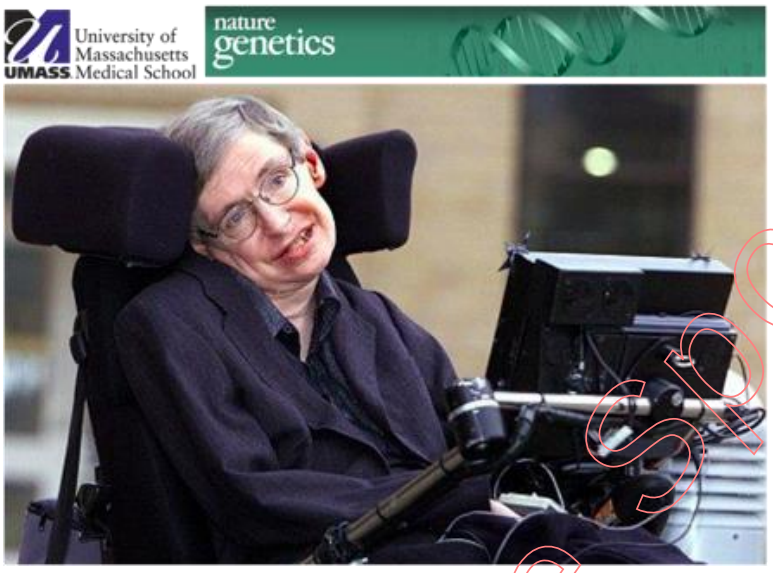
Genetic Science Spotlight

University of Massachusetts reports NEK1 gene associated with ALS



With the fund raised in the Ice Bucket Challenge that swept across the globe in 2014, a multinational collaboration led by the University of Massachusetts Medical School recently identified a significant association between amyotrophic lateral sclerosis (ALS) and the loss of function (LOF) variation of the NEK1 gene. The results were published in Nature Genetics in June 2016. The team of scientists performed whole exome sequencing (WES) for 1,022 familial ALS patients and 6,172 sporadic cases, and identified the candidate geneNEK1 with an odds ratio (OR) of 2.41, p=1.2E-7. The p.Arg261His variation of the NEK1 gene has the most prominent association among all, with an OR of 2.4, p=4.8E-5. The NEK1gene is responsible for maintaining the cytoskeleton and the mitochondrion in neurons. Despite the ambiguity in the exact pathogenesis of the NEK1 LOF variation, scientists believe that it has provided a new target for therapy treatment, and helps better understand the triggers of the disease.

http://www.nature.com/ng/journal/v48/n9/full/ng.3626.html

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