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This policy was adopted by the 8th EMSA Spring Assembly in Lisbon on 8th of May of 2022. Must be reconsidered until: 8th of May of 2026.

Rare Diseases

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The European Medical Students´ Association (EMSA) represents medical students across Europe. We envision a healthy and solidary Europe in which medical students actively promote health. EMSA empowers medical students to advocate health in all policies, excellence in medical research, interprofessional healthcare education and the protection of human rights across Europe.

Problem statement.

In the European Union (EU), a disease is considered to be rare when it affects less than 5 in 10 000 individuals. While the diseases are uncommon, rare diseases are not.There are estimated to exist between 5 000 and 8 000 rare diseases. Combined all rare diseases are estimated to affect one in 15 individuals worldwide, accounting for around 30 million residents in Europe (European Commission). However the numbers are thought to often be underestimated, as most patients who are diagnosed with a rare disease are not mentioned in epidemiological databases, as well as rare diseases being heavily underdiagnosed (WHO, 2004).

Most rare diseases have a genetic basis, and are characterised by an important heterogeneity in symptoms. Nevertheless they often share common characteristics within themselves such as their chronic, severe and degenerative course. Even though rare diseases affect patients in different ways, many struggles are shared: the long and hard journey to diagnosis, the difficult access to specialist care, the small number of patients with each condition and their dispersion around the world making them prone to not receive sufficient medical attention, limited treatment options and emotional burden (Uhlenbusch, 2019). This being said, when addressing rare diseases it is crucial that we have the patient experience in mind, especially during the pre-diagnosis stage when patients with rare diseases often experience significant trauma and we can't undervalue the role of the healthcare system in these situations.

Patients with rare diseases experience an average diagnostic delay of 7,6 year, frequent misdiagnosis, and negative interactions with healthcare professionals. Especially female presenting patients with chronic pain, commonly experience medical gaslighting (Newton, 2013), meaning that they regularly experience doctors blaming their illness or symptoms on psychological factors, or even deny their illness entirely. While being common experiences in the rare disease community these problems are rarely addressed in scientific publications.

During the post-diagnosis stage of patients with rare disease, the burden of living with a rare disease is characterised by lack of information, lack of experienced or knowledgeable healthcare providers, poor accessibility to treatment or even lack thereof. In fact, there is only a 5% or less chance of treatment for a rare disease. For the cases in which treatment exists they are usually highly expensive, and coverage is decided on individual cases.

The EU's and WHO strategic objective for rare diseases is “to improve patient access to diagnosis, information and care with some specific measures, including:

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- Improving recognition and visibility of rare diseases;
- Ensuring that rare diseases are adequately coded and traceable in all health information systems;
- Supporting national plans for rare diseases in EU countries;
- Creating European reference networks linking centres of expertise and professionals in different countries to share knowledge and identify alternative treatment options;
- Support patient organisations as they are particularly important because they provide additional incentives for developing orphan drugs to combat rare diseases;"

Overall, efforts have been made in raising awareness on rare diseases, improving patient care, developing new treatments and regulating orphan drug policies. The European Union has already acknowledged the unique needs of rare disease patients and highlighted the importance of improving their care by introducing national rare disease policies in EU member states. It is also important to state the central role of strong advocacy efforts made from patient support groups and international NGOs, whose members usually live with a rare disease and therefore are highly motivated to achieve better access to good quality healthcare.

Besides all the initiatives and efforts, there are still many remaining challenges to face in the field of rare diseases, where public funding of fundamental research remains crucial, alongside investments in dedicated infrastructure and international networks, stimulating collaboration and bringing together medical experts, reference centres and patients' groups. Equally important is the availability of an internationally recognized rare disease classification system which can help generate reliable epidemiological data.

Investment in any drug development project carries a significant risk for pharmaceutical companies, especially in rare diseases, where the risk is compounded by the fact that the target population is small and therefore the return of investment and profit for the pharmaceutical companies is limited. With all these challenges, funding of clinical trial programmes remain essential for orphan drug development, especially for rare diseases that appear less attractive for the pharmaceutical industry.

Overall, great progress has been made in raising awareness in rare diseases, improving patient care, developing new treatments and regulating orphan drug policies. The European Union has already acknowledged the unique needs of rare disease patients and highlighted the importance of improving their care by introducing national rare disease policies in EU member states. It is also important to state that all these achievements were fueled by strong advocacy efforts from patient support groups and international NGOs, whose members usually live with a rare disease and therefore are highly motivated to achieve better access to good quality healthcare.

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Our view. Aim

We, as European Medical Student Association (EMSA), strongly believe that rare diseases patients, as a vulnerable population, deserve additional efforts to ensure quality and equitable access to care. This being said, rare diseases should be viewed as a global health issue, requiring intra and inter-country efforts, especially in raising awareness and promoting research.

Being an organisation that includes and represents future researchers and clinicians, and therefore is a key point to raise awareness and advocate on the subject of rare diseases. EMSA has made continuous efforts on the matter, realising the continuous insufficient support for patients living with a rare disease in European countries as well as the lack of formal education on rare diseases within the medical schools curricula, teaching not only the particularities of RD and their diagnosis and treatment but also on the patients experience. Despite the progress made in recent years in the rare disease field, there is still much work to do at a local, national and European level. As this goal is going to be challenging and the effects not immediate, it is crucial that rare diseases receive more attention and are more commonly included in the european health agenda, with the following priorities:

- Enable a rapid diagnosis for all in order to reduce diagnostic delays and undiagnosed diseases;
- Innovate in order to treat, increase therapeutic resources through research and policies focused on improving patient experience and outcomes;
- Improve quality of life and autonomy of patients;
- Communicate and train, promoting the sharing of knowledge and expertise in the field of rare diseases and involving patient groups in the discussion;
- Modernise organisations and optimise national funding mechanisms;

