

Barriers to reducing time to diagnosis and treatment in rare disease

Wendy Everett,¹ Stacey Tatroe,² Lisa Feng,³ Paul Strumph,⁴ Josin James,³
Kelly McNeil-Posey,³ Barbara Mungin,³ Christine Rowe,³ Adrian Kielhorn*³

*Presenting author

¹Atlas Clarity, San Francisco, CA, USA; ²TwelveStone Health, Murfreesboro, TN, USA; ³Alexion, AstraZeneca Rare Disease, Boston, MA, USA; ⁴Seraxis, Germantown MD, USA

Lisa Feng, Josin James, Kelly McNeil-Posey, Barbara Mungin, Christine Rowe and Adrian Kielhorn are employees of and shareholders in Alexion, AstraZeneca Rare Disease

Wendy Everett has nothing to disclose

Stacey Tatroe receives speaker's bureau fees from Biogen

Paul Strumph is a board member at MGFA and employee of Seraxis, a cellular therapy company

Acknowledgments: Writing assistance was provided by Jon Carthy, PhD, Rebecca Spencer Martín, MSci and Rebecca Hornby, PhD, of Oxford PharmaGenesis, Oxford, UK, with funding from Alexion, AstraZeneca Rare Disease

Funding statement: This study was sponsored by Alexion, AstraZeneca Rare Disease

Background

- For patients with rare diseases, it can take years to receive a diagnosis and treatment¹
- Various barriers to timely diagnosis, and clinical, social and economic factors may prolong these time horizons^{2,3}

Objectives

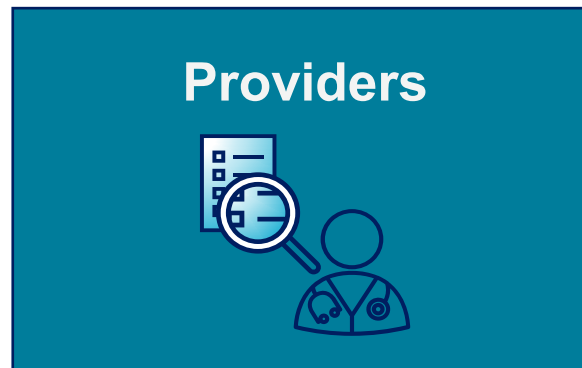
- **ASPIRE project goal:** identify, evaluate and better understand barriers to timely diagnosis and treatment among patients with rare autoimmune diseases:
 - generalized myasthenia gravis (gMG)
 - neuromyelitis optica spectrum disorder (NMOSD)

1. Black N et al. 2015. Available from: <https://piru.ac.uk/assets/files/Rare%20diseases%20Final%20report.pdf>; 2. Benito-Lozano J et al. *Int J Environ Res Public Health*. 2022; 19:6456; 3. GAO. 2021. Available from: <https://www.gao.gov/assets/gao-22-104235.pdf>.

gMG, generalized myasthenia gravis, NMOSD, neuromyelitis optica spectrum disorder.

Landscape assessment – comprised of a literature review and interviews

- Targeted literature search to assess the patient advocacy organizations, registries and online communities relating to gMG and NMOSD in the US
- Interviews conducted with representatives from:



Patient advocacy organizations, registries and online communities identified in the literature search

- Literature search conducted to identify the resources and platforms available to support patients with gMG and NMOSD

Patient advocacy organizations	Registries	Online communities
MG		
9 organizations ¹	11 patient registries ³ 1 clinical research network ³	7 communities ⁵
NMOSD		
6 organizations ²	4 patient registries ⁴ 1 clinical registry ⁴	4 communities ⁶

1. Myasthenia Gravis Foundation of America, Muscular Dystrophy Association, MG Hope Foundation, NORD, Myasthenia Gravis Association, Conquer MG, MG Minnesota, Myasthenia Gravis Foundation of Western Pennsylvania, Myasthenia Gravis Foundation of Michigan; 2. Siegel Rare Neuroimmune Association, The Sumaira Foundation, The Guthy Jackson Foundation, NORD, Child Neurology Foundation, Connor B Judge Foundation; 3. Alexion, Allstripe, ARNet, Duke Myasthenia Gravis (MG) Clinic Registry, EXPLORE MG Registry, MGFA MG Patient registry, Myasthenia Gravis Rare Disease Network, MyRealWorld MG, Patients Like Me, Picnic Health, PROMISE MG Study, Rare Disease Patient Registry; 4. CorEvitas SPHERES Registry, NMOBase, Patients Like Me, Pediatric NMOSD Observational Study, SRNA Registry; 5. Daily Strength, Myasthenia Gravis (Facebook), Myasthenia Gravis News, NeuroTalk, Smart Patients, Stuff That Works, INSPIRE; 6. Neuromyelitis Optica, Neuromyelitis Optica Net, Smart Patients, Stuff That Works.
gMG, generalized myasthenia gravis, NMOSD, neuromyelitis optica spectrum disorder.



