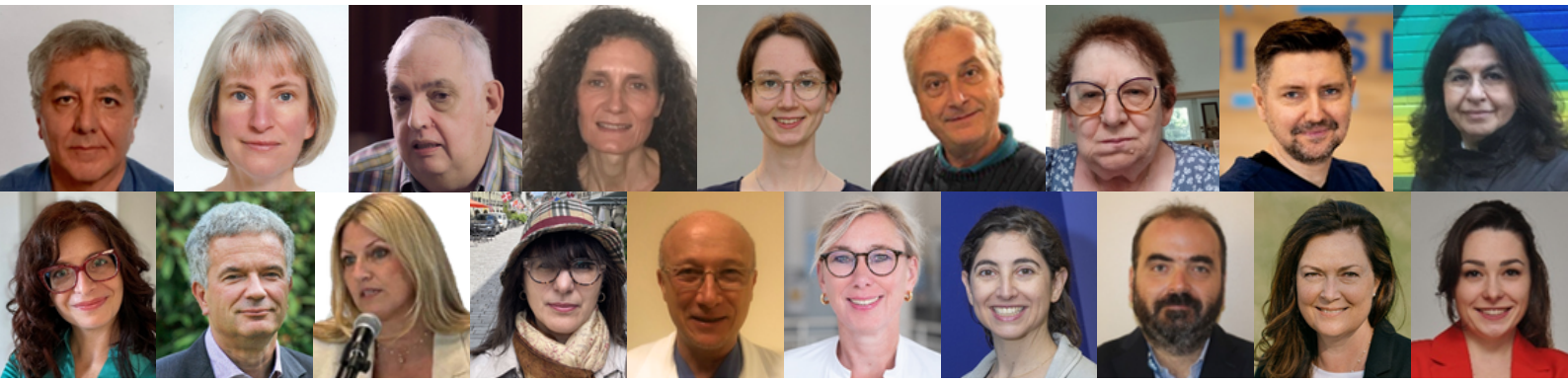
RETHINKING MYASTHENIA GRAVIS

ADVOCACY PAPER

ANNEX



Rethinking Myasthenia Gravis Care through a Brain Health Lens: Adaptive Pathways and Patient-Centred Insights

EBC European Brain Council

INSTITUTE OF MANAGEMENT Sant'Anna School of Advanced Studies – Pisa

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Interviewee Organisations and Institutions Involved

France

Centre Hospitalier Universitaire (CHU) de Limoges, Limoges, France

Groupe d'intérêt Myasthénies de l'Association Française contre les Myopathies (AFM-Téléthon), Évry, France

Association des Myasthéniques Isolés et Solidaires (AMIS), La Chapelle-en-Serval, France

Institut de Myologie, Paris, France

Germany

Charité - Universitätsmedizin Berlin, Berlin, Germany

Deutsche Myasthenie Gesellschaft e.V. (DMG), Bremen, Germany

Universitätsklinikum Essen, Essen, Germany

Italy

Associazione Miastenia OdV, Padua, Italy

Ospedale Ca' Foncello di Treviso - AULSS2 Marca Trevigiana, Treviso, Italy

Centro di Malattie Neuromuscolari e Malattie Rare Neurologiche - Asl Roma 1 San Filippo Neri, Rome, Italy

AIM | Associazione Italiana Miastenia, Milan, Italy

UO Neurologia, Azienda Ospedaliero Universitaria Pisana, Pisa, Italy

Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico di Milano, Milan, Italy

Poland

Neurologia Śląska Centrum Medyczne, Katowice, Poland

CognitivaMed Center for Psychotherapy and Psychiatry, Rybnik, Poland

Polish Association of Patients with Myasthenia Gravis `Gioconda´, Wieprz, Poland

Myasthenia Gravis Association - Face to Face, Kowale, Poland

Interviewees in France

Karima Ghorab:
Healthcare Professional, Centre Hospitalier Universitaire (CHU) de Limoges, Limoges, France

`I am a neurologist hospital practitioner, working in a university hospital that serves as a reference center for muscle pathologies. I have been a neurologist for about 20 years, and I have been working in this particular center since 2016. We have several reference centers in France for these neuromuscular pathologies. I see a lot of patients with myasthenia, so I am the one who initiates the treatment, reassesses the treatments, and follows up accordingly afterwards'.

`Collaborating closely with general practitioners and physicians is crucial. We need dedicated time slots for education, emphasising that myasthenia can be subtle and easily overlooked. Awareness meetings between neurologists and general practitioners are therefore essential. Ophthalmologists already excel in spotting eyelid droop or ocular issues and referring promptly - but diagnostic delays still occur for patients who `wander´ due to unexplained fatigue until symptoms worsen'.

