HHS Public Access

Author manuscript

JAMA. Author manuscript; available in PMC 2015 September 17.

Published in final edited form as:

JAMA. 2014 September 17; 312(11): 1097–1098. doi:10.1001/jama.2014.9799.

Quantifying a Nonnotifiable Disease in the United States:

The National Amyotrophic Lateral Sclerosis Registry Model

D. Kevin Horton, DrPH, MSPH,
Agency for Toxic Substances and Disease Registry, Atlanta, Georgia

Paul Mehta, MD, and
Agency for Toxic Substances and Disease Registry, Atlanta, Georgia

Vinicius Antao, MD, MSc, PhD
Agency for Toxic Substances and Disease Registry, Atlanta, Georgia

Public health surveillance is an essential tool for assessing, controlling, and preventing disease. In the United States, public health surveillance has evolved from a focus on monitoring infectious diseases to also tracking injuries, chronic diseases, birth defects, environmental and occupational exposures, and risk factors.[1] Despite this evolution of surveillance topics, many conditions still are not notifiable to federal public health officials nor are there surveillance systems in place to capture such conditions. The lack of morbidity data for nonnotifiable conditions makes it difficult to access accurately the populations at greatest risk and the true economic and societal burden of such diseases. New approaches are needed to more accurately quantify nonnotifiable conditions of interest in the United States, such as amyotrophic lateral sclerosis (ALS).

ALS, also known as Lou Gehrig disease, is a rare disease that affects the upper and lower motor neurons and usually leads to death within 2 to 5 years after diagnosis.[2] In 2008, the US Congress passed the ALS Registry Act,[3] intended to create a national ALS registry to better describe the incidence and prevalence of ALS, define demographic characteristics of those with ALS, and examine risk factors that may be related to the development of ALS, such as environmental and occupational exposures. Even though the act did not make ALS a notifiable disease, it did allow for the creation of a national population-based registry to collect and analyze data regarding persons living with ALS in the United States. In 2009, the federal Agency for Toxic Substances and Disease Registry (ATSDR), a sister agency of the Centers for Disease Control and Prevention (CDC), launched the National ALS Registry. This registry undertook a novel approach to quantify the burden of ALS using data from national administrative sources and a self-enrollment web portal. Data from both sources are merged and de-duplicated to ensure an accurate case count.

Corresponding Author: D. Kevin Horton, DrPH, MSPH, Agency for Toxic Substances and Disease Registry, 4770 Buford Hwy, Mailstop F-58, Atlanta, GA 30341-3717 (dhorton@cdc.gov).

Conflict of Interest Disclosures: All authors have completed and submitted the ICMJE Form for Disclosure of Potential Conflicts of Interest and none were reported.

Disclaimer: The conclusions of this article are those of the authors and do not necessarily represent the views of ATSDR, CDC, or the US Department of Health and Human Services.

To date, much of the population-based epidemiology on ALS has been derived from other public health surveillance systems, most notably the well-established European ALS registries (eg, in England, Ireland, Italy, Scotland). These registries, which have been in existence for decades, are able to identify ALS cases because the European health care systems facilitate visits by ALS patients to neurologists, who in turn report these cases to their respective national or regional ALS registries.[4] Because of differences in health care delivery in the United States, ATSDR researchers had to devise a different mechanism for identifying ALS cases to fulfill the intent of the ALS Registry Act.

Pilot testing for the registry indicated that a 2-pronged approach could be used to identify all newly diagnosed and existing ALS cases in the United States. The first approach, launched in 2009, uses large, existing national administrative databases (ie, Medicare, Medicaid, Veteran Benefits Administration, and the Veterans Health Administration) to identify patients with ALS. Because there is no blood test for ALS and the diagnosis is largely based on signs, symptoms, and neurophysiologic tests (eg, electromyograms), this approach uses a pilot-tested algorithm that, when applied to the databases, separates patients into 3 categories: definite ALS cases, possible ALS cases, and non-ALS cases. Patients with definite cases are automatically entered into the National ALS Registry. Patients for whom there is not enough information to determine whether they have ALS are considered possible cases and are reevaluated when subsequent years of administrative data become available. If these patients are later determined to be definite cases, then they are entered into the registry. Patients who are determined not to be definite cases are not included in the registry.

