

## BRIEF FROM THE ALS SOCIETY OF CANADA

August 2017

### Executive Summary

The ALS Society of Canada (ALS Canada) is pleased to participate in the pre-budget consultation process, and appreciates the opportunity to share with the members of the House of Commons Standing Committee on Finance our thoughts on the future of the Canadian economy. This submission addresses one of the important needs for maintaining productivity and competitiveness in Canada – a healthy population. Disease and illness are responsible for a significant amount of cost to the Canadian health care system and the Canadian economy.

Amyotrophic lateral sclerosis (ALS) is a fatal neurodegenerative disease and the most common motor neuron disease in adults. ALS causes progressive paralysis due to degeneration of the upper and lower motor neurons in the brain and spinal cord. Most often, death is a result of the eventual paralysis of the muscles in the throat and diaphragm as someone loses the ability to swallow, cough and breathe. In 90 to 95 per cent of cases, ALS affects people with no family history of the disease. ALS has no known cure or effective treatment yet. Approximately 2,500 to 3,000 Canadians live with the disease at any moment in time, and 80 per cent of people with ALS die within two to five years of diagnosis.

The vision of ALS Canada is to make ALS a treatable disease by 2024. That vision can only be realized through a commitment to research. The ALS Canada Research Program is the only dedicated source of funding for ALS research in Canada, and it is funded entirely by donors, the majority of whom have a personal connection to ALS and are often already burdened by the financial costs of the disease. For a fatal disease like ALS that affects a relatively small patient community, the federal government has a duty to fund research – especially research that will have immediate impact, as we will explain further.

Through the strategic investment in ALS research that we are requesting, the federal government will be supporting a unique, coast-to-coast innovation that will support Canadian industry while also accelerating the path to improving the lives of Canadians with ALS and building a “living lab” of data that can be used to study of other diseases and disorders. Our recommendation is that:

**The federal government invest \$9.1 million so that every Canadian living with ALS (approximately 3,000) and a matching number of healthy Canadians can contribute to Project MinE, an international effort to determine the genetic underpinnings of ALS.**

Furthermore, our organization supports the submission of the Health Charities Coalition of Canada (HCCC), a member-based organization comprised of 27 national health charities who represent the voice of patients at all levels of the health continuum. The collective members translate knowledge gathered through research to advocate for better public policy and better health outcomes for Canadians. As a member of HCCC, we support their recommendations for greater access to medicines for all Canadians and for the investment in research that will provide jobs and stimulate the economy, setting the stage for improved prosperity and innovation in the future. Additionally, we support the recommendation for the Government of Canada to establish a formal mechanism for meaningfully and continuously engaging patient representatives in its decision making and regulatory processes on issues related to health and health research.

ALS Canada, founded in 1977, is the only national voluntary health organization dedicated solely to the fight against ALS and support for those with the disease. ALS Canada is the leading not-for-profit organization working nationwide to fund ALS research and, with the provincial ALS Societies, we all work to improve the quality of life for Canadians affected by ALS.

### **Supporting Information**

ALS research has come further in the past five years than the previous century. In 2017, most of the leading researchers and clinicians in the field believe we are getting significantly closer to understanding how ALS is caused, which will ultimately lead to treatments for this fatal disease. The pace of making ALS treatable can be rapidly increased through a targeted investment in an unprecedented global collaboration where Canada stands poised to take a leadership role with only modest investment, yet will be able to drive cross-country innovation and collaboration.

For the first time in history, it is feasible to identify the genetic signature that can lead someone without a family history of ALS (approximately 90 to 95% of cases) to develop the disease. Whole genome sequencing (WGS) makes this possible, and it is the focus of Project MinE ([www.projectmine.com](http://www.projectmine.com)), a groundbreaking, collaborative, multi-national effort to provide WGS for 15,000 people with ALS and 7,500 controls.

Whole genome sequencing used to be incredibly expensive: the human genome project cost approximately \$2.7 billion when it finished in 2000. Over time, costs have decreased to the point that new instrumentation at McGill University has enabled whole genome sequencing to take place for between \$1,300 and \$1,500 CAD per genome. In the case of Project MinE, additional costs of logistics for shipping samples and storing data also exist. In combination these costs make the total, global project an expensive one, but within the realm of feasibility, especially when countries work together to leverage funds and resources/samples. This leveraging of samples is even more important for a rare disease like ALS where no country could ever achieve this level of statistical significance alone.

In 2016, Canada became the 17<sup>th</sup> country to join Project MinE, beginning with a commitment from ALS Canada of \$150,000 to sequence approximately 100 genomes from stored samples of people no longer living. The Canadian component of Project MinE currently brings together the four leading ALS geneticists located in Vancouver, Toronto, Montreal and Quebec City for their first ever cross-Canada collaboration. Each has led or been part of international consortia that have resulted in some of the most important genetic discoveries in the field. They also represent a geographical balance that would provide a collaborative set of Canadian samples representative of ALS cases across the country.