`This highlights a broader need for efficiency and coordination across care pathways. Patients increasingly demand rapid access to specialists; if general practitioners cannot provide clarity, they often self-refer or seek consultations independently. Simple training campaigns, awareness initiatives, and partnerships with patient associations focused on neuromuscular and myasthenia conditions can help bridge these gaps. Yet public health challenges remain, such as securing timely electromyogram appointments - sometimes requiring referrals to other centers to obtain faster testing'.

`Comprehensive care is essential, involving general practitioners, neurologists, therapeutic education, and psychologists. These patients often have unique psychological profiles - young individuals suddenly facing physical limitations like those of older adults, struggling to understand that it's not 'just a tremor'. These individuals often arrive scared and panicked, yet lack dedicated psychological support, leaving them to cope alone with an overwhelming disease'.

`Overall, we also observe more reports of urinary and sexual disorders, which were not discussed as much in the past. Assessment scales, therapeutic education by nurses, and gradual specialist involvement can help address these issues. Adding psychological support would make a real difference for patients'.

Interviewees in France

Marguerite Friconneau:
Patient Organisation Representative, Groupe d'intérêt Myasthénies de l'Association Française contre les Myopathies (AFM-Téléthon), Évry, France

`I am now 70 years old, and I got my Myasthenia Gravis (MG) when I was 29. So, I am a patient with 41 years of lived experience, and I'm a rather special case because I'm seronegative (meaning the antibodies usually found in individuals with MG are not present in my blood). Back then, it wasn't the same as it is now: the disease was completely unknown. It's a bit better now, but I have to say a 'little bit' because many doctors still don't know much about it.

When I got this disease, I decided to try to find other people living with the same disease. Someone from the hospital's social services told me about a newly formed group in France that was very near to my house, so I decided to join. This was in 1987. At the time, I was so lost—so lost that I needed to meet other people facing the same problems. I joined the group, and after a few years, I became a member of the Board of Directors. In the beginning, we were about 150. Now, the group has grown to more than 2000 members.

My MG is much better now than it used to be. A few years ago, I was in very bad shape and had to be admitted to intensive care. I couldn't go on holiday. But now, I am going on holiday next week with my sister, and we are driving to Poland. It's a long way. She will drive´.

`To optimise care pathways, we need a real, structured pathway, with clear, concise guidelines. Some already exist, but they are at the national level and are far too long. It's very difficult to create something effective, but I think we will try to achieve this in the coming years. The document has to be short, only one page´.

`What people often have difficulty understanding is that the disease is always changing. One day you feel all right, the day afterwards, you could go to the hospital. That's your life. You can learn to cope, but it's mentally exhausting; this is a real burden of the disease... You have to adapt to cope every single day. I never received any psychological support, and I think I should have´.

Interviewees in France

I am 69 years old. I am French. I worked for an international group in civil engineering for a little over 30 years, and I have had myasthenia since 2008. I am president of a patient association called Association des Myasthéniques Isolés et Solidaires (AMIS). It is an association that was created in 2008 as a non-profit, and I have been its president since 2013.

Since 2008, I have been positive for the anti-acetylcholine receptor (AChR) antibody. The severity of my MG was progressive since I reached practically stage IV (severe MG) of the Myasthenia Gravis Foundation of America (MGFA) Clinical Classification. However, at the moment, I am rather in stage IIIB (moderate MG) ´.

Pierre Boulanger: Patient Organisation Representative, Association des Myasthéniques Isolés et Solidaires (AMIS), La Chapelle-en-Serval, France

`To improve patient care pathways, faster access to specialised doctors is essential. The French 'STAMINA' study demonstrated that around 20,000 people in France live with myasthenia among a 67 million overall population – meaning it is a rare condition. As a result, fatigue reported to general practitioners is rarely linked to myasthenia, and even when suspected, neurologist appointments can take six to nine months to receive. Allowing general practitioners to consult expertise centers directly, with no need for the patient to act as an intermediary could speed up diagnosis and reduce delays'.

`Improving how the diagnosis is communicated is equally important. Upon diagnosis, most patients have never heard of the condition and retain little information due to emotional distress. Patient associations help translate medical information into accessible language and provide reassurance and guidance. For example, ours is developing a verified information tool for patients'.

`Given the emotional toll of the disease, early access to multidisciplinary support, including a relay nurse and psychological care, would be beneficial but remains rare in France. Post-diagnosis care should also include therapeutic patient education (ETP) and administrative guidance. Patient associations assist by helping patients manage 'Affection de Longue Durée' (ALD) status, 'Maison Départementale des Personnes Handicapées' (MDPH) applications, insurance, occupational follow-up, and so on'.

`Overall, our mission is to support patients intelligently and empathetically, helping them to strike a balance between non-negotiable medical constraints and personal aspirations´.

Interviewees in France

Simone Birnbaum:
Clinical Researcher and Healthcare Professional, Institut de Myologie, Paris, France

`I am a physiotherapist and researcher. I work at the Institute of Myology as a clinical research physiotherapist. A large part of my research is focused on myasthenia gravis symptoms, exercise, fatiguability, respiratory impairment, and quality of life´.

