

Conclusions

These data suggest that lncRNAs might be important players in muscle dysregulation in ALS and are candidate for the study of mechanisms contributing to ALS pathogenesis.

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119400

Diagnostic and prognostic role of plasma phosphorylated neurofilament heavy chain (pNf-H) in amyotrophic lateral sclerosis

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Background and aims

The phosphorylated neurofilament heavy chain (pNf-H) is a promising putative biomarker in Amyotrophic Lateral Sclerosis (ALS). We examined pNf-H concentrations in plasma of ALS patients to investigate its potential as a diagnostic and prognostic biomarker.

Methods

Incident ALS cases enrolled in a population-based registry were retrospectively selected and matched by sex and age with a cohort of healthy volunteers. Plasma pNf-H levels were measured by an ELISA kit (Neurofilament pNf-H-high sensitive ELISA, Euroimmun) and correlated with clinical parameters. Discrimination ability of pNf-H was tested using Receiving Operating Characteristic (ROC) curves. Kaplan-Meier (KM) univariate analysis was carried-out to determine the survival probability based on plasma pNf-H; univariate and multivariable Cox proportional hazard models were performed to estimate the effect of plasma pNf-H on survival, adjusted for clinically relevant covariates.

Results

Plasma pNf-H was significantly higher in patients compared to controls. An optimal cut-off of 39.74 pg/ml discriminated cases from controls with 75% sensitivity and specificity. Bulbar-onset cases had higher levels of plasma pNf-H compared to spinal onset ($p = 0.0033$). Furthermore, plasma pNf-H positively correlated with disease progression rate ($r = 0.19$, $p = 0.031$). Survival probability was no different among cases stratified according to pNf-H median value (101 pg/mL) (log-rank test $\chi^2 = 0.5$; $p = 0.49$). In Cox PH regression models plasma pNf-H was not independently associated with a reduced survival in cases (aHR 1, 95% CI 1–1, $p = 0.028$).

Conclusions

Our findings confirmed the potential utility of plasma pNf-H as a diagnostic biomarker in ALS. However, further evaluations in longitudinal data are needed to corroborate its prognostic value.

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119401

Professional and personal insights on neurodegenerative disorders – A world survey for professionals during the COVID-19 pandemic

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Background and aims

Neurodegenerative disorders are associated with high burden for patients, their families and the society, with overload of the professionals. We addressed the professional and personal insights and expectations of the latter, including during the COVID-19 pandemic.

Methods

A survey was developed and made available in google forms (<https://docs.google.com/forms/d/1-Gv5grtI8qb1r7C7ChpJnxs-a4ynIS6sGMu-70wm3Ss/edit#responses>), between 26/10–15/11/2020, to professionals working in the neurodegenerative field. Three sections were considered, addressing: characterization of the respondents; patient-professional relation; pathophysiology, genotype-phenotype, pharmacological perspectives. Means and frequencies were calculated. Differences between those working >15 vs ≤15 years were assessed using Fisher exact test. $P < 0.05$ was significant.

Results

Eighty-two professionals, from 5 continents, most Europeans, answered. Most were fulfilled with work, 47 worked >15 years, 67% for ≥4 days/week. Clinicians described positive impact on patients/families. Almost all had strategies to cope with the diseases' devastating nature, not affected by the COVID-19 in 44.3% or sometimes in 35.7%. Causative genetic interplay, protein aggregations and common neurodegenerative pathways, with ageing impact, justified a partial or continuous spectrum, with high levels of congruence between respondents ≤15 vs >15 years/work. Most believed in disease improvement, available within 5 years, but a cure only in the far future.

Conclusions

Professionals working in the neurodegenerative field are satisfied and motivated. Coping strategies may, nevertheless, be influenced by the COVID-19. Causative genetic interplay, protein aggregations, common neurodegenerative pathways and ageing play relevant roles, and justify partial/continuous spectrum, with high levels of congruence between professionals. A cure is not expected in the near future, but hope exists.

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119402

Clinical and genetic spectrum of amyotrophic lateral sclerosis in a Tunisian series

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