

The newly elected president of the European Alliance of Neuromuscular Disorders Associations, Boris Sustarsic, reflects on the current state of research into new therapies for neuromuscular disorders

# Overcoming obstacles

**T**he European Alliance of Neuromuscular Disorders Associations (EAMDA) works to support and improve the quality of everyday life of people with neuromuscular disorders (NMDs) – a series of rare conditions such as myasthenia gravis, Duchenne muscular dystrophy and Pompe disease that lead to muscle wastage, cramping and pain, and disability.

Founded in 1974, EAMDA brings together NDA societies from across Europe to champion the rights and interests of everyone living with a neuromuscular disorder, raise awareness and promote related research, and prevent and alleviate the consequences of associated disability.

Speaking to *Health Europa Quarterly*, the alliance's new president, Boris Sustarsic, discusses the turning point in efforts towards new treatments for neuromuscular disorders, explains why it's important to boost political support and public funding for services for people with NMDs, and highlights the role of EAMDA in bringing together patients, European NMD associations and industry.

## How would you assess the current state of research into new and effective therapies for neuromuscular disorders?

For many decades persons with neuromuscular disorders listened with great hope to the news about the results of basic research into human neurology, especially regarding individual neuromuscular disorders. Announcements of the light at the end of the tunnel sounded optimistic, but there was no information about new drugs that would tackle the progressive nature of these hereditary disorders.



**Boris Sustarsic**

An important turning point was the introduction of the drug for Pompe disease. It should be noted that it was the patients' organisations that significantly contributed to shortening the period from laboratory discovery to clinical application of the new medicinal product. Introduction of the new drug ataluren (Translarna™) for persons with Duchenne muscular dystrophy was a similar story: it was upon the appeal of patients' organisations that the medical authorities approved its provisional use. The new drug SPINRAZA® for treating spinal muscular atrophy patients raises even more realistic hopes. It represents new forms of etiological treatment, while on the other hand it signals that there will be no single treatment for all patients with NMDs but rather different drugs targeting individual genetic disorders.

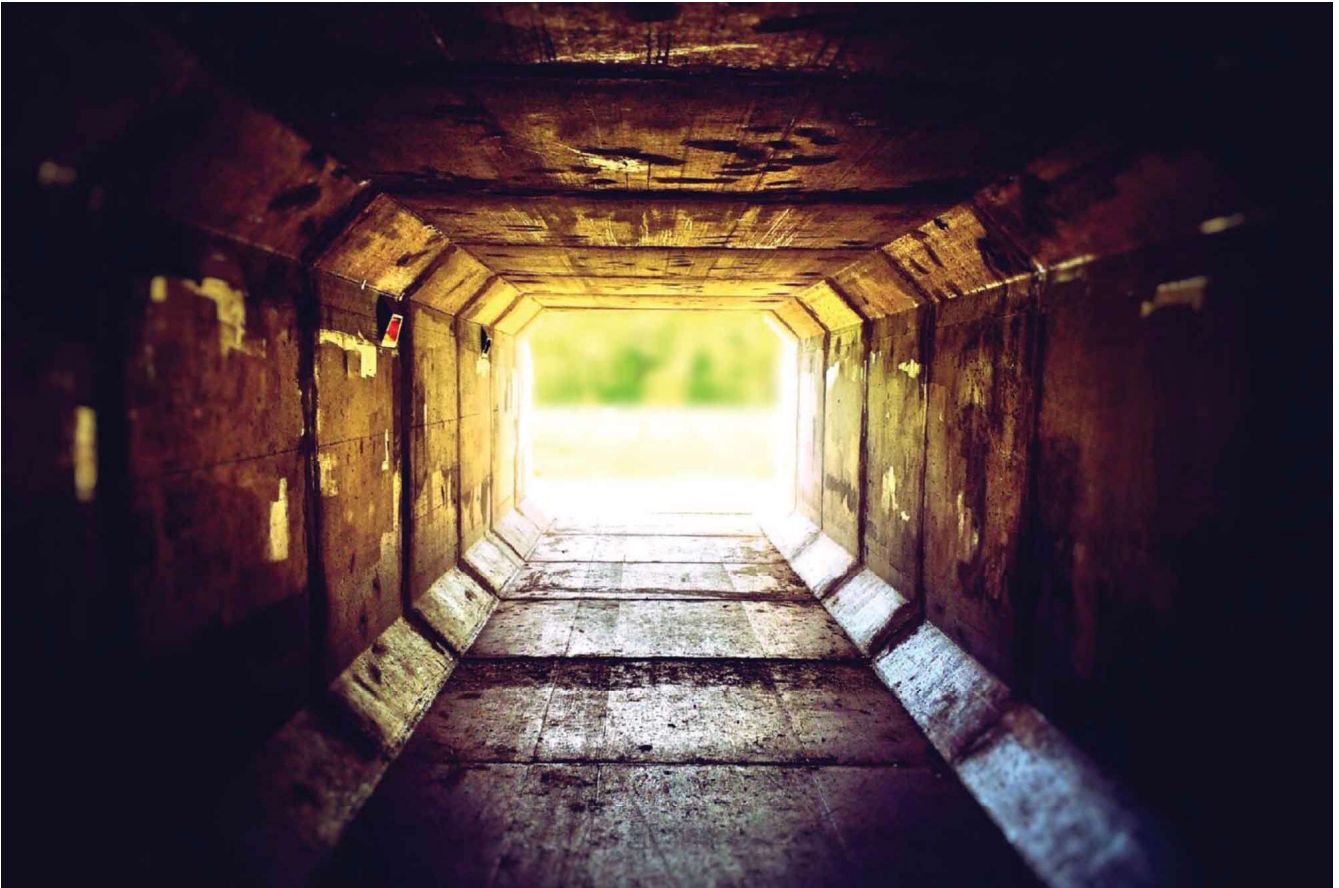
**The introduction of the drug for Pompe disease demonstrated that patients' organisations have an important role to play in translating laboratory discoveries into clinical applications**

However, a serious obstacle related to the existing and expected new drugs is their high price which leads to selective application. I hope that none of the new drugs will cause any serious secondary complications or adverse side effects. Another open question is what criteria to apply for evaluating the effects of new drugs. Namely the physicians' expectations are much higher than the expectations of persons with NMDs, who will be very happy even if the treatment only stops the progression of the disease or helps strengthen individual body functions that significantly influence the quality of our lives.

