Barriers to reducing time to diagnosis and treatment in rare disease

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DISCLOSURES

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

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Background

- For patients with rare diseases, it can take years to receive a diagnosis and treatment[1]
- 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

Providers

Policymakers and **payers**

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

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

[a]Barriers 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

[a]Key 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"

[a]Barriers are presented in no particular order.
[b]Key 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

[a]Barriers are presented in no particular order.
ER, emergency room; gMG, generalized myasthenia gravis, NMOSD, neuromyelitis optica spectrum disorder.

CONCLUSIONS

- 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

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

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?

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