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Prognostic biomarkers of Amyotrophic Lateral Sclerosis (ALS): A step forward in the understanding of the disease

Ana Cristina Calvo

University of Zaragoza, Spain

Amyotrophic lateral sclerosis (ALS) is a neurodegenerative disease of unknown origin that causes progressive muscle paralysis and motor neuron death. The need of reliable biomarkers of ALS that can be accurately monitoring along disease progression is an increasing field of research. In this sense, our main objective is to identify molecular biomarkers as key elements of the induced neurodegeneration in ALS. Previous studies in our research workgroup analysed the transcriptional expression of a group of genes, whose expression was found up and down-regulated significantly in a preliminary microarray study and in muscle biopsy samples from transgenic SOD1G93A mice, the best characterized murine model for the disease. In this study we identified five genes, *Mef2c*, *Gsr*, *Col19a1*, *Calm1* and *Snx10* as potential genetic biomarkers of longevity in this animal model. Next, we translated this study to ALS patient's samples to validate the potential nature of these biomarkers. Skeletal muscle biopsies and blood samples from sporadic and familial ALS patients were analyzed by real time PCR and Western blot to test the expression levels of fifteen genes and fourteen proteins. ROC curves, multinomial regression and time-dependent Cox regression analysis were performed. *COL19A1* gene and protein levels were identified as potential prognostic candidates in skeletal muscle samples from ALS patients. In addition, the same gene improved prognosis in blood samples from sporadic ALS patients. These findings provide an important first step towards the accurate prediction of potential biomarkers in ALS be the initial springboard to new clinical trials and promising therapeutic strategies.

Biography

Ana Cristina Calvo has completed her PhD in 2003 from the Anatomy and Human Histology Department in the University of Zaragoza in Spain and Post-doctoral studies from the ALS Unit in the Hospital 12 October in Madrid (Spain) and from Faculty of Medicine in the Universidad Autónoma in Barcelona (Spain). She is an Associate Professor in Genetics and member of the Laboratory of Genetics and Biochemistry (LAGENBIO, IA2-IIS) in the Faculty of Veterinary Sciences in the University of Zaragoza (Spain). She has published 30 papers in reputed journals and participated as a coauthor in three licensed patents.

accalvo@unizar.es

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