## What more do you think policymakers could do to support people with neuromuscular disorders to participate fully in all walks of society? What challenges do people with neuromuscular disorders encounter in this regard?

Apart from the therapeutic effect of new drugs, the current generation of persons with NMDs show a significantly longer life expectancy, which is mainly the result of internal medicine





measures, good nutrition and appropriate medical care, as well as provision of electronic technical aids which compensate for the weakened or lost muscular power. Policymakers at the national and EU level should be made aware that it is possible to introduce friendly and efficient civilisational solutions in which muscular power no longer plays the decisive role. With the technological support that is already available on the market persons with NMDs can be equal citizens in many aspects and play active social roles in private and public life. Thus, we should use the available arguments and examples of good practice to overcome the concealed negative attitudes towards persons with NMDs, including those in the medical field which can be described as therapeutic nihilism.

As a rule, persons with NMDs have an intact central nervous system, and it has been scientifically proven that in some cases they have an increased IQ level. Therefore, in my opinion, the basic challenge of persons with NMDs is to attain good quality education, if possible to the highest university degree. Nevertheless, in view of the progressive nature of our disability this seemingly simple goal is

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not easily attainable. There are many conditions that have to be met, such as:

- Regular medical supervision and the opportunity to attend rehabilitation programmes (physiotherapy, respiratory exercises, swimming in the sea/pool and other activities to preserve health);
- Access to new treatments available worldwide;
- The possibility to use 24/7 personal assistance at home;
- Accessible (barrier-free) environment;
- Access to orthopaedic devices;
- Transportation with adapted vehicles; and
- The possibility to use fully accessible public transportation.

All these are the pre-requisites for setting up the conditions for people with NMDs to participate in society, to be regularly involved in the working environment, and to have the basic conditions for creativity, employment and family life. Policymakers play the main role in ensuring that the national law supports those goals in practice. That means that the abovementioned services should always be available and financed through public funds. The levels of those services differ throughout the European countries. For example, the situation in Western European countries is better in comparison with Eastern European countries, although they are all full EU members. The situation is even worse in the candidate countries for EU membership. There are also cases where the country officially supports the services and accessibility for people with NMDs, but in real life this is not implemented, because of lacking legal

regulations. It is known that many EU countries support equal rights of people with disabilities in their national constitution. However, practice shows that it is not enough only to write down those rights in the constitution while the mechanism for supporting people with NMDs does not work, because of low political support and consequently insufficient public funds. But I believe that the situation is gradually improving. To this end, strong co-operation between NMD associations through EAMDA is essential.

**How does EAMDA work to improve the quality of life of people with neuromuscular disorders in Europe?**

EAMDA co-operates with different European NMD associations and their representatives that have connections with local decision makers. EAMDA is a member of different international associations such as the European Patients' Forum (EPF), European Disability Form (EDF), European Federation of Neurological Associations (EFNA) and EURORDIS-Rare Diseases Europe, and it also co-operates with other respective associations active in the field of neuromuscular disorders. Its main goal is to establish an accessible and inclusive environment for people with neuromuscular disorders as well as to ensure the necessary medical and social services, with the final aim to achieve their social inclusion.

EAMDA organises regular annual general assemblies (AGAs) in a different European country each year, and this is currently our main activity. As an umbrella organisation EAMDA prepares the extensive medical and social part of the conference for people with neuromuscular disorders as well as for scientists and other experts. AGAs represent an opportunity to learn about the obstacles and challenges people with NMDs are faced with in the presented country and, even more importantly, to convince the national policymakers that providing services for people with NMDs does not represent only costs but also an opportunity to build an inclusive society regardless of disability.

**You have recently been elected to a four-year mandate as president of EAMDA – what will some of your key priorities be during this time?**

EAMDA has quite a long tradition in the European Union. It was established in 1974 in London, UK. It later moved to Valetta, Malta, and

**Accessible devices, facilities and services are the prerequisites for setting up the conditions for people with NMDs to participate in society**

in 2008 to Ljubljana, Slovenia. Although Slovenia is a European country, it took quite some time for our decision makers to recognise the global importance of EAMDA and to establish its legal status according to the Slovenian and European regulations. After struggling with the local bureaucracy, we finally obtained the legal entity status.

In my new term of office, I will therefore focus on activities that will enable EAMDA to successfully compete for European projects, which is what we need in EAMDA for the further development of our activities. To that end we need knowledge exchange and information transfer between different organisations in the same field, together with the support of our member associations and assistance of individual researchers. In order to successfully regulate the quality of life of persons with NMDs, the existing patient identity should be formally complemented with the disability identity since all EU states have well-developed disability policies and protection of disabled people which provide numerous social rights that persons with NMDs need in order to achieve their active social roles.

Therefore, my main focus as EAMDA president will be to improve the quality of life of people with NMDs in terms of equal treatment, technical aids, personal assistance, disability, independent living, and social inclusion. EAMDA cannot compete in the field of basic research, but it can creatively contribute to the regulation of the social life of persons with NMDs, and even more importantly it can strengthen the organisation and increase the efficiency of national NMD organisations, especially in Central and Eastern Europe.



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