efpia
European Federation of Pharmaceutical Industries and Associations

Draft ☐ Final ✱

#WeWontRest Until We Make Treatments for Rare Diseases Less Rare

The innovative pharmaceutical industry urges policymakers to support robust incentives for research and development of orphan medicines to give hope to patients living with rare diseases

To give hope to the 30 million[i] (1 in 17) Europeans living with a rare disease, all partners in healthcare need to work together to ensure that patients get access to currently available treatments. In addition, we need to further incentivise the research and development of new treatments for patients, where, currently, no treatment options exist. Neither of these two imperatives will be addressed by jeopardising a proven, effective Regulation - they require separate approaches and different solutions.

Incentivisation of research and development of orphan medicines is a good example of how an EU Industrial Strategy can channel research and innovation towards meeting patients' unmet needs, and also strengthen the European economy. Meanwhile, any concerns on the good functioning of the Orphan Medicinal Products Regulation can be best addressed in the framework of an EU multi-stakeholder Forum for Better Access to Health Innovation.

In the context of the current evaluation[ii] of the OMP Regulation, EFPIA calls on European institutions to ensure a stable and predictable regulatory and incentives' framework. This will support further research and development into much needed new treatments for rare diseases.

The EU Orphan Regulation has brought hope to people living with rare disease

Rare diseases are often genetic in origin, mostly emerging in childhood and result in a severe burden of illness. Diseases such as amyotrophic lateral sclerosis (ALS), chronic lymphocytic leukaemia, Duchenne Muscular Dystrophy, or Wilson's disease, to name a few, are complex, scientifically challenging and heterogenous diseases in nature, which despite being rare or very rare, collectively affect 30 million Europeans[iii].

In 2000, the European Union adopted the **Regulation on Orphan Medicinal Products**[iv] ("Orphan Regulation") to address the needs of Europeans living with such a rare disease. Prior to its introduction, research in this area was limited and "*too few and did not lead to significant progress in research*"[v]. Given the small patient populations associated with each rare condition, the complexity and risk associated with drug discovery and developments, and the corresponding limited commercial viability, there was mounting concern that without incentives to encourage research and innovation, no or too few new specific treatments would be developed for these serious conditions.

The Orphan Regulation has stimulated the development of new treatments for patients living with previously untreatable rare diseases – we have seen an increase from just 8[vi] authorised products for rare diseases before the entry into force of the Regulation, to 164 today[vii].

Europe has benefited greatly from the adoption of the Orphan Regulation and the targeted incentives it established. It stimulates R&D (e.g., *via* fee reduction and protocol assistance) into treatments showing great potential, and it rewards with 10-year market exclusivity those that ultimately make it through the long and complex development process, to become a new treatment and hope for patients. The benefits of the Orphan Regulation have led to:

- Greater understanding of rare diseases which is essential to the development of new treatments:
  - the number of clinical trials focused on rare diseases increased by 88% between 2006 and 2016 in the EU region alone[viii],
  - resulting in 2,121 new orphan designations[ix], which in turn
  - led to 164 authorised new treatments for around 90 rare diseases between 2000 and 2018, compared to only 8 prior to the Regulation – an average of 9 new medicines per year;
- Raising the profile of rare disease in Europe, making it easier for patients to form networks thereby accessing crucial information and communicate their experience and expertise across the research and healthcare communities;

Since the adoption of the Regulation, 23 European countries also adopted at least 1 national rare disease plan[x].

These incentives have transformed the lives of patients and their families, improving health outcomes and contributing to a thriving economy. They are central to developing treatment options for previously untreatable

conditions, they advance medical knowledge more broadly, as well as foster a supportive research, innovation and investment environment in Europe.

95% of Rare Diseases still need treatment options

We believe that no patient should be left behind. The Orphan Regulation is critical to foster further investment and innovation in Europe to deliver new treatments for these remaining rare diseases.

Despite these significant advances, 95%[xi] of rare conditions remain without a treatment option. This is due to the many distinct rare diseases that exist globally (estimated 5000 to 8000), small and scattered patient populations associated with each condition, lack of validated biomarkers and end-points, poor diagnostics and limited clinical expertise as well as the complexity and risk associated with drug discovery and developments[xii].

