

Rare Conversations - Take Two

Keeping Rare Disease Patients In The Centre Of Discussion - In cooperation with EuropaBio -

- On February 4th, Alexion continued its **RareConversations** event series in cooperation with EuropaBio. **Maciej Gajewski, Executive Director and Head of Public Affairs and Policy at Alexion** operated as moderator for the conversation on the importance of **keeping Rare Disease patients in the centre of discussion** between panellists from the patient, regulatory and industry community.
- **Patient advocate Alastair Kent** presented a videoclip showing impactful stories of Rare Disease patients and their families. Subsequently, he was interviewed by **Walter Atzori, Alexion's Head of Patient Advocacy International**.



Mr Kent stated that the pandemic has promoted the use of tele-medicine to allow for more frequent interactions with practitioners. It also spawned creative ways of conducting clinical trials. Similar creativity is needed from regulators in the area of Rare Disease.



Stelios Kypouropoulos, Member of the European Parliament, expressed the need for research to not only focus on the frequency of Rare Diseases, but also on severity and the impact on children. The quality of life of Rare Disease patients and their social impact should be considered as well.



Simone Boselli, Director of Public Affairs, EURORDIS, supported MEP Kypouropoulos' statement and specified that the creation of knowledge is key to combatting all Rare Diseases, not just the prevalent ones. The ecosystem is already in a good place but needs to be further expanded.



Patient expert for haemophilia Dr Cees Smit highlighted positively that the life expectancy in his disease area has significantly increased over the last decades, but also recognised that there is still a lot of work to do, notably with regards to the gender aspect and the global unmet need.



Bernard Grimm, Healthcare Biotechnology Director, EuropaBio, pointed at the fact that there is still a lack of knowledge about Rare Diseases compared to regular diseases. That is why the level of investment is important, especially for SMEs.



Dr Denis Horgan, Executive Director, EAPM, noted the progress already made as well, but saw a further need to integrate research results into the healthcare systems. A system developed around the ERNs could facilitate access and provide more targeted treatments.

Key take-aways

- Persons affected by Rare Diseases should not only be viewed or treated as patients. Besides lifesaving treatment, the **impact on their quality of life should also be taken into consideration when drafting legislation.**
- Research in the Rare Diseases area has already advanced a lot in the last decades: the life expectancy has dramatically increased for some Rare Diseases and for other ones the granular expertise has increased. However, there **still exists a lack of knowledge in comparison to regular diseases. That is why the level of investments is important, especially to support SMEs.**
- Many important decisions about the life of people affected from Rare Diseases are taken without them. **Patients should not only be at the center of discussion, but also lead the discussion themselves.**
- Roughly 70% of Rare Diseases are of genetical origin. Therefore, an important measure is the utilization of new-born screenings, in order to allow families to start preparing and assessing available therapies earlier.
- Patient stories are heard by regulators, but the focus often lays on the monetary aspect of potential support measures. **It needs to be made clear that finding a treatment means that the dependency of Rare Disease patients on society and the social security systems also decreases.**
- **Defining unmet needs can be a difficult task, because prioritization could potentially lead to disparities.** Even if a treatment already exists, better and more targeted solutions can still be found. Furthermore, medicines can be repurposed for another disease to create better outcomes for patients.
- Following a multi-stakeholder approach by **leveraging the European Research Networks and the shared expertise of the community** can be crucial to facilitate access and provide for targeted treatments.
- The rapid vaccine availability for COVID has shown that collaborative and innovative efforts to combat an existing health threat are possible. The same urgency should be applied to Rare Diseases and kept in mind when reforming the regulatory framework.
- The regulatory environment is already in a good state, but there is still room for improvements, especially to address the high unmet need by creating new treatments. **Innovation needs to be further incentivized to close the knowledge gap towards regular diseases.**

