NewStatesman

Making the UK a rare disease leader

Science, technologies and infrastructure

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World-class healthcare leaves no one behind

We must do more for the 3.5m people affected by rare diseases, writes Damien Bailly, head of UK-Ireland Cluster at Shire Pharmaceuticals Ltd

An estimated 3.5 million people in the United Kingdom are affected by a rare disease at some point in their life[1]. Approximately half of people affected by rare disease are children, and 30 per cent of children with a rare disease will not live to see their fifth birthday[2]. The facts relating to rare disease are eye opening, and the personal stories of the individuals impacted are even more compelling. The Rare Disease UK patient experience examples, available online, reveal stories of diagnoses which take many years, children living with chronic pain, and those coping with the mental health issues associated with rare disease[3].

The UK Strategy for Rare Diseases sets the ambition that "no one gets left behind because they have a rare disease". Shire was delighted to partner with the *New Statesman* to discuss how we meet this ambition, and how the UK might build on its strong foundations in clinical expertise to lead the world in the field of rare diseases.

The UK's clinical leadership can be seen by the instrumental role that UK clinicians have had in establishing and leading European Reference Networks. This, in turn, is the basis for much ongoing research and development in the UK, with Shire, for instance, conducting a quarter of its European clinical trials in the UK[4].

The UK has also established a world-class programme of genomics research, with the Secretary of State for Health, Matt Hancock, recently announcing an ambitious expansion of the 100,000 Genomes Project.

However, in order to transform the UK into a post-Brexit hub for global rare disease leadership, the UK must have excellence from bench to bedside, and at present, beyond near-unrivalled scientific leadership, UK rare disease patients face two significant challenges:

1. Patients still wait too long for a diagnosis. On average, patients wait 5.6 years to receive an accurate diagnosis, typically seeing seven or eight healthcare professionals[5].

2. Research has shown that of the 143 licensed rare disease medicines in Europe, just 68 have been centrally approved for use by the NHS in England[6]. This is mostly because of an outdated method of assessing the cost-effectiveness of these treatments, which other countries have moved away from[7].

Both of these challenges can and must be addressed through collaboration between NHS England, NICE, other decision-makers, patient representatives, and the life sciences sector. In doing so, we should set a new ambition for rare disease; for the UK to be the unparalleled global rare disease leader, from bench to bedside, with enhanced clinical leadership, and with patients having a right to access all treatments which their doctors think they need.

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Real-world experience is the best research

The UK can be a test-bed for innovation, writes Stephen Twigg MP, co-chair of the all-party parliamentary group for rare genetic and undiagnosed conditions

During my time as co-chair of the all-party parliamentary group (APPG) for rare, genetic and undiagnosed conditions, an issue that has been consistently raised by patients, clinicians, and pharmaceutical companies is the challenge they face attempting to navigate NHS England's and NICE's pathways to approve funding for a new rare disease treatment.

There are many routes through which a medicine can be approved in England. NICE is increasingly using the Single Technology Appraisal route alongside the Highly Specialised Technology programme to evaluate rare disease medicines, and of course there is the Cancer Drugs Fund for treatments for rare cancers.

NHS England makes decisions on routine access differently, with a competitive funding round, where a treatment for epilepsy in children might compete with a treatment for a debilitating condition affecting adults – a very difficult trade-off.

We have to remind ourselves that this is just the decision-making process for rare disease treatments in England. At a UK level, there is further diversity as Wales, Scotland and Northern Ireland operate their own approaches, or borrow from each other. This complex, overlapping and sometimes contradictory environment is not a signal that the UK embraces innovation in biosciences. Rather, it is a worrying deterrent, at a time when we should be working to facilitate the uptake of innovation, ensuring an efficient and navigable route from research through to the NHS for safe and effective treatments.

We need the cycle of drug development to keep turning to continue innovative treatments in the NHS, to maintain standards of care and to ensure the NHS stays a relevant research environment, so that we can develop new cutting edge treatments in this country. Without the cycle of access, treatment and research turning, the UK risks disinvestment from innovative companies.