In addition to our collective responsibility to find new treatment options for patients living with rare diseases, the Orphan Regulation can play a role in a future EU industrial strategy, helping to promote sustainable innovation. According to the Economic and Societal Footprint study among biopharmaceutical companies[xiii], "(…) phasing out current incentives in Europe would have a material negative effect on their European operations, with over half [of the respondents] indicating that this scenario would lead to a reduction in their R&D and commercial footprints of more than 25%. (…) Weaker intellectual property regimes limit the willingness of companies to invest in R&D [in Europe]."

Access: a shared goal and responsibility

In view of the needs regarding access to novel orphan treatments, the research-based pharmaceutical industry is committed to working together with stakeholders to find new pricing models to finance these, thus ensuring access for patients and improving sustainability for health systems. Re-opening the Regulation, designed to support research into new treatment options, will not improve patient access - now or in the future. Finding new and collaborative approaches involving all relevant stakeholders, will.

New hope comes from new treatments and early diagnosis both in child and adult patients. The success and value of the Regulation in stimulating the development of new treatments can only be fully realised if patients get access to them.

There are challenges regarding access to novel orphan treatments and considerable inequalities between, and sometimes even within, Member States. According to the 2018 PATIENT W.A.I.T. Indicator[xiv], not only are orphan medicines less available but they also take more time to reach patients following European Medicines Agency approval in over 80% of the 28 analysed European countries.

Ensuring patient access to the new treatments of today and tomorrow is a shared goal and responsibility. It requires regulators, health system partners, patients, governments and industry sitting together to find new ways to finance these innovative treatments and to ensure access for patients and sustainability for health systems at national level. There has been some progress:

- Companies already engage in innovative arrangements, such as managed entry agreements and early access schemes, to support patients' access to new medicines.
- A number of countries across Europe have adapted HTA and reimbursement practices to best capture the value that these products bring.

EFPIA believes that is time for a different type of conversation calls on the European Institutions to implement the EU Health Coalition's call to establish a **multi-stakeholder Forum on Better Access to Health Innovation**[xv] that brings all actors together to discuss how to ensure access to new treatments and technologies today, medical innovation for tomorrow and sustainable healthcare systems in a globally competitive Europe. Only through such a dialogue a meaningful response to the challenges of healthcare systems can be identified and implemented.

[i] http://www.orpha.net/national/data/NO-NO/www/uploads/Orphanet_publication_pointprev_2019.pdf
[ii] https://www.who.int/medicines/areas/priority_medicines/BP6_19Rare.pdf
[iii] https://ec.europa.eu/info/research-and-innovation/research-area/health/rare-diseases_en
[iv] https://ec.europa.eu/health/sites/health/files/files/eudralex/vol-1/reg_2000_141_cons-2009-07/reg_2000_141_cons-2009-07_en.pdf
[v] https://ec.europa.eu/health/sites/health/files/files/orphanmp/doc/orphan_inv_report_20160126.pdf
[vi] Mark L Flear, Anne-Maree Farrell, Tamara K Hervey and Therese Murphy (eds) (2013) European law and New Health Technologies. Oxford Studies in European Law.
[vii] https://www.ema.europa.eu/en/documents/other/orphan-medicines-figures-2000-2018_en.pdf
[viii] https://www.pugatch-consilium.com/reports/Benchmarking_success.pdf
[ix] https://www.ema.europa.eu/en/documents/other/orphan-medicines-figures-2000-2018_en.pdf
[x] http://www.europlanproject.eu/NationalPlans?idMap=1
[xi] Global Genes, Rare diseases: Facts and Statistics, https:// globalgenes.org/rare-diseases-facts-statistics/
[xii] https://www.pugatch-consilium.com/reports/Benchmarking_success.pdf and https://www.who.int/medicines/areas/priority_medicines/BP6_19Rare.pdf
[xiii] https://www.efpia.eu/media/412939/efpia-economic-societal-footprint-industry-final-report-250619.pdf
[xiv] https://www.efpia.eu/media/412747/efpia-patient-wait-indicator-study-2018-results-030419.pdf
[xv] https://www.euhealthcoalition.eu/recommendations/