

UCSD/Egyptian Collaboration to Identify Genetic Causes of Mental Retardation

November 21, 2005 |

A promising collaboration between researchers at the University of California, San Diego (UCSD) School of Medicine and the Human Genetics and Genome Research Division at Egypt's National Research Centre in Cairo aims to help children with inherited mental retardation.

Brain development is genetically complex, with nearly half of all genes being expressed in the developing human brain. This complexity provides an ideal framework for understanding the roles of key genes during neuronal development, and the application of human genetics to the diagnosis and treatment of children with inherited mental retardation.

Autosomal recessive mental retardation, a neurological disorder caused by genetic factors that are not gender-linked, is common in Egypt because of the high number of first-cousin marriages, between 30 to 40 percent of all unions. This results in high rates of mental retardation and other inherited genetic diseases in that country. However, while the availability of genetic diagnostic tools has improved in Egypt in recent years, molecular research into autosomal recessive mental retardation and other related neurological disorders has remained limited.

The new partnership aims to change that, by enabling scientists in the two countries to work together to better understand the causes of these inherited diseases, and improve the ability of the Egyptian scientists to identify families at risk.

At UC San Diego, Joseph Gleeson, MD, Assistant Professor of Neurosciences and Director of the Center for Cerebellar Malformations, and his research team coordinate diagnosis and treatment of children with neuronal abnormalities, and study families of children with inherited disorders of brain development. In January, Gleeson headed a U.S. team based in the UC San Diego School of Medicine which visited the National Institute of Neuromotor System in Egypt. The Institute treats children most severely affected by inherited mental retardation and brain malformations. There, he met with Dr. Abdel Hamid Abaza, head of Egypt's General Organization of Teaching Hospitals and Institutes (GOTHI) to discuss plans for collaboration.

"MR is a complex problem faced by populations the world over. The Center for Cerebellar Malformations at UC San Diego was established to investigate the genes involved in childhood neurological disorders including mental retardation and autism. Now we have the opportunity to share what we have learned with Egyptian scientists. At the same time their contributions will expand our own research horizons," said Gleeson.

By October, researchers had taken the first official steps in establishing a collaboration to investigate the genetic basis of a common type of mental retardation.

"With our recent expansion in facilities there is a renewed emphasis in Egypt on the importance of proper diagnosis and treatment of these children," said Dr. Hatem. "We are excited to collaborate with Dr. Gleeson and his team on this important research project."

In the first stage of the proposed project, scheduled to be completed in 2006, Egyptian researchers will visit UC San Diego to learn genetic analysis methods for the study of their patients. In the second phase, the Egyptian team will establish a parallel facility there, assisted by U.S. team members who will travel to Cairo.

"Currently, families are being tested for genetic linkage of the recessive gene for mental retardation in our laboratory at UC San Diego. Our goal is to transfer this technology to Egypt, allowing the Egyptian research group to screen patients directly," said Gleeson. Intended as a long-term collaboration, the project will enable Egyptian scientists to later adapt the same genotyping technology to test patients and families with other genetic diseases.

The Egyptian research group is headed by Alice Abd El-Aleem, MD/PhD, and Maha S. Zaki, MD/PhD. Additional team members from the Human Genetics & Genome Research Division of the National Research Centre in Cairo include Ghada Abd El-Salam, MD, PhD, Mahmoud Yousry, BS, Eman Abbas, BS, and Hussam Ahmed, BS.

Researchers on the team from UC San Diego include Jennifer L. Silhavy, MS, Sarah E. Marsh, MS, and Carrie L. Louie, BS, all from the Neurogenetics Laboratory in UC San Diego School of Medicine Division of Neurology.

The study is funded by The National Institutes of Health with additional funding requested from the U.S.-Egypt Joint Science & Technology Fund, Joint Grants Division.

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