National ALS Registry:
A Multi-faceted Research Platform

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**Agency for Toxic Substances and Disease Registry**

Disclosures

- **The findings and conclusions in this presentation have not been formally disseminated by the Centers for Disease Control and Prevention/the Agency for Toxic Substances and Disease Registry and should not be construed to represent any agency determination or policy.**

Outline of Presentation

- **Multi-faceted ALS Research Platform**

- **ALS Registry Act/Reportable vs. Notifiable/Federal ALS Research Initiatives**

- **National ALS Prevalence Estimates**

- **Risk Factor Surveys**

- **National ALS Biorepository**

- **How We Support Clinical Trials & Epidemiological Studies**

- **Funded Research**

- **Impact of the Registry**

- **Conclusion**

National ALS Registry – A Multi-faceted Research Platform
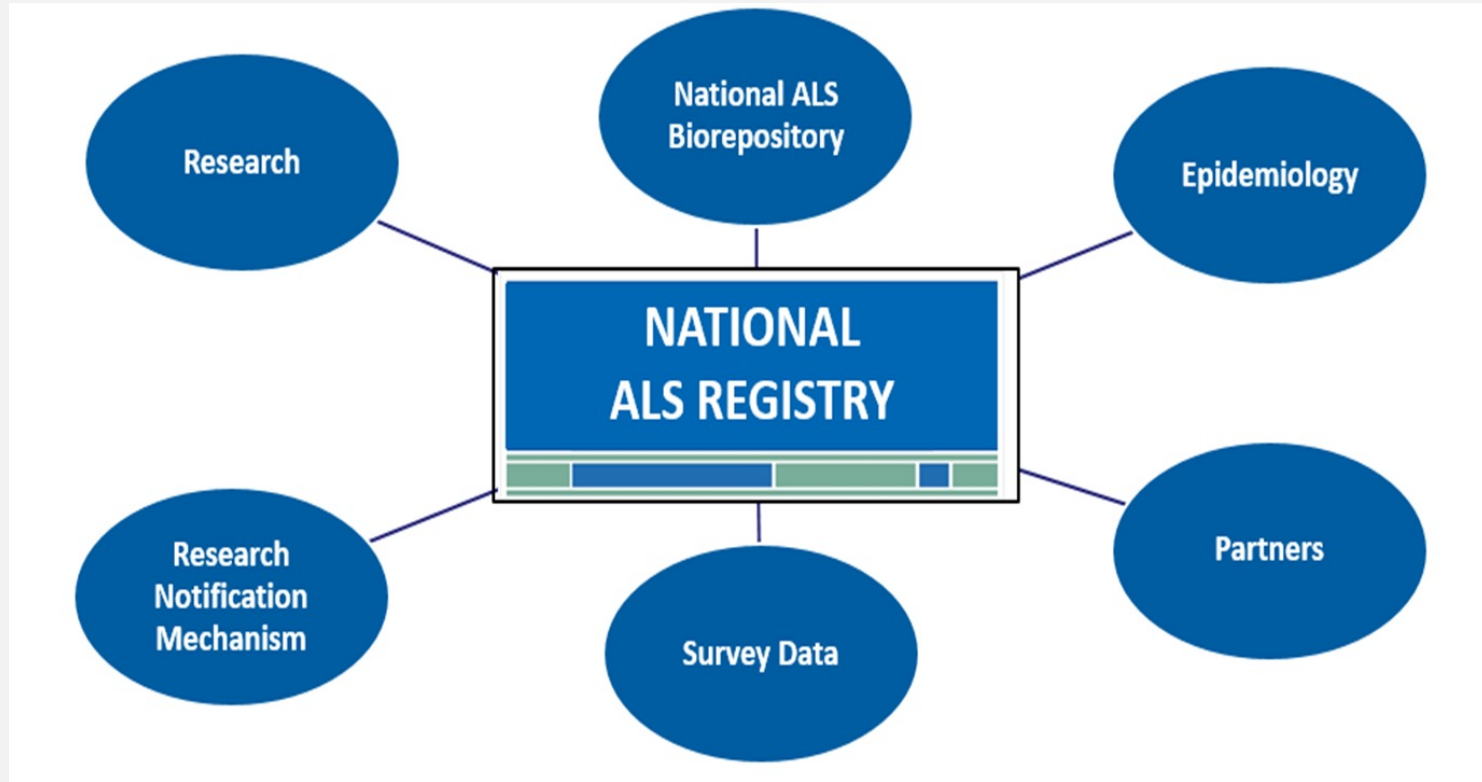
U.S. ALS Registry Act

- **Enacted as Public Law 110-373 (October, 2008)**

- **Directed CDC/ATSDR to create a population-based U.S. registry**

- **National ALS Registry launched in October, 2010**

- **Registry purposes (as specified by the Act)**
  - Describe incidence and prevalence of ALS
  - Describe the demographics of ALS patients
  - Examine risk factors for the disease

- **ALS is non-notifiable to CDC in the United States**

Reportable Versus Notifiable

- **Reportable diseases are mandatorily reported to jurisdictions by individuals in the health care community, including providers, facilities and laboratories[1]**
  - Each state determines which diseases/conditions must be reported
  - ALS is reportable in the state of MA, VT, ME, but not to the Registry
- **Notifiable diseases are reported to the CDC on a voluntary basis by each jurisdiction[1]**
  - Case records are de-identified and include limited information about the patient and the case
  - 120 diseases/conditions currently monitored
- **Novel case-finding methods are needed**

How many cases of ALS are there in the United States?

