

Lay Summary

BRAIN UK Ref: 17/001

Pathological study of two cases with SLC52A3 mutation

Prof. Tamas Revesz, UCL Institute of Technology

Brown-Vialetto-Van Laere syndrome is a rare neurological disorder, named after three scientists, who first described this devastating condition. It affects the body's nervous system. In this disease, degeneration of the nerve cells takes place in parts of the brain which are essential for maintaining fundamental functions, such as movement and breathing. Most patients die at a young age because of the destruction of vital centres in the brain.

Recently, mutations in several genes have been identified in a group of patients with this syndrome. A gene is the basic unit of heredity, which determine the characteristics you inherit from your parents. Genes, which are made up of DNA, act as instructions to make molecules in the body called proteins. The proteins encoded by the mutant genes are known to be involved in normal function of mitochondria – small structures inside the cells, which are the main energy providers (also called powerhouses of the cells). In this study we are investigating whether the disease changes in the patients with this syndrome mimic the changes seen in patients with mitochondrial diseases.