The algorithm used to identify cases includes components such as the *International Classification of Diseases* (*ICD*) code for ALS (*ICD-9* 335.20), frequency of neurology visits, and prescription drug use (specifically for riluzole, the only US Food and Drug Administration–approved medication for ALS). Pilot testing of the algorithm (based on 4754 total patients: 1596 with definite ALS, 1432 with possible ALS, and 1726 with non-ALS) yielded a sensitivity of 86.8% (95% CI, 85.0%–88.4%) and specificity of 85.4% (95% CI, 83.6%–87.0%).[5]

For the second approach, ATSDR created a secure web portal that was launched in 2010 to identify cases that may not have been captured in the national databases,[6] because not all patients with ALS can be identified through existing national databases. Eligibility requirements may limit the capture of some ALS cases; for example, nonveterans will not be captured through the Veterans Affairs databases. The web portal allows patients to self-register into the system by answering prevalidated screening questions (eg, have you ever been told by a neurologist that you have ALS?). If the screening determines that a person has ALS, he/she can complete an online consent form, answer brief enrollment questions, and take risk factor surveys including measures such as occupational and environmental exposures, smoking and alcohol use history, disease progression, and physical activity. ATSDR is currently working with large ALS support groups in the United States (eg, the ALS Association, the Muscular Dystrophy Association) to raise awareness among ALS patients of the self-enrollment part of the National ALS Registry.

The July 25, 2014, issue of the CDC's *Morbidity and Mortality Weekly Report* includes the ATSDR's first national prevalence estimate of ALS in the United States.[7] Based on a total of 12 187 persons meeting the surveillance case definition of definite ALS identified by the registry, the estimated prevalence was 3.9 ALS cases per 100 000 persons in the US general population. Overall, ALS cases were more prevalent among whites, men, non-Hispanics, and persons aged 60 through 69 years. Although these estimates are comparable to those in Europe, US incidence estimates are not available from the registry because the national administrative databases do not contain a date of diagnosis for ALS and the date of diagnosis for patients who self-report in the web portal cannot be validated at this time. However, the registry is using active surveillance to determine incidence in 3 states and 8 metropolitan areas. The state/metropolitan data are also being used to assess the completeness of the registry data, along with capture/recapture methodology.

The National ALS Registry is also being used as a patient recruiting source for researchers who are conducting clinical trials and epidemiological studies. To date, 9 US institutions have used the registry to send e-mail messages to registry enrollees about ALS research studies in which they are eligible to participate. Additionally, a feasibility study is examining the creation of a national biorepository to collect biological specimens (eg, blood, saliva, and tissue) from registry enrollees. Linking the specimens to the survey information already being collected from registry participants could make the registry even more useful.

Even in 2014, many conditions are not surveilled in the United States, making it difficult to accurately assess their disease burden. The approach used for the National ALS Registry provides a novel method to quantify prevalence for a nonnotifiable condition on the national level in the United States. Moreover, it is possible that this approach, in part or whole, could be used to help determine the prevalence of other nonnotifiable conditions in the United States (such as Parkinson disease or multiple sclerosis).

References

1. CDC. CDC's Vision for Public Health Surveillance in the 21st Century. MMWR 2012. Jul 27; 2012 61(suppl):1–40. http://www.cdc.gov/mmwr/pdf/other/su6103.pdf.
2. Mitsumoto, H.; Chad, DA.; Pioro, EP. Amyotrophic Lateral Sclerosis. Philadelphia, PA: FA Davis Co; 1998.
3. US Public Health Service. ALS Registry Act; Washington, DC. 110th Congress. Pub L 110–373; October 2008; 42 USC 280g–7. http://wwwn.cdc.gov/ALS/Download/ALS%20Registry%20Act%20(Public%20Law%20110-373).pdf
4. Logroscino G, Traynor BJ, Hardiman O, et al. EURALS. Incidence of amyotrophic lateral sclerosis in Europe. J Neurol Neurosurg Psychiatry. 2010; 81 (4):385–390. [PubMed: 19710046]
5. Kaye WE, Sanchez M, Wu J. Feasibility of creating a National ALS Registry using administrative data in the United States [published online March 6, 2014]. Amyotroph Lateral Scler Frontotemporal Degener. 10.3109/21678421.2014.887119
6. [Accessed July 12, 2014] National Amyotrophic Lateral Sclerosis (ALS) Registry. http://www.cdc.gov/als
7. CDC. Prevalence of amyotrophic lateral sclerosis—United States, 2010–2011. MMWR Morb Mortal Wkly Rep. 2014; 63(SS–7):1–13. http://www.cdc.gov/mmwr/pdf/ss/ss6307.pdf. [PubMed: 24402465]