The APPG has asked Genetic Alliance UK to deliver a new vision for access to rare disease medicines. If we were starting from scratch, we certainly would not have what we have now, and I am not convinced that we can maximise improvements by continuing our current approach of making small adjustments to the current systems.

Genetic Alliance UK's challenge will be to find a model that increases and hastens access to rare disease medicines while ensuring there is continued confidence in the value of the treatments that we deliver through the NHS.

A key factor in delivering this new vision will be real-world evidence. Instead of trying to make an assessment of the cost to benefit ratio of a treatment based on a small population clinical trial, this empowers decision-makers to examine and assess the treatment in the NHS itself.

Our collective challenge is to deliver a future where the UK is the nation of choice for medical research and the launch of innovative treatments. What we need now is a clear line of sight and the vision to embrace this challenge and drive transformative change.

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Shire and the *New Statesman* brought industry experts and policymakers together to discuss how to harness the UK's life sciences excellence in the fight against rare diseases

How genomics is transforming rare disease treatments

While their incidence may be less common than other conditions, rare diseases affect around 3.5 million[1] people in the United Kingdom, and a minority group which still represents millions of people is not one that can be ignored. An estimated 30 per cent of children born with a rare disease do not live beyond their fifth birthday[2].

Chronic conditions can impact significantly on people's quality of life, in many cases causing disability, and the National Health Service, seemingly under perennial pressure to do more with less, must not be caught out. The UK's health system, then, should be proactive, rather than reactive, and rare disease research and treatments deserve to be propelled into mainstream discourse. Earlier this year, global biotechnology company Shire and the *New Statesman* gathered industry experts and policymakers for an event in Westminster, to discuss the science, technologies and infrastructure needed to make the UK a world-leader in treating rare diseases.

Vicky Ford MP, member of the science and technology select committee, noted in her opening address the success being spearheaded by the UK in the field of genomics. Name-checking the 100,000 Genomes Project[3] – managed by Genomics England, a company owned by the Department of Health and Social Care – Ford said that the UK must keep hold of its "cutting-edge" and "ground-breaking" research, while ensuring that "life sciences formed a key part of any industrial strategy". The MP for Chelmsford said that the UK should aim to keep "[as much of the] supply chain in the UK as possible." She explained: "The government wants the UK to be a world-leader in life sciences. That means being able to support clinical trials here, and being able to market new medicines here."

The 100,000 Genomes Project, whereby the genes of NHS patients suffering with rare diseases are sequenced and analysed across 13 specialist NHS Genomics Medicine Centres (GMCs), was launched in 2013. Ford said that data science had a "crucial role to play in shaping the

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Data can inform earlier diagnoses

future of healthcare. The GMCs have a chance to work with these fantastic pools of information, and by having these pools it means that the NHS can have a point of reference for rare diseases that didn't exist before."

For Paul Pambakian, associate director of government relations at Shire, one of the crucial challenges to overcome in treating rare diseases, was the speed and accuracy of the diagnostic processes. He said: "Part of the problem for people [who suffer from rare diseases] is that they don't actually know what they've got for several years. You've got instances where a patient can see, on average, five doctors, and receive three or four misdiagnoses. Imagine the stress that a patient and their family goes through."

Dr Richard Scott, clinical lead for rare diseases at Genomics England, agreed that having access to long-term data banks, providing a "library" of conditions, could help doctors to reach diagnoses quicker. He said: "Genomics has been a real game-changer in that regard, and on top of diagnoses, the opportunity to monitor genetic conditions, shows how data is being used to manage people's long-term health."

Using risk and probability assessments, Scott explained, the 100,000 Genomes Project was helping to create insights into early detection, and hereditary conditions. "This can give families a peace of mind. It's an extra reassurance. Some of these families are terrified to have another child, because they worry about whether a disease could be passed on. In some cases, finding the cause allows us to reassure them that the chance of having another child with the condition is low. In others, if we can predict that a child has a high risk of having [a rare disease] early enough, then it may be possible for us to institute some changes, to help manage that."

Pambakian suggested, meanwhile, that other European countries had perhaps more welcoming assessments frameworks when it came to funding new medicines for rare diseases. Ironically, Pambakian said, "UK universities are world-class", but "those innovations are too often not funded or are too slow out of the blocks". A report from the Office of Health Economics (OHE) last year[4] found that orphan medicinal products (OMPs) – medicines used to treat rare diseases – were less readily available in the UK.

