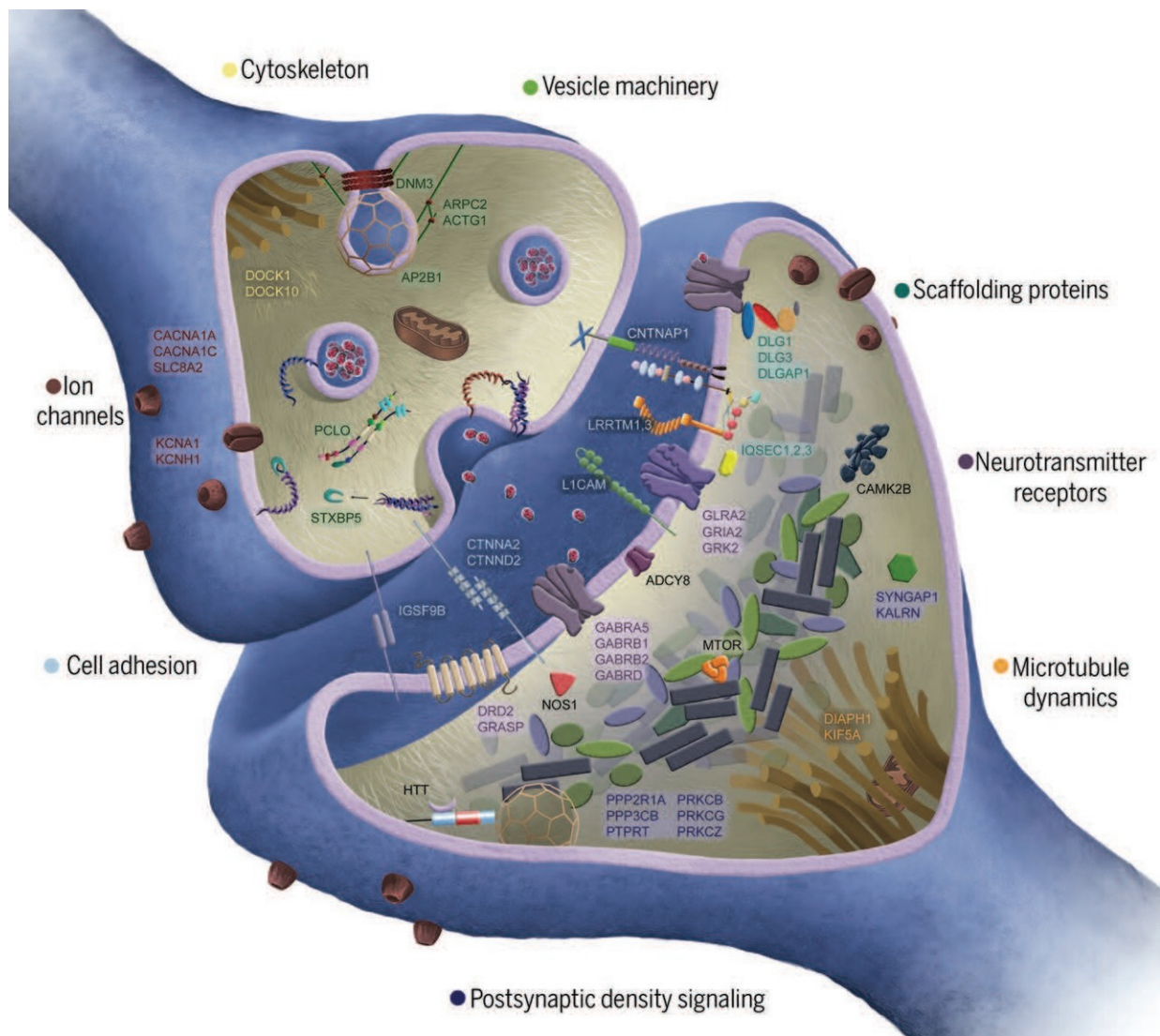


New clues into the genetic origins of schizophrenia

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Genes of the brain synapse with damaging mutations in schizophrenia. Credit: Gulsuner et al., *Science* (2019)

The first genetic analysis of schizophrenia in an ancestral African population, the South African Xhosa, appears in the Jan. 31 issue of the journal *Science*. An international group of scientists conducted the research, including investigators from Columbia University Mailman School of Public Health and New York State Psychiatric Institute, as well as the University of Cape Town and the University of Washington.

The study was carried out in the Xhosa population because Africa is the birthplace of all humans, yet ancestral African populations have rarely been the focus of genetics research. (There is no evidence that the Xhosa have an unusually high risk of schizophrenia). The researchers analyzed blood samples collected from 909 individuals diagnosed with schizophrenia and 917 controls living in South Africa. Their study revealed that participants with schizophrenia are significantly more likely to carry rare, damaging [genetic mutations](#) compared to participants without schizophrenia. These rare mutations were also more likely to affect brain and synaptic function. Synapses coordinate the communication between [brain nerve cells](#) called neurons; the organization and firing of neuronal synapses are ultimately responsible for learning, memory, and brain function.

The genes and pathways identified by this research inform the understanding of schizophrenia for all [human populations](#), the researchers say. Further studies in African populations might also suggest potential mechanisms for the design of more effective treatments.

"The presence of only a few DNA variations damaging to synaptic function could have an outsized effect on schizophrenia," says co-author Ezra Susser, MD, DrPH, professor of epidemiology and psychiatry at the Columbia Mailman School, Columbia University Irving Medical Center,

and New York State Psychiatric Institute. "While these variants differ from person to person, we believe they may disrupt neural pathways that elevate risk for schizophrenia."

The relative lack of genetics studies in Africa leaves a major gap in understanding [human genetics](#). Almost 99 percent of [human evolution](#) took place in Africa after the first modern humans originated and before humans migrated from Africa to Europe and Asia 50,000 to 100,000 years ago. Because of the lack of studies in Africa, many generations of human genetic history are missing from our understanding of human adaptation and of human disease. Studies of ancestral African populations like the Xhosa have more diverse background DNA, which facilitates the identification of truly rare mutations.

More information: S. Gulsuner et al., "Genetics of schizophrenia in the South African Xhosa," *Science* (2020).
[science.sciencemag.org/cgi/doi ... 1126/science.aay8833](https://science.sciencemag.org/cgi/doi/10.1126/science.aay8833)

Provided by Columbia University's Mailman School of Public Health

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