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Researchers track Huntington's disease progression using PET scans

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Investigators at The Feinstein Institute for Medical Research have discovered a new way to measure the progression of Huntington's disease, using positron emission tomography (PET) to scan the brains of carriers of the gene. The findings are published in the September issue of *The Journal of Clinical Investigation*.

Huntington's disease causes the progressive breakdown of nerve cells in the brain, which leads to impairments in movement, thinking and emotions. Most people with Huntington's disease develop signs and symptoms in their 40s or 50s, but the onset of disease may be earlier or later in life. Medications are available to help manage the symptoms of Huntington's disease, but treatments do not prevent the physical, mental and behavioral decline associated with the condition.

Huntington's disease is an inherited disease, passed from parent to child through a mutation in the normal gene. Each child of a parent with Huntington's disease has a 50/50 chance of inheriting the Huntington's disease gene, and a child who inherits the gene will eventually develop the disease. Genetic testing for Huntington's disease can be performed to determine whether a person carries the gene and is developing the disease even before symptoms appear. Having this ability provides an opportunity for scientists to study how the disease first develops and how it progresses in its early, presymptomatic stages. Even though a carrier of the Huntington's disease gene may not have experienced symptoms, changes in the brain have already taken place, which ultimately lead to severe disability. Brain imaging is one tool that could be used to track

how quickly Huntington's disease progresses in gene carriers. Having a better way to track the disease at its earliest stages will make it easier to test drugs designed to delay or even prevent the onset of symptoms.

Researchers at the Feinstein Institute used PET scanning to map changes in brain metabolism in 12 people with the Huntington's disease gene who had not developed clinical signs of the illness. The researchers scanned the subjects repeatedly over a seven-year period and found a characteristic set (network) of abnormalities in their brains. The network was used to measure the rate of disease progression in the study participants. The Feinstein Institute investigators then confirmed the progression rate through independent measurements in scans from a separate group of Huntington's disease gene carriers who were studied in the Netherlands. The investigators believe that progression networks similar to the one identified in Huntington's disease carriers will have an important role in evaluating new drugs for degenerative brain disorders.

"Huntington's disease is an extremely debilitating disease. The findings make it possible to evaluate the effects of new drugs on disease progression before symptoms actually appear. This is a major advance in the field," said David Eidelberg, MD, Susan and Leonard Feinstein Professor and head of the Center for Neurosciences at the Feinstein Institute.

More information: Metabolic Network as a Progression Biomarker of Premanifest Huntington's Disease, *J Clin Invest.* 2013;123(9):4076–4088. DOI: 10.1172/JCI69411

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