`In France, we have the 'Protocoles Nationaux de Diagnostic et de Soins' (PNDS), which serves as a reference guide for healthcare professionals. It is currently being updated, and the revised version is expected to be published soon. It is a very useful resource, though we know that healthcare professionals don't always have time to read about every condition in detail'.

`In my view, it is not essential for every clinician to be an expert on the disease. What matters most is empowering patients to understand and manage their condition. With myasthenia and its fluctuating symptoms, patients really need to know themselves well. That's why, in the long term, I believe patient empowerment is the key'.

`Also, I believe the psychological aspects are extremely important - especially when it comes to understanding and coping with a new diagnosis, or managing a chronic condition through different stages of life. Situations like pregnancy, for instance, raise specific concerns not only about therapies but also about overall disease management. These life transitions can be challenging, and knowing where to turn for support is vital. We're fortunate to have patient associations, which many patients can turn to for guidance and help´.

Interviewees in Germany

`I'm a senior neurologist based in Berlin, Germany, with over 30 years of experience in clinical neurology and research. My motivation as a neurologist is to better understand neurological diseases and ultimately improve patient care. I began working more closely with myasthenia gravis over 20 years ago, when I first took responsibility for MG patients in an outpatient setting´.

Andreas Meisel:
Healthcare Professional, Charité-Universitätsmedizin Berlin, Berlin, Germany

`As an Integrated Myasthenia Gravis Centre certified by the German Myasthenia Gravis Society, we provide multidisciplinary care in close cooperation with thoracic surgeons, performing over 100 thymectomies per year, as well as specialists in women's health and obstetrics, especially regarding pregnancy and family planning, haematologists, and neuropathologists. Neuropathology enables advanced analysis of intercostal muscle samples to identify neuromuscular junction pathologies, crucial for research and for specific patient groups, such as seronegative cases'.

`We care for over 1,000 MG patients each year - more than 250 outpatients, 250 inpatients and around 50 in intensive care. This high case volume underpins our active involvement in clinical research, including trials for new treatments, investigator-initiated studies, biomarker research and burden-of-disease assessments'.

`Maintaining contact with the nationwide network of integrated myasthenia centres is essential, for instance, when seeking second opinions. We also collaborate within a regional network of neurologists, which helps ensure that patient needs are met through coordination beyond the hospital setting. In addition, cooperation with external psychologists is important, as we often recommend psychotherapy for patients who are open to it. This support plays a key role in preventing and managing psychiatric comorbidities such as anxiety or post-traumatic stress disorders´.

`I believe in guiding patients and involving them as active partners in their care. They need to understand the reasons behind each medical decision and be encouraged to self-educate and learn through patient guidelines, since general practitioners, or even non-specialist neurologists, often lack MG expertise. MG management is complex. Specialists are essential, and patients themselves should also become informed advocates and, in a sense, experts in their own condition´.

Interviewees in Germany

Anja Hoffmeister:

Patient Organisation Representative, Deutsche Myasthenie Gesellschaft e.V. (DMG), Bremen, Germany

`I have had myasthenia for 11 years now. It began when I was 13, and I'm 24 now. I'm currently completing my master's in medical physics and will soon begin my PhD in physics. I've also been part of the Deutsche Myasthenie patient association for 11 years, with active involvement over the past five and a half years. I first joined by organising meetings for a regional group, and after a year or so, I started organising meetings for young people with myasthenia – which I continue to do today. I am now also involved in the executive board, in international activities, and on the user council board for the German Myasthenia registry.

In the beginning, my myasthenia was severe. I spent several years in a wheelchair and later relied on a walking frame. I also experienced breathing difficulties and required frequent hospital visits. Now, my condition has improved significantly - I no longer need the walking frame and can even participate in sports. I still experience persistent diplopia (double vision) and occasional difficulties with walking, speaking, swallowing, and drooping eyelids, but overall, things are much, much better.

Physical exercise has helped me a lot. I practice archery and occasionally swim. I also enjoy walking – slowly, just at my own pace, not running, just walking´.

`One advantage of being diagnosed with myasthenia at a young age is that I was able to choose a career that I can continue despite my condition. Many other young people I've met studied or worked for several years before developing symptoms and being diagnosed, which often leads to major challenges in their professional lives´.

´My treatment is covered by health insurance, which is essential because it's extremely expensive - without insurance, I wouldn't be able to get this treatment´.

`There is very little information on how contraception works with myasthenia. Neurologists are often unsure which methods work best, and gynaecologists typically don't know about myasthenia, so it's a bit difficult. I'm currently working to improve access to reliable information on topics such as sexuality, contraception, and family planning for people with myasthenia'.

