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Amyotrophic Lateral Sclerosis Descriptive Studies: Not Only Rates and Frequencies

Giancarlo Logroscino

Neurodegenerative Disease Unit, Department of Neurosciences and Sense Organs, University of Bari, Bari, and Department of Clinical and Research Neurology, Azienda Ospedaliera Pia Fondazione Panico, Tricase, Italy

In this issue of *Neuroepidemiology*, Drigo et al. [1] assess the incidence and prevalence of amyotrophic lateral sclerosis (ALS) in Friuli Venezia Giulia, a north-eastern region in Italy, with a population of about 1.2 million, bordering Slovenia to the east and Austria to the north. This is an aging population with about 22% of subjects over 65 years old.

The study design was retrospective within a population-based network of multiple sources. The period of observation lasted almost a decade, from 2002 to 2009. The methodology used in the Friuli registry is similar to the registries of EURALS, the consortium of European population-based registries [2–6] in two main characteristics: (1) the reference population is large in a well-defined geographic area, and (2) multiple sources were used for case ascertainment, including the Regional Health Database, a good example of the efficacy of administrative data for epidemiological research. The authors did not use the El Escorial criteria for the classification of cases, and the design was not prospective. However, the use of an extensive search for sources for case inception made the final results of this study on incidence (2.7/100,000) similar to the incidence found in most recent studies in Europe from the Piedmont (2.90/100,000) [7] and Ireland (2.0/100,000) [8].

All recent studies show that the incidence is quite homogeneous in Europe and the USA [9]. The most likely estimates of the incidence are between 2 and 3 cases per 100,000 person-years. The cluster of Guam disappeared some years ago, and the rates in Guam since the late 1980s are similar to most of the other regions in the world [10]. A recent study showed the highest incidence in the Kiji peninsula in Japan, in the city of Oshima, with 9.45/100,000 (95% confidence interval 7.39–26.29). This report, however, is based on a long period of observation in a very small base population (20,000 inhabitants).

An incidence rate of 1.4/100,000 was reported both in Catalonia [11] and in Sicily [12] similar to what has been reported previously in Apulia (1.7/100,000). Overall, the incidence in these southern registries is about half of what has been reported in Friuli and Piedmont. It is possible that the incidence of ALS in some areas around the Mediterranean is lower than in central and northern Europe.

The reported prevalence in this study is around 5/100,000, lower than we expected based on other studies and the median survival of ALS patients. Similarly to the incidence, the prevalence peak is in the older subjects between the ages of 65 and 74 years. A recent survey from the province of Modena (Italy) indicates almost doubling of the prevalence figures in recent years (from 5.8/100,000 in the year 2000 to 11.2/100,000 in the year 2009). All these data confirm that the ALS epidemiological figures are expected to go up in the future years with the aging of the general population or hopefully also due to the improved survival. The prevalence is important for the organization of resources in secondary and tertiary health systems to plan the health care of these patients.

Based on this experience, in Friuli the authors are building up a population-based registry that will join EURALS. In an ideal setting at least part of these population-based registries should be the basis of a system of permanent surveillance of ALS. Surveillance is defined as ‘the ongoing systematic collection, analysis, interpretation, and dissemination of data, reflecting the current status of a community or population’ [13]. This may be considered excessive for a rare disease like ALS, but this is the basis to disentangle the etiology and also to find a better therapy.

Population-based studies have been useful in defining clinical characteristics and prognostic indicators in ALS [14]. However, many new epidemiological questions remain that probably cannot be resolved by any of the existing population-based data sets. The recent reports about C9ORF76 and the heterogeneity of the clinical phenotype [15, 16] indicate that the attention of future registries should shift from pure motoneuron disease to more complex features involving cognitive impairment. Unfortunately, although not insurmountable, the obstacles for such studies both in terms of implementation and interpretation will be considerable. Many hints from the study of Drigo et al. [1] show that ALS descriptive studies are not only about rates and frequencies.

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