Brain Structure and Function in Genetic Disorders
UCLA - Departments of Psychiatry, Psychology, Neurobiology, and Genetics

UCLA has research opportunities for individuals over 18 years old with 22q11.2 Deletion Syndrome (Velocardiofacial Syndrome).

Background:
Many people with 22q11.2 Deletion Syndrome (Velocardiofacial Syndrome) have a greater chance of having attention deficits, learning disabilities, and particular psychiatric conditions such as autistic spectrum disorders. 22q11DS is caused by mutations in specific genes which affect brain development and can cause developmental delays and learning disabilities as well as certain physical problems like heart defects. One of the major obstacles in treating the disease is that scientists cannot study directly the nerve cells in the brain of individuals with 22q11DS. The purpose of the research is to create human induced pluripotent stem cells (iPSC) from a small skin sample in order to model the disease in a laboratory and better understand the structure and growth of nerve cells and their connections.

In this research, your skin will be sent to Stanford University and induced to become iPSC. The iPSC will then be induced to become nerve cells in order to model 22q11DS and possibly identify future therapies.

Study aims:
• To examine emotional adjustment, thought processes such as memory and attention, and brain structure and activity in individuals with 22qDS, as compared to those without the disorder.
• To create human induced pluripotent stem cells (iPSC) from skin samples. The iPSC will be induced to become nerve cells in the laboratory in order to model 22q11DS and possibly identify future therapies.
• To determine whether variation in the specific genes affected by 22qDS is related to differences in brain structure, function, and behavior.

Study procedures:
Study procedures include an MRI brain scan, clinical interview, tests of memory and attention, blood/saliva sample, skin sample and review of medical records. Participation may take between one to two days (~6-7 hours for all study procedures) per visit and includes 3 visits in total. All procedures will be explained carefully and all participation is completely VOLUNTARY.

Compensation:
Participants will be compensated up to $310 for full participation in all study visits and will receive a brief report about the test results from the clinical interviews and cognitive assessments.

For more information:
Please call Leila Kushan at (310) 825-3458 or email beardenlab.ucla@gmail.com