Interviewees in Germany

Claudia Schlemminger:
Patient Organisation Representative, Deutsche Myasthenie Gesellschaft e.V. (DMG), Bremen, Germany

`I'm 54 years old, and I come from Germany. I have been living in North Rhine-Westphalia since 2018, after previously resided in the Berlin-Brandenburg region. After finishing school, I earned a teaching degree, but because I had studied in the former German Democratic Republic (GDR), my qualifications were not officially recognised, so I had to start over. I then trained as a dental assistant, which I truly enjoyed.

Everything changed when I developed myasthenia. I went from being a workaholic and sports enthusiast to someone who could barely move. Walking became impossible, and I eventually had to use a wheelchair or a walker. I had to retire because of my condition - a huge shock for me, as I was diagnosed at 39 and had always considered myself completely healthy before then´.

`You could say that, for nearly 20 years, not much progress happened in treatment. The standard therapy was immunosuppression with azathioprine, and if that wasn't tolerated, there were only two or three alternative options. Now, however, a wide range of new medications is available - so many that it can be difficult to keep up - and even more are on the way. The great thing is that these newer drugs are far more targeted. While older treatments like cortisone or azathioprine suppressed the immune system with a 'sledgehamme' effect, the new therapies act more precisely, fine-tuning specific mechanisms. This makes a big difference for patients, both in terms of effectiveness and acceptance'.

'That said, these new medications are extremely expensive, which creates challenges. Many physicians worry about reimbursement claims from health insurance companies if costs are questioned. In addition, the growing number of infusion-based therapies brings logistical issues such as scheduling infusion slots, ensuring nursing supervision, and managing time effectively. There are also new subcutaneous treatments that require patients to be properly monitored and informed about their use´.

Interviewees in Germany

Heike Hoffmeister:
Caregiver, Mother of Patient Organisation Representative Deutsche Myasthenie Gesellschaft e.V. (DMG), Bremen, Germany

`I'm 52 years old and trained as a bank clerk. I spent several years at home caring for my children and later, for my parents. After they passed away, I returned to work – not in banking this time, but at a rehabilitation clinic.

My daughter has had myasthenia since 2014. It began one day when she called me from school, saying she couldn't see anymore. I picked her up and took her to the paediatrician, who initially suspected multiple sclerosis. She was later found to be seronegative, meaning she doesn't have detectable antibodies, and the standard tests were only partially conclusive.

At one point, doctors suggested she see a psychologist. The psychologist agreed that psychological support could help her cope, but after assessing her, he confirmed there was nothing psychologically wrong - she wasn't imagining her symptoms. Unfortunately, some doctors still didn't take her seriously and continued to doubt her condition.

It was particularly hard for her as a teenager, being in a wheelchair or using a walker. Using a walker felt even worse, as it's typically associated with elderly people, not young girls´.

`The doctors and others always offered support, but in the end, I handled most of it myself. I was the one driving my daughter everywhere, accompanying her to appointments, and staying with her when needed. For instance, during her thymectomy in Berlin, she was in the hospital for about two weeks. I wasn't there the entire time - I went home for a couple of days in between - but then returned to be with her again'.

`At that time, I was a housewife and not working, which allowed me to be there for her as much as possible. Still, the experience left its mark. We offered a lot of support - especially me - but the negative encounters with some doctors at first, when she was not taken seriously, were very difficult for her. Those experiences are something she won't forget´.

Interviewees in Germany

`I'm a paediatric neurologist and have been working in Essen for about 19 years. I have been working in the field of myasthenia for roughly 25 years, starting when I began treating patients in Bochum before moving on to other places. Today, I head the Paediatric Neurology Department, where we care for a large number of neuromuscular patients up to the age of 18, as our focus is on child neurology´.

Ulrike Schara-Schmidt:
Healthcare Professional,
Universitätsklinikum Essen, Essen, Germany

`The youngest patient we have had diagnosed with myasthenia gravis was 15 months old. We also see many infants with ocular or secondary myasthenia'.

`One of the challenges we face is the limited range of medications available for children compared to adults. In adult care, treatment options have improved considerably, and when the first therapy works well, everything usually goes smoothly. However, in paediatric cases, some patients do not respond to initial treatment, and we must try alternative medications to find the right one'.

`Looking ahead, I believe we need more time and resources for patient care, as well as expanded telemedicine options to maintain contact between visits. There are already significant advances in treatment - checkpoint inhibitors, complement inhibitors, neonatal receptor modulators, improved surgical methods, and more precise antibody detection. Unfortunately, most of these developments are not yet available in the scope of paediatric care, though studies involving adolescents are underway. My hope is that in a few years, children and adolescents will have access to a broader range of therapies similar to those available for adults, reflecting the real progress we've seen in the field´.

Interviewees in Italy

`I'm from Italy, and I'm the vice president of Associazione Miastenia, a patient organisation based in Padua. I work as a chemist and technician at the University's Department of Pharmacology and Pharmaceutical Sciences, where I'm involved in both research and education. I like playing the guitar, and I love photography.

