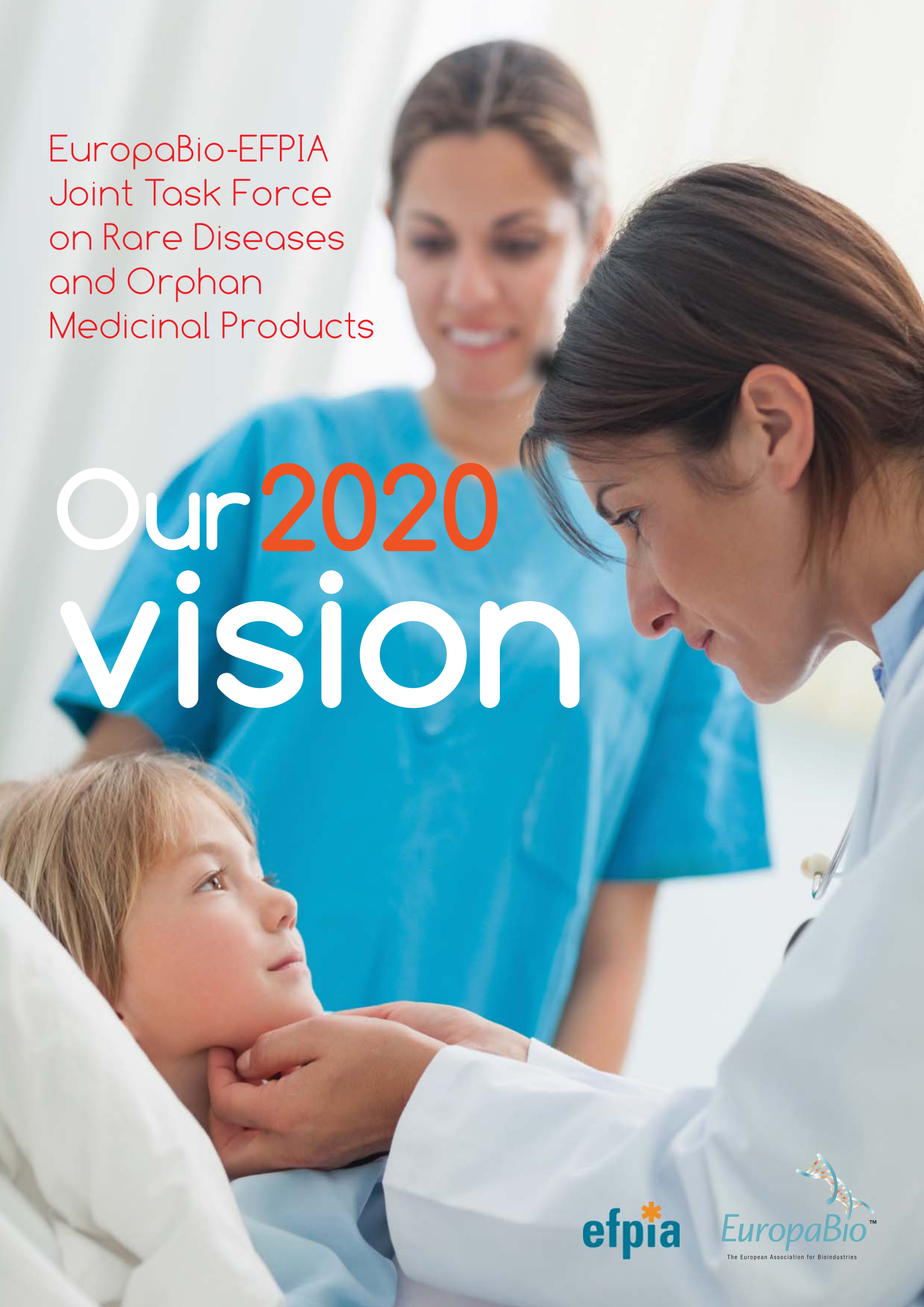
EuropaBio-EFPIA Joint Task Force on Rare Diseases and Orphan Medicinal Products

Our 2020 vision



efpia

EuropaBio™

The European Association for Bioindustries

Our 2020 vision:

Rare diseases is maintained as a **health policy priority** to ensure timely and equitable patient access

Unmet medical need is increasingly met with **effective new treatments**

Rare disease patients experience **the best possible clinical outcomes**

There is **timely and accurate diagnosis** with adequate use of the latest technology

