Identification of genetic mechanisms at the basis of the disease and, as critical objective, of the discovery of possible therapies

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During the period under review, we observed 18 patients suffering from ALS (6 men and 12 women). All the patients met the "El Escorial" requirements for the defined, clinically possible or possible with laboratory support ALS diagnosis. For each patient, we collected their personal details, sex, date and place (bulbar/spinal) in which the symptoms began, duration of the disease, from its beginning to the last visit. In addition, for each patient, the working history was collected, including the type of job, the possible risk factors they were exposed to, as well as the family history to discover any link with the disease. The follow-up envisages check-ups every 2 months with the recording of clinical parameters, functional status using the ALS-FRS-R scale as well as the assessment of the SVC.

For this reason, a database was created for the examined patients, containing all the phenotypic characteristics to correlate to bioumoral data.

The average age of the sample is 68.22 years ± 13,54 (SD). In 1 patient the disease had a bulbar beginning, whereas in 17 it had a spinal beginning. All patients gave their informed consent.

The development of the disease was calculated using the performance of the ALS-FRS-R scores. Patents were subdivided into thirds which proved useful to group patients with different development stages of the disease.

Accordingly, blood samples were collected from patients and from a control group (people suffering from neurodegenerative diseases such as Parkinson's disease, multiple system atrophy and progressive supranuclear palsy). Each sample was coded in order to maintain the privacy of patients and checks and was sent to the OIRM ASO unit of the Turin Sant'Anna hospital for analysis.

In addition, to study the possible pathogenic role of the genes identified as potential risk factors for ALS, the expression of such genes is being studied. This is achieved measuring the levels of mRNA or proteins (if there is an available antibody) in the peripheral blood mononuclear cells (PBMC) of patients suffering from ALS. They are compared with data obtained from the control population. The mRNA is measured via the real time PCR and the 'immunoblot' proteins. This study is very useful because if changes in potentially susceptible gene expression were found in the peripheral blood of ALS patients, it could be possible to characterise the pathogenic mechanisms of ALS and develop laboratory markers to diagnose the disease.