I was diagnosed with myasthenia in 1993, so I've lived with it for most of my life. Today, I'm doing well and enjoying every moment - just last night, I was playing with my band in a pub. Early on, things were more challenging since I have generalised myasthenia affecting almost all of my muscles, but I received a quick diagnosis because I tested positive for acetylcholine receptor antibodies, the only known type at the time´.

Andrea Pagetta:
Patient Organisation Representative, Associazione Miastenia OdV, Padua, Italy

`When I was diagnosed, I was relieved that doctors had found something treatable. No one told me it was a chronic condition or explained the fluctuations. I thought it was like having a cold or the flu - you take your medicine, wait a bit, and then recover'.

'(...) I began to accept that myasthenia wasn't something that would go away, but rather something I would have to live with – as a part of my life. Through a support group with other patients, psychologists, and neurologists, I learned to speak openly about my symptoms and feelings, which helped me accept the disease and move forward´.

`Reducing diagnosis time should be a top priority. I was lucky - it only took a few months - but many others wait years, often being told their symptoms are psychological, which is wrong and deeply frustrating'.

`I also recall the story of a friend, a young woman studying architecture in Venice, whose symptoms were dismissed as psychological. When I was a young man and said I couldn't jump while playing volleyball, everyone believed me right away; no one suggested it was `in my head'. These gender stereotypes, where women's symptoms are more easily dismissed as psychological, are an important issue to address if we want to improve diagnostic pathways and patient care´.

Interviewees in Italy

Domenico Marco Bonifati:

Healthcare Professional, Ospedale
Ca' Foncello di Treviso - AULSS2
Marca Trevigiana, Treviso, Italy

`I am the head of the Neurology Unit at Ca' Foncello Hospital in Treviso, and I am a neurologist by training. My work has focused mainly on myasthenia gravis since my residency, during which I managed an outpatient clinic for several years. Before moving to Treviso in 2015, I worked at a hospital in Trento´.

`The most challenging symptom to interpret is fatigue. Many patients report feeling tired, but fatigue can result from numerous causes, including psychological ones. It's essential to distinguish between general fatigue and true fatigability, which is specific to myasthenia. Other symptoms like double vision, drooping eyelids, or a nasal voice are easier to recognise. During a consultation, for example, asking the patient to count to 50 can sometimes reveal voice changes´.

`One of the main challenges is always time. None of us works exclusively in myasthenia care - even as head of the clinic, I have many other responsibilities, so dedicating enough time to patients or research is difficult. That's an area where improvement is clearly needed'.

'Digital technologies could help a great deal, but our public health system still lags behind when compared to how private companies manage client support using advanced, real-time digital tools. Greater investment in innovative digital solutions would make a significant difference, helping us use our limited time more effectively´.

Interviewees in Italy

`I work in the Neurology Department of San Filippo Neri Hospital in Rome, part of ASL Roma 1, and have been treating rare neurological diseases for about thirty years. I trained in Rome and Verona, beginning in neuropathology before specialising in nerve and muscle disorders.

Our centre serves as the regional reference centre for rare neurological diseases in Lazio and, in recent years, has focused particularly on developing and applying new therapies for neuromuscular junction disorders´.

Elena Maria Pennisi:
Healthcare Professional, Centro di Malattie Neuromuscolari e Malattie Rare Neurologiche - Asl Roma 1 San Filippo Neri, Rome, Italy

`Until recently, available treatments often weren't sufficient to manage the disease burden, which affects not only motor function but also emotional well-being. Today, new therapies offer important opportunities, especially for patients who do not respond to conventional treatments, providing good efficacy with fewer side effects. These innovations are particularly valuable in managing acute phases of the disease´.

`Additional staff would make a major difference - more professionals, especially younger ones, could help expand care and ensure future expertise. Passing on knowledge to the next generation of neurologists is essential'.

`Our system is quite efficient; there are no long waiting times. Electromyography and antibody tests can usually be done within one or two weeks. While more time per patient would be beneficial, the overall organisation functions well thanks to its multidisciplinary and integrated approach'.

`Overall, Italy is well-positioned for myasthenia care. We have several specialised centres nationwide, which is not a sign of fragmentation, but of strength; it ensures accessibility across the country and reduces the need for long-distance travel, stress, and costs´.
`The most important goal is early diagnosis. The sooner it happens, the lighter the disease burden. Early detection allows for better therapy control and helps prevent serious complications, which, fortunately, are now quite rare. I think we're clearly on the right path´.

Interviewees in Italy

Giuseppe Bignone:
Caregiver, Husband of Patient
Oragnisaton Representative,
AIM | Associazione Italiana
Miastenia, Milan, Italy

`I am a caregiver for my wife, who has both myasthenia gravis and Stiff-Person Syndrome (SPS) - two serious conditions. My role is simple: I'm her husband, partner, and companion in everything. My wife's myasthenia fluctuates, but it's rather serious. For example, she can't go out alone and relies on a wheelchair because she gets tired immediately, making independent living impossible. At home, we've adapted our environment - every room has an armchair or recliner so that when the first signs of fatigue appear, she can immediately lie down´.

