is no cure or efficient treatment options for individuals suffering from SCZ. The most successful treatment options are anti-psychotics, the most common of which are chlorpromazine, risperidone, fluphenazine and haloperidol. However, treatment can often be poorly effective and may have adverse drug events. It is therefore important to identify potential genetic contributors and other factors that predispose individuals to SCZ, with the purpose of improving clinical outcome.

Useful website and resources:

•UCT Human Genetic website:
www.humangenetics.uct.ac.za

•Royal College of Psychiatrists website:
www.rcpsych.ac.uk

•Patients like me website:
www.patientslikeme.com

•National Institute of Mental Illness (NIMH): Schizophrenia overview

Support groups

• www.sadag.org

•www.sabda.org.za