To date, Project MinE has procured funding for 38% of the sequences and important discoveries have already been made. However, much more data is needed for the project to have the statistical power needed for many key genetic aspects of ALS. This means that unlike funding laboratory research projects, Canadian investment in Project MinE will quickly lead to results that will accelerate ALS research. Canada has achieved 7% of its goal for the project, giving the federal government an opportunity to play a leadership role that will enable the scientific community to better understand ALS and develop targeted treatments – while also supporting Canadian collaboration and innovation and building expertise and infrastructure that will lead to insights into other neurological conditions.

The federal government should provide this one-time investment in Project MinE because:

1. **The investment will provide a legacy for terminally-ill Canadians.** ALS is invariably fatal and not everyone qualifies or has access to clinical trials of experimental therapeutics, leaving many Canadians to die without the opportunity to contribute in some way to a future without the disease. Through investment in Project MinE, every Canadian diagnosed with ALS, despite their stage of disease, could provide a blood sample and have their genome sequenced.
2. **Project MinE would be a unique, coast-to-coast innovation that would support Canadian industry while also building expertise and infrastructure for future efforts in many diseases.** The nature of the Canadian healthcare system means that the majority of people living with ALS will be diagnosed and monitored at one of the 16 multi-disciplinary, networked ALS clinics across the country. Federal funding for Project MinE would therefore support Canadian industry and innovation as the sequencing would happen on Canadian soil, keeping funds in Canada while both investing in and showcasing Canadian innovation on an international stage. Collectively, executing this project would be simple, provide immediate impact and results that could be shared globally.
3. **Canada has an opportunity to provide critical research dollars that don't exist elsewhere.** Every year (with the exception of 2014), ALS research in Canada is almost entirely supported by grassroots fundraising initiatives. However, the pool of funds that can be raised from the community is usually very small, given the small patient population, the personal financial burden of the disease and the fact that there is no survivor community. When compared to larger chronic diseases, like cancer and cardiovascular, which have a large patient population and survivor community, and corresponding fundraising activities, it becomes quickly apparent that fundraising to support research is not a realistic option for ALS. The low prevalence also makes ALS less attractive to pharmaceutical companies and makes competition for CIHR grants more difficult, all resulting in a need for directed support from other sources like the federal government. The federal government's investment in Project MinE would single-handedly boost the initiative to nearly two-thirds of its goal (from 38% at present) and immediately lead to genetic discoveries that would help us to understand ALS better, leading to more rapid development of treatments.
4. **Whole genome sequencing of control samples can be used for studies in a wide variety of other conditions.** Federal funding for Project MinE would not only impact ALS significantly, but would provide valuable control data for other national and international initiatives. Already, Project MinE data have been shared with a Harvard study looking at human genetics, a stroke study in Maryland, a cardiac disease initiative in the Netherlands, and a large group called the Haplotype Reference Consortium that is examining genetic phenomena to understand human genetics in a non-disease capacity as well as how diseases relate to each other.

Undoubtedly, having Canadian controls in this study will be valuable as they will not only represent the diverse ethnic backgrounds of our country, but the open access nature of the data means it could be shared amongst many Canada-specific initiatives. Given this, federal government support of Project MinE will serve to not only help the world solve ALS, but it will provide a strong Canadian contribution to studies that will advance our knowledge of genetics in many other ways.

5. **Federal investment in ALS research now will reduce the need for or amount of future investment.** Many researchers believe the discovery of treatments to significantly slow ALS is a matter of "when" not "if" and that "when" depends of funding. Once there are treatment options for ALS and people

are able to live longer and better with the disease, a threshold will be crossed and opportunities will emerge that exponentially advance the ability to raise funds, boost investment in Canadian research from outside sources, and discover even better treatments. A population living longer with ALS means more access to grassroots funding and less reliance on government dollars. Industry starts to recognize ALS as a space with better potential for return on investment and begins to bring dollars to Canada. The massive burden on the healthcare system will be lightened as people will be better able to live in their homes and emergency room visits would lessen significantly. The ALS research community would achieve greater funding through industry partnership and as a result of government funding to a more prevalent population. All of these will further accelerate ALS treatment advancement in a positive feedback cycle resulting in further productivity of Canadians.

**Recommendation**

**The federal government invest \$9.1 million so that every Canadian living with ALS (approximately 3,000) and a matching number of healthy Canadians can contribute to Project MinE, an international effort to determine the genetic underpinnings of ALS.**

The \$9.1 million will allow for 3,000 ALS and 3,000 control samples to be collected in 16 clinics across Canada and then shipped to Quebec for analysis. This is a one-time investment and the collection and analysis is expected to take 24 months.

**Financial Breakdown**

| <b>Activity</b>                        | <b>Notes</b>                          | <b>Cost</b>        |
|--|---------------------------------------|--------------------|
| DNA extraction                         | \$30 per sample x 6,000 samples       | \$180,000          |
| DNA sequencing                         | \$1,473.15 per sample x 6,000 samples | \$8,838,900        |
| Shipping to Genome Quebec (in batches) |                                       | \$80,000           |
| <b>TOTAL</b>                           |                                       | <b>\$9,100,000</b> |