`The hardest part is helping my wife cope with the disease and the sense of limitation that comes with it. It's difficult to come to terms with a body that no longer functions as it once did. Losing her job as a teacher created a deep void. Part of my role is to support her emotionally, finding ways to help her rediscover her strengths and focus on what she can still do rather than on what she has lost'.

`On a practical level, I've adapted as well – I can push my wife's wheelchair, even uphill, without difficulty, and those daily tasks don't tire me. The real challenge is balancing caregiving with work. My life has changed, too, especially in my professional role as a teacher at a public science high school. My principal has often reminded me that my 'Law 104/1992' leave (paid, covered leave for employees to care for family members with certified severe disabilities) should be scheduled in advance. I always explain that while I can plan for therapies, I can't predict sudden health emergencies. When these emergencies happen, I simply notify the school immediately by certified email. Despite performing my duties excellently, I was removed from my positions as department director and coordinator because I benefit from around twenty days of leave per year. This has also affected my income and pension´.

`I hope to see a truly holistic, interdisciplinary care system in the future - one that brings together all key professionals, from neurologists to pulmonologists, physiotherapists and psychologists, working collaboratively and treating the patient as an equal partner. Ideally, the neurologist would coordinate this teamwork, while caregivers would be recognised as essential members of the care team - something that, unfortunately, is still lacking in our area´.

Interviewees in Italy

`I'm Maria Bonaria, but everyone knows me as Maya. I'm from Italy, born and raised here, and I'm a pharmacist living with myasthenia gravis. For me, it's a roller coaster. My myasthenia initially presented as mild, but my diagnostic journey was very long: it took six years before I finally received a diagnosis. During that time, without treatment, my symptoms fluctuated but remained relatively manageable. Later, the condition became more severe, and although I take medication, my response still varies throughout the year. It's a highly unpredictable disease; today, for instance, I feel fine, but I never know how long that will last´.

Maria (Maya) Bonaria Uccheddu: Patient Organisation Representative, AIM | Associazione Italiana Miastenia, Milan, Italy

`A fast response is essential at the first signs of a myasthenia exacerbation - neurologists must act quickly, as the condition can worsen rapidly. Patients need accessible, timely communication, and telemedicine could help greatly, especially for those living far from specialised centres´.

`Because of my health, I've often had to take time off or work at reduced capacity. In another job, I might have been dismissed - but being self-employed also meant my business couldn't grow the way I had hoped'.

`Even before my diagnosis, the symptoms had already forced major changes in my life. Driving became difficult, so I bought a house closer to work and stopped using a car altogether. On an island like Sardinia, where public transport is unreliable, not owning a car made daily life and work far more challenging'.

`Living with an unpredictable, undiagnosed condition was extremely hard – both socially and emotionally. I'd swing between periods of being active and sociable when I felt well and times of isolation during flare-ups. Explaining my situation was nearly impossible, especially since my symptoms were invisible and I didn't yet have any answers myself´.

`What I find most frustrating is that I've run out of treatment options for my type of myasthenia gravis. Because I'm seronegative, I don't have access to the same medications available to patients with known biomarkers. Those who test positive have more options, while I'm currently on an off-label treatment that isn't officially approved for myasthenia gravis. It's not that these therapies don't exist - they do - but I simply don't have access to them. This isn't a medical problem, but one of equity and access, as seronegative patients are often excluded for complex regulatory reason´.

Interviewees in Italy

`I'm a neurologist based in Pisa, where I work in a major, specialised centre for myasthenia gravis that treats patients from across Italy. I've been diagnosing and managing myasthenia gravis for nearly 20 years and have seen many cases - our centre follows an estimated 3,000 to 4,000 patients. We're a team of neurologists, and I currently lead the myasthenia gravis research and treatment group at Pisa´.

Michelangelo Maestri Tassoni
Healthcare Professional, UO Neurologia, Azienda Ospedaliero Universitaria Pisana, Pisa, Italy

`Digital tools could play an important role in monitoring myasthenia gravis by helping detect early signs of deterioration through regular 'Myasthenia Gravis Activities of Daily Living' (MG-ADL) assessments and helping physicians prioritise patients in need. However, each case is unique: some patients who seem stable may still struggle, while others with persistent symptoms may feel better than before. These tools have limitations, particularly for older patients less comfortable with technology, and should complement rather than replace in-person visits. Still, they can greatly enhance communication and continuity of care, especially for patients distant from specialised centres´.

`This question of distance and access ties into a broader challenge: improving awareness of myasthenia gravis among general practitioners and non-specialised neurologist. Delayed recognition can postpone diagnosis and treatment, with significant consequences for patients. Not all hospitals in Italy offer the same level of expertise or treatment options, making timely referral to specialised centres crucial. While mild cases can often be managed locally, complex cases require specialised follow-up, which remains difficult to access'.