Patients have **equal and timely access to existing treatments** across Europe

Patients can enrol **quickly in clinical trials**

1. Key Facts about Rare Diseases and Orphan Medicinal Products

Rare diseases

A rare disease is considered in the EU as a **life-threatening or chronically debilitating condition** affecting not more than five in 10,000 persons in the Community.[1]

This means that today, of the current population in the EU of 740 million people, up to 370,000 people may be suffering from one of the 6,000 to 8,000 rare diseases identified so far. Half of these rare diseases affect children.[2]

Role of orphan medicinal products

Orphan medicinal products have been proven to have a significant impact on patients' lives and well-being, as they provide solutions for the diagnosis, prevention or treatment of rare diseases. They are often the sole therapeutic option for patients and so help address unmet medical need. As such, they are life-saving or can have a significant impact on the quality of life of individual-patients.

Regulation (EC) No 141/2001 on orphan medicinal products states that patients suffering from rare conditions deserve the same quality, safety and efficacy in medicinal products as other patients. It also recognises that due to rarity, patients with rare diseases have not benefited from medical innovation to the same extent as patients with common diseases and concludes that regulatory and economic incentives are required to ensure that in future patients suffering from rare conditions can benefit from advances in medical innovation.

National Plans for rare diseases: improving patient access

Although Regulation (EC) No 141/2001 has had a direct positive impact on the number of orphan medicinal products approved in the EU, patients face significant challenges in getting access to approved medicines and there are inequalities evident across the EU.

To address some of the issues related to patient access, in June 2009, the Council of the European Union unanimously adopted a Recommendation on an action in the field of rare diseases. In the Recommendation, EU Member States agreed to establish and implement plans or strategies for rare diseases to aim to ensure that patients with rare diseases have access to high-quality care, including diagnostics, treatments, habilitation for those living with the disease and, if possible, effective orphan medicinal products. Many EU Member States have now developed and adopted a national plan for rare diseases, which is having a tangible impact on the lives of patients with rare diseases. We encourage **Member States to fulfill the commitments made in the Council Recommendation on an action in the field of rare diseases to adopt and implement a national plan for rare diseases, with sufficient funding**.[3]

1. http://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:L:2000:018:0001:0005:en:PDF
2. http://ec.europa.eu/health/rare_diseases/policy/index_en.htm
3. http://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:C:2009:151:0007:0010:EN:PDF

2. Innovation and Discovery

The development of orphan medicinal products and associated research in rare diseases has an important impact on medical innovation, through the better understanding of diseases and the development of new cutting-edge technologies. The orphan medicinal products model also serves as a precursor for the future of healthcare innovation through more targeted and personalised treatments. Regulation (EC) No 141/2001 has had a significant positive impact on the level of research and development dedicated to orphan medicinal products. Before the adoption of the Regulation, there were almost no approved treatments for rare diseases in the EU. **Since the Regulation came into force, more than 90 new treatments for patients with rare diseases have been approved at European level.**[4] Many patients with rare diseases have benefited as a result of this Regulation. **The EU regulatory framework for orphan medicinal products provides clarity, predictability and is an enabler of innovation.**

> “Before the adoption of the Regulation, there were almost no approved treatments for rare diseases in the EU

Despite the success of the Regulation, the development of orphan medicinal products remains a risky undertaking for those investing in the research, development and commercialisation of these treatments. Due to the rarity of these diseases, there is often only partial knowledge of the disease, scarce medical expertise and recruitment for clinical trials is a challenge. In addition, orphan medicinal products are complex products, they are often biologics and often first-in class, with lengthy development plans. **It is crucial that the appropriate legislative and clinical development framework exists to support innovation in the field of rare diseases; in particular the enrolment of patients in clinical trials.**

3. Diagnosis

It is crucial to increase cooperation among various stakeholders and support the further research that is needed to ensure accurate diagnosis of rare diseases at an early stage. Despite progress made in this space, there is a need to better understand the obstacles patients and caregivers within the rare disease community face in obtaining a correct and timely diagnosis.

