friends of europe

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IMPROVING ACCESS, CARE AND DIAGNOSIS FOR RARE DISEASES

REPORT

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In association with

IMPROVING ACCESS, CARE AND DIAGNOSIS FOR RARE DISEASES

Around 30 million Europeans suffer from rare diseases such as cystic fibrosis, Duchenne muscular dystrophy or Hunter syndrome. As there are more than 6,000 known rare diseases and therapies exist for only 400 or so, there remains very high unmet need in rare diseases.

Because 80% of rare diseases have a genetic component, patients cannot influence or cope with their disease through lifestyle changes or using other drugs, as there are often no alternative therapies. Furthermore, more than half of rare disease patients are children; of those, 30% will die before their fifth birthday.

Even where treatments exist, patients are often not getting access to them because of budget pressures, bureaucratic obstacles to investment or problems of scale, panellists told a Friends of Europe 'Café Crossfire' lunch debate on Wednesday 21 June. They called for more collaboration between doctors, industry, insurers, patients and EU governments to solve this issue.

The European Commission has already introduced incentives for pharmaceutical companies to develop new drugs. This has increased the number of therapies available to treat rare diseases from eight to over 130 in the last 15 years. A series of European Reference Networks (ERNs) have also been set up to bring patients and specialists together to help speed up and aid treatment. But more 'soft incentives' are needed to encourage more evidence-gathering, to invest in prevention, and to train doctors to diagnose rare diseases.

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Director-General of the European Federation of Pharmaceutical Industries Associations (EFPIA)

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Chief Executive Officer of the European Organisation for Rare Diseases (Eurordis)

NOT ‘BUSINESS AS USUAL’

Rare diseases affect fewer than five people in every 10,000 with very rare diseases affecting only one in 100,000). Because they are rare, there is often a shortage of expertise, money and treatments available, though the picture differs across EU countries. “Clearly this is an area where there is a need for critical mass,” said Xavier Prats Monné, European Commission Director-General for Health and Food Safety. “Patients are the ones we should be thinking of.”

The problem for industry, said **Nathalie Moll**, Director-General of the European Federation of Pharmaceutical Industries Associations (EFPIA), is that public budgets remain under pressure . “We are not living in a ‘business as usual’ environment,” she said. “It’s not even only the healthcare budgets that are strained, it’s the member states’ budgets,” she said, adding that for industry the problem of investment was much larger than people realised. “You can’t do this in your garage and then scale it up with some investment – the odds are 10,000 to one,” she explained. “You start with 10,000 compounds, and you might come out with one – that is not a good story for an investor.”

According to **Yann Le Cam**, Chief Executive Officer of the European Organisation for Rare Diseases (Eurordis), the problem is also one of access to medicines. “What does it mean to invest in treatments and get them to market authorisation, with sophisticated work by industry and by regulators, if, at the end, it doesn’t get to the patient?” he said, pointing to reports of health services in Spain, the Netherlands and the United Kingdom rejecting new drugs to treat rare diseases.

In central and eastern Europe, there is also a problem of lack of funding for national health systems, with around six per cent of GDP spent on healthcare in many countries in the region, compared to more than ten per cent in France and Germany. For instance, in Poland, with the health system strapped for cash and staff, some therapies for multiple sclerosis are not reimbursed by the state.

INCENTIVES WORK

In 2000 the European Commission introduced incentives to encourage the development of new drugs to treat rare disease, including ten-year market exclusivity and access to scientific advice from the European Medicines Agency (EMA). “The incentives are there to actually stimulate innovation – I think they are working,” said Kim Stratton, Head of International Commercial at Shire. “It would be a shame to spend energy unpicking and reinventing something that’s not broken. What is broken is how to actually get these medicines to our patients,” she told the Friends of Europe event.

Stratton outlined how the lack of large datasets for orphan drugs, and the often limited knowledge of the diseases, can make it hard to demonstrate treatment value at time of launch following standard appraisal criteria. This leads to patients being denied access to innovative therapies, she said.“If we don’t fix this soon, that takes away the incentive to put more products into the pipeline.”

Presenting a new proposal, Stratton said, “Innovative patient access solutions such as our ‘immediate access’ model can enable timely patient access while providing flexibility to manage evidential and financial uncertainty when evaluating orphan medicines.”

The EU should do more to encourage 'soft incentives', Yann Le Cam added – including data collection, neonatal screening and training to help healthcare professionals prevent and diagnose rare diseases much earlier. He called for a structured dialogue between payers and industry. "Industry needs a return on investment and good rewards, in order to attract investment – they need predictability."

COLLABORATION IS KING

In 2013 the European Commission created an expert group on rare diseases. This group, which has met eight times, advises the bloc on anything from health technology assessments (HTAs) to cross-border genetic testing. But the real success story for the EU has been the European Reference Networks (ERNs), 24 centres of excellence which began work in March this year. Their purpose is to link up 900 specialist teams across Europe with people seeking treatment for rare diseases. "The ERNs are bringing the knowledge and the expertise to the patient rather than expecting the patient to go out and find that through the system," said Tamsin Rose, Senior Fellow at Friends of Europe, who moderated the debate.

Maurizio Scarpa, Director of the Horst Schmidt Klinik Institute for Rare Diseases and Coordinator of the European Reference Network for Hereditary Metabolic Diseases (MetabERN) said the networks have heralded a "revolution" in rare disease treatment that sees the patient as a whole person, not as a single specialty. "The big goal, which is to change the quality of life of patients, to change the natural history of diseases, to find new therapies, needs collaboration, needs effort, needs vision, needs mission and needs passion," he said. With the help of the ERNs, he is confident of reaching the goal of the International Rare Diseases Research Consortium (IRDiRC) to create 1,000 new drug therapies by 2027 (with the 2020 goal of 200 therapies already having been met).

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CALL TO ACTION

Panellists agreed that there was a unique opportunity to build on the success of the ERNs and rally people together. "If there's one area where it must be easy to show EU added value, and therefore the need for EU investment, it is rare diseases and ERNs," said Xavier Prats Monné. "Let's not make the mistake that was done with the Brexit debate, where all those who knew about the benefits of European cooperation were just too silent or, rather, were not shouting loud enough," he said.

For patients, what is needed is a series of concrete goals, said Yann Le Cam, including diagnosis within six months and adding ten years to life expectancy. "What is success is critical mass, so quantity means quality," he said, pointing to progress made for cystic fibrosis, Duchenne and haemophilia sufferers. "That's a nice goal to have, to help us focus things."

The ERNs can also be a gateway to help develop therapies for more frequent diseases, added Maurizio Scarpa. "Rare diseases, most of the time, can explain the pathogenesis of common diseases," he explained. "We need to have coordination in order not only to cure, but also to prevent diseases."

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European Commission Director-General for Health and Food Safety

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