`Hospital capacity adds another layer of difficulty. At our centre, we can only admit patients experiencing, or at risk of, a myasthenic crisis, which makes it challenging to hospitalise those who would nevertheless benefit from closer monitoring'.

`Access to newer therapies presents additional barriers. Their use in Italy remains limited due to legal, bureaucratic, and cost-related barriers. These treatments are currently reserved for patients unresponsive to steroids or conventional immunosuppressants, although there is ongoing scientific debate about whether they should be introduced earlier in the disease course. Despite their proven efficacy, high costs continue to restrict their use'.

`To address some of these challenges, we established a network with other centres across Italy to support patients who live far from Pisa. Newer subcutaneous formulations represent a promising development, reducing hospital visits and easing some logistical and financial burdens for patients´.

Interviewees in Italy

Monica Sciacco:
Healthcare Professional, Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico di Milano, Milan, Italy

`I am a neurologist with extensive experience in neuromuscular disorders, working at a university hospital in Italy. I have been following patients with these conditions, including myasthenia, for several years. My work involves both initial clinical assessment and long-term follow-up in outpatient and inpatient settings.

Patients are first seen in our outpatient clinic for an initial evaluation, after which we conduct targeted diagnostic tests - most performed on-site, except for serum antibody quantification, which is outsourced to another hospital. Once the diagnosis is confirmed, follow-up plans are tailored to each patient's age, comorbidities, and treatment needs. When required, surgical and inpatient care are also available within our hospital, ensuring comprehensive and coordinated management´.

`We're now beginning to use new treatments such as complement inhibitors and receptor blockers. These therapies are promising, but limited to patients with specific antibody profiles. Many of our patients are seronegative, so they're not eligible, and must continue with traditional immunosuppressive therapies, which carry side effects and cannot be used indefinitely´.

`Regarding diagnosis, one improvement would be to perform antibody testing directly in our hospital instead of sending samples elsewhere - this is the only step we currently outsource'.

`Another possible improvement concerns treatment administration. Some patients would prefer to self-administer subcutaneous therapy at home, but hospital policy doesn't allow it. Personally, I'm not entirely convinced it's the best option, since medical supervision ensures safety'.

`For those who find it difficult to come regularly, remote evaluations could be introduced. Once patients are well known to the team, virtual follow-ups could be feasible for stable cases, while in-person visits would remain essential for assessing any clinical worsening. These adjustments could make care more flexible without compromising quality´.

`Patient involvement in treatment decisions varies greatly. Younger patients are often more proactive - they read about the disease online and want detailed explanations of proposed treatments to ensure these align with their work, travel, or personal lifestyle. Older patients, by contrast, tend to be less focused on the details and more concerned with stability and quality of life, especially when dealing with multiple health conditions. In general, patients trust my decisions, but they value open communication. They appreciate when I take their individual circumstances into account rather than applying treatment guidelines mechanically´.

Interviewees in Poland

`I'm a specialist neurologist from Katowice, Poland, and I work at Neurologia Śląska Medical Centre, which is a private site that has been conducting and performing clinical trials for 25 years´.

Marek Śmiłowski
Healthcare Professional, Neurologia Śląska Centrum Medyczne, Katowice, Poland

`I think one of the main challenges for patients is finding a doctor who can provide a timely and accurate diagnosis. Many struggle to access specialists in neuromuscular diseases, particularly for myasthenia gravis, and often spend months consulting different doctors before receiving the correct diagnosis. Even once diagnosed, identifying appropriate treatment remains difficult, as access to advanced biological therapies is still limited, making effective disease management more challenging´.

`At our centre, we are well equipped to diagnose and treat patients with myasthenia gravis. As a private facility specialised in this condition, our processes allow us to reach a diagnosis more quickly. In contrast, patients who turn to general neurology clinics usually face longer waiting times before receiving proper evaluation and treatment'.

`While diagnostic capabilities across Poland - including in public outpatient clinics - are generally comparable to those in other European countries, access to biological treatments varies significantly from country to country. In Poland, only a few therapies are reimbursed and available through the public system. In the private sector, reimbursed treatments are unavailable since private clinics are excluded from the National Health Service system. To bridge this gap, we provide patients with the opportunity to participate in clinical trials, which often can grant access to new and promising therapies'.

Interviewees in Poland

Anna Kwiaton-Szopa:
Healthcare Professional, CognitivaMed Center for Psychotherapy and Psychiatry, Rybnik, Poland; Polish Association of Patients with Myasthenia Gravis `Gioconda´, Wieprz, Poland

`I hold a master's degree in psychology with a specialisation in neuropsychology and cognitive neuroscience from Jagiellonian University in Kraków. Later, I completed postgraduate studies and several courses in psychodietetics. My professional experience includes working in a psychiatric ward and as a school psychologist, where I supported children, adolescents, adults, and older adults.