The European Commission's Orphan Medicinal Products Regulation, set up in 2000, intended to incentivise the development and

Patient groups offer emotional support

marketing of new treatments for rare diseases. Marketing authorisation granted to OMPs, however, is only one part of their journey; patients have access to health technology assessment (HTA) decisions are implemented by national health systems. Nodding to the OHE's report[4], Pambakian said: "There were 143 orphan medicines with a central license at the time of this study… 133 of them have been made available routinely in Germany, 116 are available in France, and the figure for England is just 68. That is really disappointing, and presents a real challenge for the whole rare disease community."

While data science and genomics represent the advanced aspect of early detection of rare diseases, Dr Jayne Spink, chief executive at Genetic Alliance UK, said that at a basic level the UK should "reconsider its policy on newborn screenings". Currently, the NHS employs a "blood spot" test, more commonly known as the "heel prick" test[5]. Newborn babies have their heels pricked and a small sample of their blood is checked for nine rare conditions. "If you look at Italy, which screens for 40 [conditions]," Spink said, "or Austria which screens for 28, then you have to say that the UK is lagging here. Screening at birth gives a head start for treatments, and the longer someone goes undiagnosed, the more difficult it gets [to treat them]."

Social and emotional support, according to the chief executive of Genetic Disorders UK Caroline Harding, is as important as the science or investment involved in the fight against rare diseases. Genetic Disorders UK is a registered charity with a vision to improve the lives of individuals and families affected by rare hereditary conditions, by offering online information, bonding and fundraising events, and a phone-in helpline. As these conditions can be "emotionally exhausting", Harding said that patient-led organisations were a good way of coping, by offering a support network through shared experiences.

These groups, she said, can "help the exacerbated mum or dad… they will feel better if they know that there are other people going through the same thing, and they can get advice." She added: "Patient registries and data are one thing, but having a group to be able to be a part of… a centralised space for patients and the families of patients, that's a lot more personal."

The success of any initiative, Shire's director of market access and external affairs Berkeley Greenwood argued, hinges heavily on its representation in government. The remits for the UK's health ministers, he said, were too broad. For something as specific as rare disease strategy, Greenwood suggested, then, "a specialist minister, exactly for them." He said: "In the UK, we have a health secretary, we have a minister of state for health and we have parliamentary undersecretaries. But these roles have to cover a lot. Rare diseases are a full-time job, or at least they should be, for someone.

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We need to make that happen."

There has been an "absence of joined up thinking at policy-level," Greenwood said, "which doesn't mirror the roles in industry." He continued: "In NHS England, you've got people who decide whether a particular product should be used, you've got officials who update the rare disease strategy and gauge whether or not it's being achieved. But what you don't have is a person in parliament, who is accountable, specifically, to oversee rare disease treatments and strategy. An overarching political champion, if you like, could oversee the policy aspect of this issue… they could look at skills, funding, jobs, specifically relating to rare diseases. The other ministerial roles in health are having to contend with lots of other things."

"A political champion is needed"

Rare diseases, the round table concurred, are not rare enough. That millions of people suffer from conditions for which there is relatively little treatment is an aberration that the NHS and the UK's life sciences industry must work even harder to overcome. Making the most of innovative research, the key to fighting rare diseases, then, lies in early detection, swifter diagnoses and more coordinated treatment. After all, a minority that still accounts for millions of people is one that needs support.

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Rare diseases by the numbers

Genetic conditions represent some of the most complex and challenging issues facing the NHS

3.5m

The number of people in the UK who will have a rare disease at some point in their life[1]

5.6 years

The average wait for people with rare diseases before receiving a correct diagnosis[2]

30

The percentage of children with a rare disease who will not live to see their fifth birthday[3]

5m

The number of genomes that will be sequenced in the UK in the next five years[4]

68 of 143

The number of rare disease medicines to have been centrally approved for use by the NHS in England[5]

1 in 17

The number of people in the UK who will have a rare disease at some point in their life[1]

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