DUCHENNE MUSCULAR DYSTROPHY (DMD) IN CANADA

BACKGROUND

In May of 2011 the Canadian Pediatric Neuromuscular Group published a study of 773 DMD patients in clinics across Canada who had been genetically confirmed[i]. This paper demonstrated a prevalence estimate of 1.5 per 10,000 males in Canada.

2015 population statistics from Statistics Canada indicate a population of 5,290,800 males between aged 0 – 24 years. This would suggest a population of approximately 794 Duchenne muscular dystrophy cases in Canada.

The Canadian neuromuscular disease registry (CNDR) is a national database of adults and children diagnosed with neuromuscular disease who provide informed consent for information to be abstracted from their medical records. Participants are enrolled at participating neuromuscular disease clinics across Canada and through a self-registration process mediated by their treating physician (e.g. family doctor, community neurologist). Currently, specialty neuromuscular programs in 7 provinces (BC, AB, SK, ON, QC, NB and NS) participate in the registry. The registry is in the process of acquiring all remaining neuromuscular clinics delivering specialized care in two additional provinces (NL and MB). Patients from Prince Edward Island are seen by specialty teams from the other Maritime provinces. Patients from the Yukon are seen in BC. Patients from Nunavut are seen in Ottawa, ON and patients from the Northwest Territories are seen in Edmonton, AB.

The Canadian Neuromuscular Diseases Network (CAN-NMD) is a new national collaboration amongst clinical teams, clinical and basic science researchers, and neuromuscular disease patients and families. The Network aims to enhance connectivity and collaboration at a national level to facilitate increased research capacity; enhanced clinical care and consistency of clinical care and increased educational opportunities for patients and providers. The Network involves almost 150 members across Canada to date.

CAN-NMD and the CNDR collaborated to examine the prevalence of Duchenne muscular dystrophy (DMD) across known Canadian treatment programs and to further examine the frequency of cases involving nonsense mutations.

METHODS

The National Director of Operations of the CAN-NMD individually emailed clinical care teams and members of the Network known to be caring for DMD patients to inquire about total male DMD cases, known nonsense mutation cases and any additional clinical care programs in the local region that might be seeing patients with DMD. Clinics participating in the CNDR were provided a report of their current CNDR DMD statistics and asked to verify whether these statistics represented total enrollment of their cases. Clinics not participating in the CNDR were asked to

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provide accurate statistics through information contained in clinic databases or other accurate sources (e.g. genetics clinic information). Statistics from all clinics were compiled and added to relevant CNDR data to create an aggregate national report.

RESULTS

REGION	# CLINICS REPORTING	TOTAL LIVING MALE DMD	KNOWN NONSENSE	% NONSENSE
Alberta	2/4	51	7	13%
British Columbia	2/2	73	7	10%
Manitoba	2/2	15	0	0
New Brunswick	1/2	11	0	0
Newfoundland	1/2	11	Less than 5	9%
Northwest Territories	0	0	0	0
Nova Scotia	2/2	21		
Nunavut	1/1	0	0	0
Ontario	10/13	243	27	11%
Prince Edward Island	1/1	Less than 5	0	0
Quebec	6/6	155	8	5%
Saskatchewan	1/1	Less than 5	0	0
Yukon Territory	1/1	Less than 5	0	0
TOTAL	29/37	584	50	9%

DISCUSSION

This study has some known limitations. The CAN-NMD and CNDR networks currently do not have participation from the pediatric neuromuscular program in Edmonton, nor the pediatric neuromuscular program in Saskatchewan. Assuming Southern Alberta and Northern Alberta have roughly the same proportion of cases, Edmonton might represent another approximately 40 cases and this number might include 1 or 2 cases from the Northwest Territories. The Nova Scotia pediatric clinic did not have a way to review nonsense mutations without conducting an exhaustive chart review and resources were not available for this. However, the frequency of nonsense mutations approximates everything covered in the literature and therefore it is likely that Nova Scotia has approximately 2 nonsense mutations in their known 21 patients. The final known limitation to this study is that in the provinces of Saskatchewan, Manitoba and Quebec there are a substantial number of practicing community neurologists who may see patients with Duchenne muscular dystrophy and these patients are not subsequently referred into our clinic network. It is not possible to estimate the number of cases this may represent. Further, our

Toronto clinic team reported an awareness that some adult DMD cases may be seen at a rehabilitation clinic outside our network. All known nonsense mutations reported in this study are known cases confirmed by genetic testing. With recent advances in genetic testing some older cases may not have had their testing repeated so there is still a chance that this number underestimates the true nonsense cases. However, as indicated the overall proportion is consistent with reported literature.

[i] Mah, J.K.; Selby, K.; Campbell, C.; Nadeau, A.; Tarnopolsky, M.; McCormick, A.; et al. (2011). A population-based study of dystrophin mutations in Canada. *Canadian Journal of Neurological Sciences 38(3):465-474.*

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