MedicalXpress

New registry to accelerate research on fragile X syndrome

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(PhysOrg.com) -- As researchers delve further into the genetic basis for disease, they face a conundrum: finding enough affected people who can fill out a true picture of mutations that can vary from one person to another. A case in point is fragile X syndrome, a genetic mutation that affects approximately one infant boy in 3,600 births, and one infant girl in 4,000-6,000 births.

Fragile X syndrome is the leading inherited cause of intellectual disability. Caused by a repetitive genetic error on the X chromosome, it is passed through families and can occur more frequently or severely in future generations. Beyond causing cognitive difficulties, scientists have in recent years discovered that people who carry the fragile X gene mutation can develop associated disorders, such as early menopause in women, and a neurological condition that mimics the movement disorder Parkinson's disease in adults as they age.

A new research registry, growing from a collaboration between the Waisman Center at the University of Wisconsin-Madison and the Carolina Institute on Developmental Disabilities at the University of North Carolina-Chapel Hill, is an attempt to "to streamline the process of connecting scientists with a large number of families affected by fragile X syndrome and its associated conditions," says Susan Vial, Waisman's coordinator for the registry.

Another objective is to help federally funded research centers work together and reduce the number of separate registries.

The registry, funded by the National Institute for Child Health and Human Development, will offer affected families the opportunity to take part in research at both centers on a wide range of topics, including family adaptation, brain development, language and genetic studies, Vial says.

Behavioral and educational studies may include parent surveys, individual assessments and observations. Medical studies may involve things such as DNA testing, brain imaging and clinical trials.

"Promising pharmacological and psychosocial treatments for fragile X syndrome are emerging, but their evaluation has been hindered by a lack of access to large and diverse samples of research participants," says Leonard Abbeduto, associate director of behavior science and director of the Center for Excellence in Developmental Disabilities at the Waisman Center. "We are taking steps to address this problem and are serving as a national demonstration project."

The registry is expected to set the stage for development of shared research registries for other neurodevelopmental disorders such as Down syndrome, Vial says.

More information: www.fragilexregistry.org/

Provided by University of Wisconsin-Madison