

Amyotrophic lateral sclerosis (ALS) is a degenerative disease of the nervous system which selectively damages motor neurons I and II. ALS is still considered a rare disease and affects both male (slightly more frequently) and female patients of all ages – but mainly during the seventh decade of life. The disease causes a progressive deficit of spinal motor functions (atrophy, cramps, fasciculations, spasticity and loss of muscular strength affecting the four limbs), respiratory functions (affecting the diaphragm and intercostal muscles) and bulbar functions (i.e. the muscles innervated by cranial nerves causing dysphonia, dysarthria and dysphagia ).

Median survival is about 3-4 years as of the onset of symptoms. Death is caused by respiratory failure for pneumonia – often ab ingestis (ingestion of food through the respiratory tract) – or progressive failure of respiratory muscles.

According to the ALS Register of Piedmont and Valle d’Aosta, the incidence of ALS is of 3.0 cases per 100,000 people (that is, around 130 new cases per year in Piedmont and Valle d’Aosta) and its prevalence is 8.0 cases per 100,000 people (approximately 450).

Approximately 10% of cases have genetic origin: numerous causal genes are known nowadays – the most frequent are *C9ORF72*, *SOD1*, *FUS/TLS*, *TARDBP*. 10-15% of patients are likely to develop frontotemporal dementia.

The cause of ALS is still unknown and no etiologic treatment is available, with the only exception of riluzole which, however, proved effective only to slow down the progress of the disease. Nevertheless, numerous symptomatic therapies are available and adequate care can be provided to ALS patients as long as they are totally assisted by specialized centers.

Over the past years, CRESLA – the Turin-based Regional Center Specialized in ALS run by Professor Adriano Chiò – has been developing its activity enormously both in terms of workload and diversification of targets. As of its creation, its main operational goals have been care and clinical research:

- a) The number of scientific publications is constantly increasing, featuring high-level productivity both quantity and quality-wise. Moreover, numerous scientific contributions have been submitted during significant national and international conferences.
- b) Several in-house research projects are underway, involving collaboration with both the interdisciplinary research group of CRESLA-Turin and external bodies such as the Italian study group on ALS, EURALS, NIH and others. CRESLA is focusing in particular on the following topics: studies on molecular genetics and genome wide association screening (GWAS); epidemiologic clinical studies (prospect population registry; risk factors like sport – in particular soccer; studies on stress and traumas assessment; studies on body weight and lipid asset); clinical studies on the evaluation of respiratory and diet parameters; psychological clinical studies; neuropsychological and quality of life clinical studies; research studies on diagnosis and prognosis markers; neurophysiologic studies; experimental clinical studies (see bullet point d).
- c) The number of out-patients and patients treated at home providing an institutionalized service of interdisciplinary specialist home-care is constantly increasing. This has been the first home-care service provided in Italy and is currently sponsored by a project of the Region Piedmont. The number of patients acceding from outside the Region is increasing as well, amounting to 1,100 outpatients and 500 home-care visits per year. Furthermore, hospitalization for diagnostic and therapeutic purposes and for executing procedures like radiologic percutaneous gastrostomy is a large part of care activities. The CRESLA team was the first in the world to demonstrate that percutaneous gastrostomy is most suitable for ALS patients.
- d) Numerous national and international therapeutic trials are currently underway.

In order to support CRESLA activities, the Vialli and Mauro Foundation funded a position for a university researcher in Neurology at the University of Turin. The competition note for two positions of researcher in Neurology – one of which funded by the Vialli and Mauro Foundation – code 725\_R (scientific sector MED 26) was published for projects aimed at researching lateral amyotrophic sclerosis (ALS).

Dr. Andrea Calvo was the assignee of the position and started his work as a university researcher in Neurology at the University of Turin on December 15, 2010. Thanks to this position, Dr. Calvo could continue his research and care activity on lateral amyotrophic sclerosis (ALS) started in 1997 under the supervision of Professor Chiò.