Patient-level barriers to timely diagnosis and treatment identified by representatives of patient advocacy organizations^{a1–8}

Lack of access to specialists



Inadequate insurance coverage



Limited/no access to telemedicine



Difficulty getting time off



Attitudes about health



Language barriers and health literacy



Lack of trust in physician/healthcare system



Fear of diagnosis



^aBarriers are presented in no particular order.

1. Global Genes. 2021. Available from: <https://globalgenes.happyfox.com/kb/article/272-diversity-of-odysseys/> (Accessed Mar 23, 2023); 2. Avalere. 2021. Available from: https://avalere.com/wp-content/uploads/2021/07/Diagnostic_Journey_for_RD_Patients-June-2021.pdf (Accessed Mar 23, 2023); 3. NORD. 2020. Available from: https://rarediseases.org/wp-content/uploads/2020/11/NRD-2088-Barriers-30-Yr-Survey-Report_FNL-2.pdf (Accessed Mar 23, 2023); 4. GAO. 2021. Available from: <https://www.gao.gov/assets/gao-22-104235.pdf> (Accessed Mar 23, 2023); 5. Isono K et al. *PLoS One*. 2022;17:e0265847. DOI: 10.1371/journal.pone.0265847; 6. Bogart K et al *Orphanet J Rare Dis*. 2022;17:196. DOI: 10.1186/s13023-022-02343-4; 7. Eurodis. 2009. Available from: https://www.eurordis.org/wp-content/uploads/2009/12/EURORDISCARE_FULLBOOKr.pdf (Accessed Mar 23 2023); 8. Amezcua L et al. *JAMA Neurol*. 2021;78:1515–1524. DOI: 10.1001/jamaneurol.2021.3416.



Interview findings^a from patients and patient advocacy organizations

“If patients had access to their whole medical record this could reduce the time to diagnosis...it took over 16 centers [hospitals or clinics] for my two brothers and I to receive a diagnosis”

Patient

“The starting point is conducting research that honors the perspective of patients as much as it honors clinicians... patients are the greatest untapped resource in healthcare”

**Patient advocacy
organization**

^aKey quotes represent consistent topics identified from the interviews.

RESULTS – PROVIDER LEVEL BARRIERS

Barriers to timely diagnosis and treatment identified by providers^a

Lack of knowledge and misdiagnoses



Biases



Shortage of specialists



Not using guidelines



Lack of best use of technology



Overcrowding in ERs



Interview findings^b from providers



“COVID has changed care: providers are now used to video visits but they're not adequate for doing a complete neuro exam”

Providers

“Rural community physicians are not keeping up their knowledge base and don't know or understand rare diseases”

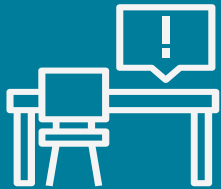
^aBarriers are presented in no particular order.

^bKey quotes represent consistent topics identified from the interviews.
ER, emergency room.



Barriers to timely diagnosis and treatment identified by policymakers and payers^a

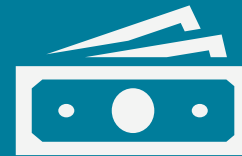
Lack of priority for rare diseases relative to other diseases



Long wait times to access clinical centers of excellence



Lack of adequate insurance



Prior authorization



^aBarriers are presented in no particular order.
ER, emergency room; gMG, generalized myasthenia gravis, NMOSD, neuromyelitis optica spectrum disorder.

- Multi-dimensional barriers that may prolong time to diagnosis and treatment for patients with gMG and NMOSD were identified by patients, patient advocacy organizations, providers, policymakers and payers
- Future studies should consider how these barriers contribute to diagnosis delays
- Understanding how these barriers affect diagnosis will support:
 - development of strategies to shorten time to an accurate diagnosis
 - timely patient treatment

Proposed solutions that are likely to reduce barriers and improve equity for gMG and NMOSD



Improve the referral process to rare disease specialists



Develop precise diagnostic criteria for primary care and acute care practitioners



Change the Medicaid payment scale for rare disease care



Improve access to care within underserved communities



Publish longitudinal utilization and quality data on equity of care



Educate beneficiaries about managing their health

**Key take
away**

What actions can we all take to promote health equity?

Thank you to everyone who
contributed to this study

Contact

Adrian.Kielhorn@alexion.com