Amyotrophic lateral sclerosis: New genetic insights
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(PhysOrg.com) -- In order to better understand the causes of amyotrophic lateral sclerosis (ALS), also known as Lou Gehrig’s Disease, a group of scientists at the Centre hospitalier de l'Université de Montreal Research Centre (CRCHUM) studied 29 genes involved in the development of motor neurons in 190 ALS patients from Quebec and France. The results of this large-scale study were recently published in the online version of Archives of Neurology.

"This is the first time a large-scale genetic study is carried out in the field of ALS. By focusing on this group of 29 genes specifically expressed in motor neurons, we found a high frequency of rare non-synonymous mutations in our cohort of ALS patients." says senior author and researcher at CRCHUM, Dr. Guy Rouleau.

"The significant excess of these harmful non-synonymous mutations that change the structure of proteins, suggests the presence of ALS specific mutations," adds Dr. Rouleau, also a professor at Université de Montréal (UdeM). Proteins are gene products.

This group of 29 genes has been recently identified as specifically involved in the development of mouse motor neurons. "The analysis of these 29 genes led us to identify several promising novel genes for ALS. However, these genes require careful evaluation for the moment as further studies are needed to definitely confirm their implication in ALS," says lead author Dr. Hussein Daoud, postdoctoral fellow at CRCHUM and UdeM.

"Our study nevertheless provides new insights into the genetic causes of ALS, which could open novel avenues for research into the pathogenesis of this devastating disease."

Amyotrophic lateral sclerosis is a neurodegenerative disease that leads to a progressive paralysis due to the selective loss of motor neurons in the brain and spinal cord. It typically leads to death three to five years after symptoms onset. Approximately 2,500 to 3,000 Canadians live with ALS. Although the discovery of several genes has led to significant new insights into the causes of ALS, the basic pathogenic mechanism and genetic cause of most ALS cases remain unknown.


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