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Poster Abstract: Diagnostic

The Search for Pompe Patients in Canada: Assessing Feasibility of a National Disease Registry to Facilitate Research

Lawrence Korngut[1,*], Megan Johnston[1], Craig Campbell[2], Angela Genge[3], Alex MacKenzie[4], Anna McCormick[5] and Shannon Venance[6]
[1]*Department of Clinical Neurosciences, University of Calgary Hotchkiss Brain Institute, Calgary, Canada*
[2]*Department of Pediatrics and Clinical Neurological Sciences, Western University, London, ON, Canada*
[3]*Clinical Research Unit, Montreal Neurological Institute, Montreal, QC, Canada*
[4]*Department of Pediatrics, Children's Hospital of Eastern Ontario Research Institute, University of Ottawa, Ottawa, ON, Canada*
[5]*Department of Physical Medicine and Rehabilitation, Children's Hospital of Eastern Ontario, University of Ottawa, Ottawa, ON, Canada*
[6]*Department of Clinical Neurological Sciences, Western University, London, ON, Canada*

BACKGROUND

The Canadian Neuromuscular Disease Registry (CNDR) is a national database of Canadian adults and children with neuromuscular disease. Since June of 2011, using a clinic-based enrollment methodology, the registry has established a network of 28 specialty neuromuscular clinics in 8 provinces, and has enrolled over 2000 individuals.

Pompe disease is a rare neuromuscular disease affecting approximately 1 in 40,000 live births.[1] Clinical presentations with a wide variation in severity of symptoms occur during two main onset periods in infancy or in adulthood. Infantile onset forms are more severe and often fatal. Adult onset may occur as early as the teenage years or as late as the sixth decade of life. A successful enzyme replacement therapy treatment exists, but evidence suggests that many Pompe cases may be going undiagnosed and without access to this treatment.[1] With over 70 different causal genetic mutations, a registry would facilitate rapid identification of patients for research and could collect valuable medical data that would enhance the diagnostic knowledge of the condition. We examined the feasibility of adding detailed medical data collection on Pompe patients to the CNDR.

*Correspondence to: Lawrence Korngut, Department of Clinical Neurosciences, University of Calgary Hotchkiss Brain Institute, Calgary T2N 1N4, Canada. E-mail: korngut@gmail.com.

METHODS

We conducted a brief survey across the registry's existing clinical care Network. A web survey tool Survey Gizmo [http://www.surveygizmo.com] was used to facilitate the survey.

RESULTS

The survey was circulated to all existing CNDR Investigators and other key contacts, including genetic and metabolic clinic contacts, that were known to the CNDR in the Spring of 2014. Based on per clinic incident and followed cases, we estimate the total number of cases within our existing network to be between 20 and 120. Strong evidence exists that many cases are seen and/or diagnosed in metabolic clinics operating separately from specialty neuromuscular clinics. There was very little consensus on what dataset items should be collected in a registry dataset.

CONCLUSIONS

Willingness to collect data on Pompe disease patients in the registry network is high; however, the addition of genetic and metabolic clinics in some regions will be necessary to ensure that all Pompe disease

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patients will be captured. More investigation into the willingness of these clinics to participate in the CNDR is required. Some evidence exists that incorporation of Pompe disease into the registry might help to facilitate improved relationships between the genetic, metabolic, and specialty neuromuscular clinics across Canada, which may have important impacts beyond the Pompe disease audience alone.

REFERENCE

[1] Manganelli F, Ruggiero L. Clinical features of Pompe disease. Acta Myol Oct 2013;32(2):82-84.