Early View

MOVING ALONG

International Parkinson and Movement Disorder Society

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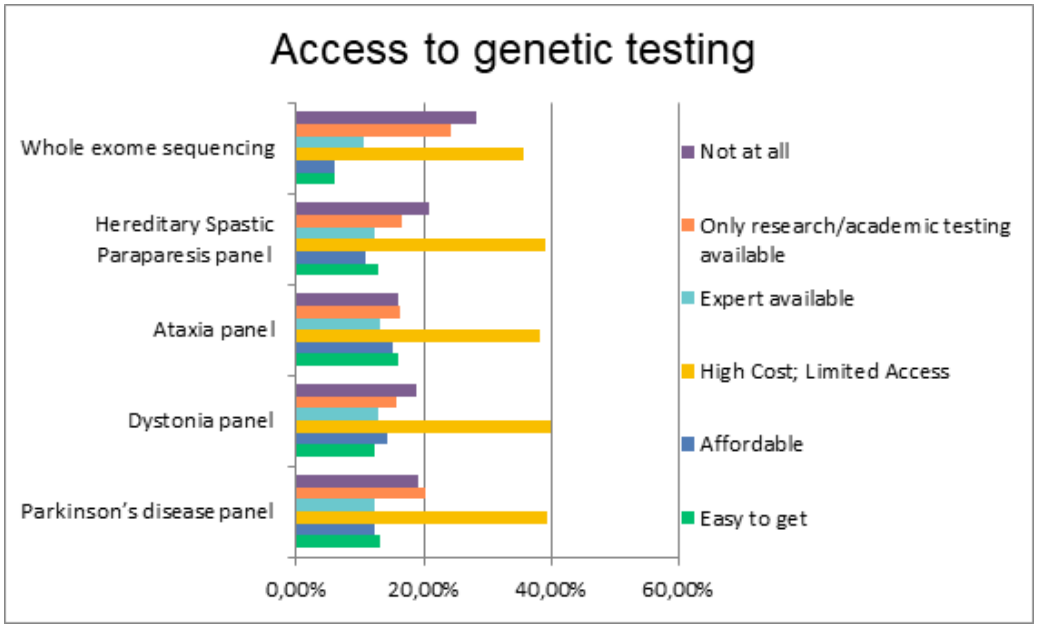
MDS Rare Movement Disorders Study Group - Global Genetic Testing Survey: Exploring the Unmet Needs

– Emilia Gatto, MD, FAAN, Chairman, Department of Neurology, Sanatorio Trinidad Mitre, Buenos Aires, Argentina; Chair, MDS Rare Movement Disorders Study Group

One of the aims of the Rare Movement Disorders Study Group (RMDSG) of the International Parkinson and Movement Disorder Society (MDS) is to improve the diagnosis of rare movement disorders. The emergence of new gene testing technologies constituted a tremendous advance in the field of rare diseases. These advances have enormously increased our knowledge on mechanisms and new promising therapeutic pathways, and at the same time expanded the genotype/ phenotype spectrum of rare diseases. Although, in the last years, next-generation sequencing (NGS) has enabled a cheap genetic diagnosis, the availability and accessibility to genetic tests remains a great challenge and account for enormous healthcare worldwide.

Members of the RMDSG designed a 21-question online survey which was electronically mailed to all members affiliated with MDS. The main objective of this survey was to better understand the access to genetic testing in different regions of the world. This survey was sent to 7,815 members of MDS who were invited to complete the survey 1,269 (93% fully completed) from 109 countries responded. 53.8% of respondents reported >10 years in clinical practice and 45.4% identified themselves as movement disorders specialists or consultants. Furthermore, 39.4% practiced in a university setting, 26.3% in a combined (private practice + university setting), 20.7% in a government setting, 9.9% in private practice, and 3.7% in another setting. Almost 90% were primarily involved in clinical care. Movement disorders specialists or pediatric neurologists were ten-fold less accessible than general physicians, whereas genetic specialists were 25-33-fold less accessible. Access to specific tests (whole exome sequencing and genetic panels for parkinsonism, dystonia, ataxia and other disorders) were limited and considered “expensive” for 35-40% of respondents. Genetic tests for Huntington disease and ataxias were the most commonly available tests at the respondents’ home institutions (available to 43%-40%). For other than these tests the average availability was 26%. University and academic centers (52.8%), provided the major genetic tests, followed by private institutions (40.8%), and by government or public institutions (36%). A national network for genetic testing was reported in only 36.7% of respondents. Despite some limitations, this survey highlights the major global challenges and disparities for access to movement disorders specialists, genetic clinics and for genetic testing availability.

These preliminary results encourage the RMDSG to increase the educational efforts and to promote the creation of networks that contribute to reduce the gap among different regions worldwide.


Access to genetic testing
Whole exome sequencing
Hereditary Spastic Paraparesis panel
Ataxia panel
Dystonia panel
Parkinson's disease panel
0,00%
20,00%
40,00%
60,00%
Not at all
Only research/academic testing available
Expert available
High Cost; Limited Access
Affordable
Easy to get