> “ It is important to make the right diagnosis early in life and to treat at the right moment

Most symptoms of rare genetic diseases appear during childhood. However, for many conditions they may not occur or might be missed in the first days or months following birth, even though irreversible damage might have already been done. In these instances, it is important to make the right diagnosis early in life and to treat at the right moment to achieve better health outcomes for the child. Newborn screening could support this goal. Most EU Member States provide newborn screening for fewer than ten conditions, even though the use of new technologies has made screening for a wide variety of disorders much more affordable. Newborn screening policies of EU Member States also vary significantly with respect to the conditions screened for, and the processes and criteria by which new conditions are added to their screening panels.[5]

The EU has recognised the need for collaboration among Member States in order to improve the detection, diagnosis and treatment of rare diseases.[6] **We encourage the EU institutions to support increased cooperation between Member States to improve rare diseases diagnosis and develop and harmonise the criteria to decide which conditions countries should add to their newborn screening panels.**

4. http://ec.europa.eu/health/documents/community-register/html/alforphreg.htm

5. The number of conditions screened for varies from one or two to more than twenty conditions. In some countries, such as Italy, there is also regional variability in the number of conditions screened. See more: Vittozi L. et al. Executive Report to the European Commission on Newborn Screening in the European Union (2012) EU Tender “Evaluation of population newborn screening practices for rare disorders in Member States of the European Union”.

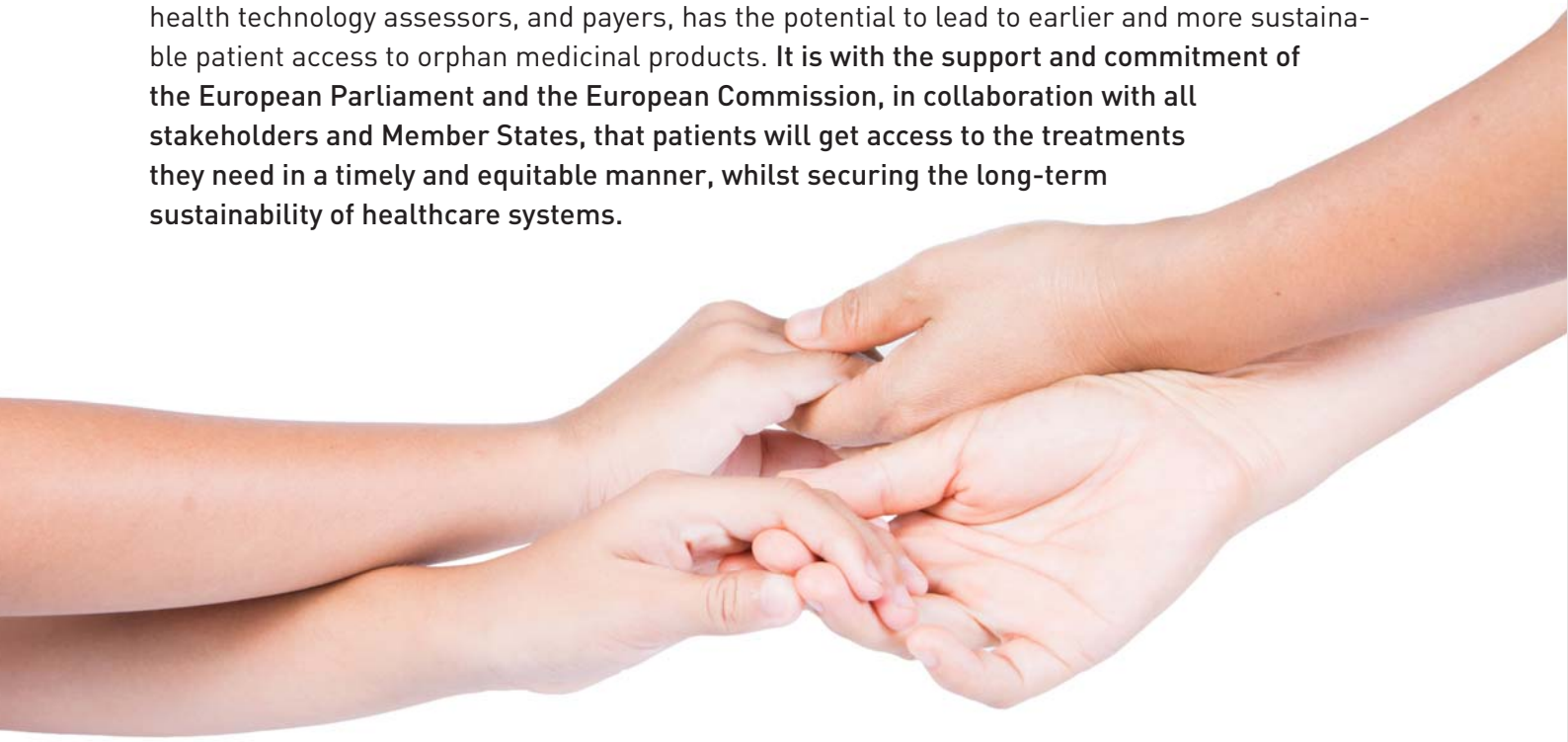
6. The framework for this collaboration is outlined in two documents: the 2008 Communication from the Commission to the European Parliament, The Council, The European Economic and Social Committee and The Committee of the Regions on Rare Diseases: Europe’s Challenges and the Council Recommendation of 8 June 2009 on Action in the Field of Rare Diseases.

