MedicalXpress

New genetic cause of warburg micro syndrome identified

November 14 2013

A collaborative team of researchers led by researchers at the Medical College of Wisconsin and the University of Edinburgh has identified a gene responsible for Warburg Micro syndrome, a rare genetic disease characterized by eye, brain and endocrine abnormalities. Patients with Warburg Micro syndrome are severely physically and mentally challenged, unable to learn how to walk or speak and become blind and paralyzed from an early age.

The findings are published in *The American Journal of Human Genetics*. Lead co-authors are Ryan Liegel, Ph.D., postdoctoral student in cell biology, neurobiology and anatomy at the Medical College of Wisconsin; and Mark Handley, Ph.D., postdoctoral researcher at the University of Edinburgh. Corresponding author is Duska J. Sidjanin, Ph.D., associate professor of cell biology, neurobiology and anatomy and member of the Human and Molecular Genetics Center at the Medical College of Wisconsin.

In this study, the researchers became interested in a gene called TBC1D20, which is known to cause blindness and sterility in mice, because of that similar phenotype. The research team evaluated a cohort of more than 70 families with Warburg Micro syndrome, and found five distinct loss-of-function mutations in TBC1D20, thus establishing those mutations as a cause of the disease.

"These findings have implications not only for families affected with Warburg Micro syndrome, but also provide novel information about the

genes and molecular pathways essential for human development that is relevant for more common developmental disorders such as epilepsy and autism," said Dr. Sidjanin.

The four genes do not comprise the full causative picture for Warburg Micro syndrome; in about half of the cases, the causing mutation was in none of those genes, which means there are additional novel genes contributing to the disease.

The researchers plan to continue to search for additional genes, and will also model the disease in tissue cultures with a hope of understanding the underlying molecular and cellular events in which TBC1D20 is involved.

Provided by Medical College of Wisconsin