Title: National Amyotrophic Lateral Sclerosis (ALS) Biorepository Feasibility Study

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Background: The Agency for Toxic Substances and Disease Registry (ATSDR) launched the congressionally mandated National ALS Registry to identify ALS cases in the United States. To enhance the Registry, ATSDR coordinated a pilot study to find the best way to collect and store biological samples for future ALS related research.

Objective: Determine the feasibility of developing a biorepository linked to the National ALS Registry.

Methods: The pilot study included two specimen collection components: 1) in-home collection of blood, urine, hair, and nails, on two occasions approximately six months apart; and 2) postmortem collection of brain, spinal cord, CSF, bone, muscle, and skin. A participant could do both components. Blood and urine specimens were processed into smaller aliquots. Brain and spinal cord were processed into fixed and frozen sections. Eligible participants must have enrolled in the National ALS Registry and consented to be contacted about research projects.

Results: Three hundred and thirty-nine people consented from all 50 states to donate biological specimens and 330 provided specimens at least once. Approximately 18% of participants did not complete the second draw due mostly to death and illness. Participants’ ages ranged from 31 to 87 years and 60% were male. Fifty-four percent of participants lived 50 or more miles from an ALS specialty/referral center. Thirty people consented to postmortem tissue donation from 18 states. Postmortem participants were 43 to 75 years of age and 50% were male. Nineteen donations have been completed. The first 18 cases examined showed neuropathologically-confirmed ALS with TDP-43 inclusions (ALS-TDP) with a mean age of 63 years (range: 43-76), 61% female, and mean brain weight of 1268g. Forty-four percent of subjects had co-morbid pathological diseases, including the neuropathological changes of Alzheimer disease, Lewy body disease, vascular disease with infarcts, encephalitis, and tumor.

Discussion/Conclusions: Creating a geographically diverse biorepository had unique challenges. However, based on expert input, ATSDR determined that, with some modifications to the pilot project, it is feasible to incorporate a biorepository into the National ALS Registry. These specimens will be a valuable resource for researchers who will be able to request specimens for ALS studies.