4. Patient Access

While the majority of patients in Europe do have access to innovative medicines to treat rare diseases, there remain significant challenges, particularly in low-income countries where patients often face long delays in getting access to approved treatments. Therefore, access to orphan medicinal products may vary greatly from one European Union country to another.

> "There remain significant challenges, particularly in low-income countries

Various initiatives have been developed to address the challenge of patient access, which is recognised by stakeholders throughout the healthcare system: better and earlier collaboration between all actors in the value chain, with industry together with regulators, health technology assessors, and payers, has the potential to lead to earlier and more sustainable patient access to orphan medicinal products. **It is with the support and commitment of the European Parliament and the European Commission, in collaboration with all stakeholders and Member States, that patients will get access to the treatments they need in a timely and equitable manner, whilst securing the long-term sustainability of healthcare systems.**



5. The sector's contribution to society

The development of orphan medicinal products is of significant value to society and the economy as a whole. With adequate treatment, patients and parents of young patients affected by rare diseases will be able to better contribute to society and the economy. Better access to orphan medicinal products can also contribute to reducing the average hospital stay for rare disease patients or decreasing patients' reliance on carers' help. With a better quality of life, individual patients are better contributors to society as a whole. The orphan medicinal products sector also generates growth and jobs, including a number of fast-growing SMEs in this sector.

The overall budget impact of orphan medicinal products is limited and sustainable. The budget impact is estimated to have grown from 0% in 2000 to 3.3% by 2010. From 2010 to 2016 this growth is predicted to continue, however the rate of growth is likely to slow and will reach a plateau of approximately 4.6% of total pharmaceutical market expenditure by 2016.[7]

7. Ref : Schey et al. Estimating the budget impact of orphan medicines in Europe: 2010 – 2020. Orphanet Journal of Rare Diseases 2011, 6:62 http://www.ojrd.com/content/6/1/62

A call to Europe

(Member States, European Commission and European Parliament)

- It is crucial that the focus on rare diseases is maintained as a health policy priority and that decision-makers work with all stakeholders, to **develop smooth pathways at EU and national level that bring innovative therapies to patients**.

- Member States are encouraged to **fulfill the commitments made in the Council Recommendation on an action in the field of rare diseases to adopt and implement a national plan for rare diseases, with sufficient funding**.

- We support **increased cooperation between Member States to improve accurate early diagnosis of rare diseases and develop/harmonise the criteria to decide which conditions countries should add to their newborn screening panels**.

- We support a more **holistic, joined-up approach that addresses the challenges of patient access to treatments**. The orphan medicinal products industry is fully committed to making this approach a reality. We also support the adaptation of Health Technology Assessment processes, methodologies and decision-making frameworks to the specificities of orphan medicinal products, so that rare disease patients have equitable access to treatments.

- We encourage policy makers to **maintain their commitment on what EU health systems can and should deliver in terms of preventing and treating rare diseases**. Maintaining such an ambition will ensure that new products continue to be developed, contributing to a healthier and more productive society in the future, while creating high-quality jobs and adding value to the European economy.

September 2014

Joint EFPIA-EuropaBio Task Force on Rare Diseases and Orphan Medicines

The Joint Task Force between EFPIA (the European Federation of Pharmaceutical Industries and Associations) and EuropaBio (the European Association for Bioindustries) on Orphan Medicinal Products and Rare Diseases is a European alliance of over 45 member companies committed to the development of orphan medicinal products (OMPs) and improving the lives of patients with rare diseases.

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