- **New method used to estimate the number of missing cases in the U.S.**
  - This is a first.

- **Revised estimates are as follows:**
  - Close to 32K (31,843) cases as of 2017
  - Rate of 9.9 per 100,000 U.S. population

- **Estimate we are missing 14,043 cases**
  - Missing patients receive care outside CMS/VHA/VBA*
  - Private insurances (e.g., PPOs, HMOs)**
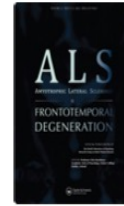
Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration

ISSN: (Print) (Online) Journal homepage: https://www.tandfonline.com/loi/iafd20

Prevalence of amyotrophic lateral sclerosis in the United States using established and novel methodologies, 2017

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*CMS: Centers for Medicare and Medicaid Services
*VHA: Veterans Health Administration
*VBA: Veterans Benefits Administration
**PPO: Preferred Provider Organization
**HMO: Health Maintenance Organization

Self-Administered Risk Factor Surveys

- **Helps researchers find clues about ALS:**

  - Occupations: are certain occupations at higher risk for ALS?

  - Military service: why are veterans at a greater risk of getting ALS?

  - Physical activity, smoking and drinking: are these risk factors?

  - Residential history and residential pesticide use: are these risk factors?

  - Traumatic brain injuries: are these risk factors?

  - Disease progression: how is ALS progressing (fast or slow) and what is affected?

  - Open ended: patient perspectives on disease cause

  Take surveys in any order (caregiver help is always appreciated)

  ## HELPS US SOLVE THE MYSTERIES BEHIND ALS

National ALS Biorepository Overview

Help researchers in the area of ALS:

- **Genetics:**
  - Identification of genes such as SOD1 and C9orf72 and others

- **Biomarker identification: indicator of a biological state or condition**
  - Help us to examine disease progression (how fast or slowly ALS progresses)
  - See if certain drugs will work or not work
  - Possibly identify if a simple blood test could catch ALS early

- **Environmental factors (analyzing patient's blood and saliva):**
  - Heavy metals (lead, arsenic, and others)
  - Persistent organic pollutants (e.g., pesticides, insecticides, PCBs, DDT): tough to degrade/breakdown

Details on the National ALS Biorepository

- **Biorepository is significantly different from others**
  - Extensive risk factor survey data linked with biosamples
  - Nationally representative, that is, beyond referral centers
  - User-friendly to person with ALS, that is, phlebotomists come to your home to collect samples
  - Specimens collected specifically for biorepository, that is, we do not use leftover study samples to constitute biorepository
  - Pre/post-mortem samples kept in one central biorepository
- **No charge for patients - user friendly - we come to your home**
- **Largest collection of pristine ALS samples for research, e.g., genetics, biomarkers, disease progression**

We Support Patient Participation in Clinical Trials and Epidemiological Studies

- **Patient recruitment for research can be difficult and challenging**

- **Approx. 95% of Registry PALS want to participate in research**

- **Registry links PALS with scientists who are recruiting for research (e.g., clinical trials, studies)**

- **Domestic and international researchers are using the tool for recruitment purposes**

- **Over 70 institutions have used it**

NATIONAL ALS REGISTRY

ALS
research
counts on you.

CDC ATSDR

U.S. Department of
Health and Human Services
Centers for Disease
Control and Prevention
Agency for Toxic Substances
and Disease Registry

Registry Funds Research

- **ATSDR is funding extramural research to learn more about ALS etiology and risk factors**

- **24 research studies have been funded to date**

- **Information gleaned also will help ATSDR prioritize topics for future risk factor surveys**

- **Funded 2 awards in September 2022**

- **New funding opportunity announcement in 2023**

Areas of Research Funded

- **Studies range from:**

  - Identifying environmental risk factors such as cyanobacteria (harmful algal blooms)

  - Analyzing pesticides and pollutants

  - Identifying biomarkers and genes

  - Evaluating the human virome (400 viruses) for antibodies

  - Genetics: why certain populations have lower rates of ALS than others

  - Military service and the risk for ALS

  - Evaluating traumatic brain injuries and chronic neuroinflammation

  - Oxidative stress and environmental risk factors

  - Evaluating the gut microbiome

  - Conducting whole genome sequencing of Biorepository samples

Current and Past Funded Institutions

Northwestern University *(Siddique)*	Harvard University *(Weisskopf & Ascherio)*
Columbia University *(Schneider & Mitsumoto)*	Dartmouth College *(Stommel)*
University of Michigan *(Feldman & Murdock)*	University of Pittsburgh *(Talbott)*
Trinity College *(Hardiman)*	Karolinska Institute *(Fang)*
Stanford University *(Nelson)*	University of Missouri *(Song)*
University of Miami *(Benatar)*	National Institutes of Health *(Traynor)*

Impact of the National ALS Registry

- **Largest database of ALS patients in the United States for research**
- **Collaborating with pharmaceutical companies and academia to provide recruitment assistance for clinical trials and epidemiological studies**
- **National ALS Biorepository is advancing ALS research on biomarkers, genetics, and environmental exposures**
  - User friendly and free system for patients to participate in research
- **Funding research grants for leading academic institutions in order to learn more about risk factors and possible etiologies**
- **Determining national epidemiological trends such as how many cases of ALS in the United States**
- **Looking to solve the who, the what, and why of ALS**

Comments/Questions

www.cdc.gov/als

For more information, contact NCEH/ATSDR
1-800-CDC-INFO (232-4636)
TTY: 1-888-232-6348   www.atsdr.cdc.gov   www.cdc.gov
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