Prospective epidemiological registers: a valuable tool for uncovering ALS pathogenesis

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The understanding of the cause of amyotrophic lateral sclerosis (ALS) remains elusive, despite the increase in the number of published papers on this disease in the two last decades. Epidemiological studies have given important insight into ALS frequency and clinical characteristics. It is now established that ALS is similarly distributed in industrialised countries, including Europe, USA and Japan; its incidence has not changed over the two last decades and its classification as an age-related disease (ie, the product of pathogenic noxae acting at specific ages) versus an ageing-related disease (ie, a selective impairment of specific neuronal systems/pathways related to brain ageing) has now been established.1

In this context, the paper by Huisman and colleagues2 (see page 1165) represents an excellent application of a modern epidemiological approach, based on sophisticated methodologies, including a prospective design, the use of multiple sources of data, an ongoing diagnosis verification and an accurate follow-up of cases. This study design represents a decisive advancement compared with the classical epidemiological methodology, in particular for the evaluation of rare disorders. The completeness of case ascertainment related to this methodology allows the analysis of the full clinical spectrum of the ALS population, in particular the old or very old cases, and those with atypical/incomplete clinical presentations.3 Moreover, this methodological approach avoids the selection of particular subpopulations of patients, a typical bias related to the clinical series from tertiary referral centres. This bias may also greatly influence the results of studies on biological markers, as demonstrated by the largely different findings on the putative protective effect of the KIFAP3 gene polymorphisms in series from referral centres and from epidemiological series.4

ALS epidemiological registers are now rapidly moving beyond their original aims (assessing the incidence and prevalence, the temporal and geographical trends and the environmental risk factor of ALS) to become the core of collaborative international biorepositories of clinical data and biological specimens. These epidemiological biorepositories, which will encompass the whole spectrum of ALS phenotypes, will be instrumental for the study of the biological bases of ALS, through integrative analyses of genomics, proteomics, metabolomics and exposomics data, as well as clinical and environmental factors, to get closer to the goal of developing novel effective therapies for this devastating disease.

Competing interest None.

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