I currently work as a psychologist at the Municipal Social Welfare Home in Rybnik, providing support to seniors, and at the CognitivaMed Center for Psychotherapy and Psychiatry, where I offer individual psychological and psychodietetic consultations. I also work with the Polish Association of Patients with Myasthenia Gravis "Gioconda", where I voluntarily provide individual psychological support to help people cope with the challenges of living with myasthenia´.

`When it comes to myasthenia gravis, patients often struggle with anxiety and depression. They must come to terms with their diagnosis and accept that they may never fully regain their previous level of physical ability. While vulnerability to anxiety can stem from personality traits such as neuroticism, the specificity of the myasthenia itself also contributes - breathing difficulties, for example, can heighten anxiety. Some patients also suffer from Post-Traumatic Stress Disorder (PTSD), often linked to multiple hospitalisations, including stays in intensive care units and the use of respirators, and so on. In my opinion, the fact that myasthenia is a rare disease further intensifies patients' difficulties. Many patients report that even experts, including neurologists and mental health professionals, have a limited understanding of their condition and what they are going through´.

`Psychological support is invaluable. Too often, we overlook that 'health' includes mental health as well. In my country, Poland, access to professional psychological care is limited, and patients frequently face long waiting times for therapy'.

'Many times, I have heard from patients with myasthenia that they would be extremely happy to have an integrated care model for rare diseases. In - this single centre, they could access comprehensive support, including diagnosis, medical treatment, and psychological or psychiatric care, provided by professionals who truly understand the realities of living with myasthenia gravis´.

Interviewees in Poland

`I am a patient living with multimorbidity, including myasthenia gravis (MG). My lived experience of navigating complex healthcare systems informs my engagement in patient advocacy, medicines research and development, and health policy discussions at both national and European levels'.

`I am the founder and president of the Myasthenia Gravis Face to Face Association in Poland. I also serve as a Municipal Representative for Persons with Disabilities, working mainly remotely due to my health condition. Despite living with a chronic illness and being on a disability pension, I remain professionally and socially active, combining institutional work with grassroots patient advocacy´.

Sylwia Łukomska
Patient Organisation Representative , Myasthenia Gravis Association - Face to Face, Kowale, Poland

`Almost twenty-nine years ago, I was unable to wake up properly after a caesarean section. The procedure was planned - a full-term twin pregnancy, large babies. Doctors did not know why a 20-year-old woman was having problems regaining consciousness and breathing after surgery. I was connected to oxygen'.

`After surgery, patients are expected to get up and start walking as soon as possible. Nurses noticed that I had difficulty walking. I was dragging my right leg, and my gait was abnormal. I constantly felt that my muscles were strangely weak. I was afraid to hold my daughters in my arms - I felt I might not be able to support them'.

`No one took my experience seriously. I knew what the doctors and nurses were thinking: a young mother after a difficult birth, overwhelmed by caring for new-borns. I was told that I should pull myself together, stop focusing on myself, that it was nothing serious - just psychosomatic symptoms. That was the message I left the hospital with...´.

`After diagnosis, frustration, fear, and grief grew. I was afraid I would always have to explain myself - that when I walked unsteadily, I was not drunk; that when I couldn't do something, it wasn't laziness but extreme fatigue. Getting out of bed often ended in a fall. Healthy people experience muscle weakness after a marathon - I sometimes experience it after walking between rooms'.

`Because of the invisibility and fluctuation of the disease, myasthenia gravis places a heavy psychological burden on patients. Symptoms are often misunderstood by society and even by close relatives´.

`My illness involved a significant financial burden - private diagnostic tests, medications, rehabilitation, physiotherapy, psychological support, and frequent long-term absences from work, sometimes lasting months, which significantly reduced my socio-economic quality of life´.

Interviews were conducted by Laura Guiso, Tao Isabel Buck, and Dieyenaba Faye on behalf of the European Brain Council (EBC).

Thank you to all patients, caregivers and healthcare providers for their valuable time and contribution to the Advocacy Paper, as part of the second phase of the Rethinking Myasthenia Gravis project. Rethinking Myasthenia Gravis (MG) is a 2-year multistakeholder research-driven project offering policy recommendations to make tangible changes with the aim to improve the lives of people living with Myasthenia Gravis across Europe. The project officially kicked off during EBC's Rare Disease Day 2025 event, `Towards a Rare Brain Disease Ecosystem`.

Read the Advocacy Paper

Learn more about the project

RETHINKING MYASTHENIA GRAVIS

ADVOCACY PAPER

ANNEX

Rethinking Myasthenia Gravis (MG) is a 2-year multistakeholder research-driven project offering policy recommendations to make tangible changes with the aim to improve the lives of people living with Myasthenia Gravis across Europe. The project officially kicked off during EBC's Rare Disease Day 2025 event, `Towards a Rare Brain Disease Ecosystem`.

Phase II: Rethinking Myasthenia Gravis Care through a Brain Health Lens: Adaptive Pathways and Patient-Centred Insights

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