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Japanese Monkeys Might Help In Curing A Fatal Inherited Disorder

A recent study published on July 23 in the journal *Neurobiology of Disease* has presented evidence about how a naturally occurring disease in a Japanese macaque monkey could serve as the first non-human primate model that will lead to a better understanding of the fatal neurodegenerative disorder called the Batten Disease.

Batten Disease- named after the British pediatrician *Frederick Eustace* Batten who first described it in 1903- and also known as neuronal ceroid lipofuscinoses (NCLs), is a fatal but rare nervous system disorder that usually starts in childhood. It is caused by genetic abnormalities that lead to the accumulation of fats and proteins called lipopigments in the cells of the brain, central nervous system, and retina in the eye, causing damage overtime.

Researchers have made a great progress in trying to identify the defective enzymes and mutated genes causing this disorder. Yet till this very moment there is no proof on how the gene mutations cause this build-up of lipopigments.

Scientists at the Oregon National Primate Research Center discovered a naturally occurring disease in a small population of Japanese macaque monkeys that is believed to be the only known model for batten disease among non-human primates in the world.

Batten Disease is caused due to a homozygous frameshift mutation in the CLN7 gene (CLN7−/−). In the Japanese macaques, which have been observed, CLN7−/− reduced the retinal thickness and retinal function, followed by profound atrophy in the cerebral and cerebellar over a five to six-year disease course, leading to progressive neurological deficits such as visual impairment, tremor, in coordination, ataxia and impaired balance.

In this study, scientists were able to identify a cohort of ten Japanese macaques bearing a haplotype that includes a frameshift mutation within exon 8 of the CLN7 gene (CLN7−/−).

Jodi McBride, Ph.D., assistant professor of neuroscience at the primate center, the lead author on the study, believes that this model should accelerate the ability to develop and test a gene therapy strategy to replace the normal version of the protein that is missing in this disease.

Although an enzyme replacement strategy delivering cerliponase alpha (Brineura™) helps to replace the TPP1 enzyme, it was only recently approved by the FDA in April, 2017. The NCLs treatments remain palliative.

“We don’t have great imaging biomarkers for this disease aside from the gold standard of MRI and so we’re also interested in using this new model to develop imaging techniques that will allow us to determine how successful we are at clearing out the buildup of cellular debris in the brain with potential treatments,” McBride said.

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