Recommendations

EMSA calls on the World Health Organisation, European Commission and United Nations to:

- Involve Patient organisations, and their members, in all future work, drawing on the extensive patient experiences;
- Support research on the patients experiences;
- Provide technical support to governments in the form of evidence based guidelines and policies;
- Coordinate international cooperation and dissemination of best practices in Rare Diseases national plans;

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- Promote international collaborations between researchers to conduct meaningful studies on rare diseases;
- Promote the development of rare diseases treatments through national and international orphan drug policies and foster inter-europe collaborations in negotiations of orphan drug prices with pharmaceutical companies;
- Meaningfully engage, encourage and support youth initiatives and organisations working on rare diseases, including them in discussions and the decision-making process.

EMSA calls on the Rare Diseases International, EURORDIS, Rare Diseases Activists and Associations to:

- Continue to demand that their members experiences get heard, and are the main focus point of any future work;
- Increase their efforts on creating a multi-sectoral approach to overcome psychological, financial, and social burdens of people living with rare diseases.
- Facilitate and encourage networking amongst patient groups;
- Provide a safe space for patients to share their trauma - private, and medical;
- Promote the sharing of common experiences and common knowledge of the rare disease community;
- Raise awareness on social burden of rare diseases amongst patients, families, caregivers and public;
- Develop educational materials empowering undiagnosed patients and their caregivers to seek diagnosis;

EMSA calls on EFPIA and IFPMA to:

- Initiate collaboration and cooperation between patient organisations, scientists, healthcare workers and policy-makers to drive research in developing treatments;
- Share existing knowledge and understanding of rare diseases to prevent duplication that slows down the process;
- Continue their dialogue with international and national authorities on orphan drug legislations that foster an innovation oriented ecosystem.

EMSA calls on EU Member States, National Governments, Health Ministries & other relevant Ministries to:

- Establish national inter-disciplinary working groups on rare diseases and develop evidence based national policies and strategies for improvement of care for patients living with rare diseases;
- Support and promote the engagement of patients and patient support groups at every step of the consultation, drafting and implementation of Rare Disease plans and strategies;

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- Allocate funds to assist the work of patient support groups and finance basic, translational and clinical research and education on rare diseases.

EMSA calls on Human Rights Organizations, NGOs, especially NGO Committee for Rare Diseases, and Civil Society Organisations to:

- Continue to support rare disease patients on a local, national and european level, including advice on access to specific patient support groups, social services, etc
- Support international collaborations between patient support groups to share expertise and promote collaborations;
- Continue to advocate for the rare disease community on a local, national and european level.

EMSA calls on European medical faculties and universities to:

- Establish a curriculum for undergraduate education of medical students that includes the particularities of Rare Diseases and their diagnosis and treatment but also the patients experience;
- Capacitate professors and educators on Rare Diseases;
- Promote exchanges of experience and expertise in teaching about rare diseases though conferences and collaborations.

EMSA Calls on Healthcare workforce and Healthcare facilities to:

- Continuously keep up to date with new developments in rare diseases;
- Organise workshops and lectures to promote professional development in the area of rare diseases amongst healthcare workers;
- Conduct research into social and clinical aspects of rare diseases and offer medical students an opportunity to get involved.

EMSA calls on EMSA members and Healthcare students to:

- Support Rare Disease Associations and raise awareness about rare diseases, and the experiences of rare disease patients;
- Actively seek opportunities to learn about rare diseases, including rare disease patients, learning with their expertise about their condition and the struggles they face;
- Get involved in clinical, scientific and social science research on rare diseases and provide constructive feedback on current rare disease curriculum, making suggestions for improvement to the teaching staff based on own experience and research.

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References

1. De Vrueh, R., Baekelandt, E. R. F., & De Haan, J. M. H. (2013). Background paper 6.19 rare diseases. *World Health Organization, Geneva*. Retrieved March 3, 2022, from https://www.who.int/medicines/areas/priority_medicines/BP6_19Rare.pdf

2. Ministry of Solidarity and Health, Ministry of Higher Education, Research and Innovation. French National Plan for Rare Diseases 2018-2022. Retrieved March 3, 2022, from https://solidarites-sante.gouv.fr/IMG/pdf/pnmr3_-_en.pdf

3. Moliner, A. M., & Waligora, J. (2017). The European Union Policy in the Field of Rare Diseases. Advances in Experimental Medicine and Biology, 1031, 561–587. Retrieved March 3, 2022, from https://doi.org/10.1007/978-3-319-67144-4_30

4. Newton, B. J., Southall, J. L., Raphael, J. H., Ashford, R. L., & LeMarchand, K. (2013). A narrative review of the impact of disbelief in chronic pain. *Pain Management Nursing*, *14*(3), 161–171. Retrieved March 3, 2022, from https://doi.org/10.1016/j.pmn.2010.09.001

5. *Orphanet: About rare diseases*. (2012, October 25). Orpha.Net. Retrieved March 3, 2022, from https://www.orpha.net/consor/cgi-bin/Education_AboutRareDiseases.php?lng=EN

6. Rare diseases. (n.d.). European Commission. Retrieved March 3, 2022, from https://ec.europa.eu/health/non-communicable-diseases/steering-group/rare-diseases_en

7. Uhlenbusch, N., Löwe, B., & Depping, M. K. (2019). *Perceived burden in dealing with different rare diseases: A qualitative focus group study*. *BMJ Open*, *9*(12). Retrieved March 3, 2022, from https://doi.org/10.1136/bmjopen-2019-033353

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