Genetic discovery may help determine effectiveness of Huntington's disease treatments

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A new genetic discovery in the field of Huntington's disease (HD) could mean a more effective way in determining severity of this neurological disease when using specific treatments. This study may provide insight for treatments that would be effective in slowing down or postponing the death of neurons for people who carry the HD gene mutation, but who do not yet show symptoms of the disease.

The work was led by researchers at Boston University School of Medicine (BUSM) and currently appears in *BMC Medical Genomics*.

HD is a fatal, inherited neurological disease that usually manifests between 30 and 50 years of age. The disease is caused by a genetic defect that is passed from parent to child in the huntingtin gene. Having too many repeated elements in the gene sequence causes the disease and an increasing number of repeats leads to earlier onset and increased severity of the disease.

The researchers studied the brains of people who died from HD and those who died of other, non-neurological diseases and identified a very specific genetic signal that strongly correlates disease severity and extent of neuronal, or brain cell death. The genetic signal, also called a microRNA, silences certain genes in the DNA. Genes that lead to the toxic effects of the huntingtin gene may be silenced by these microRNAs, in particular the miR-10b-5p microRNA.

"The findings that we found most interesting were the microRNAs that reflect the extent of the neuron death in the brain, since it is this process that causes the debilitating symptoms of the disease and eventually leads to the death of the individual," explained senior author Richard H. Myers, PhD, Director of the Genome Science Institute at BUSM.

According to the researchers these findings may represent a more effective way to tell whether or not HD treatments may be slowing down the pace of the death of brain cells. "If miR-10b-5p measurements can provide a faster and more effective way to determine whether or not a specific treatment is protecting brain neurons, it may be possible to study more potential treatments for HD more quickly. Equally importantly, it may become feasible to perform these trials in people who are HD gene carriers, but who do not yet show symptoms, by giving evidence for which trials may postpone onset and provide more healthy years of life," added Myers.

These findings also suggest that other microRNAs may also be important markers of severity for other neurological diseases such as Parkinson's disease and Alzheimer's disease. Further research is already being conducted in Parkinson's Disease by Myers and his colleagues.

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