Cynulliad Cenedlaethol Cymru | National Assembly for Wales
Y Pwyllgor Materion Allanol a Deddfwriaeth Ychwanegol | External Affairs and Additional Legislation Committee
Y goblygiadau i Gymru wrth i Brydain adael yr Undeb Ewropeaidd | Implications for Wales of Britain exiting the European Union
IOB 41
Ymateb gan Genetic Alliance UK
Evidence from Genetic Alliance UK

Introduction

1. Genetic Alliance UK is the national charity working to improve the lives of patients and families affected by all types of genetic conditions. We are an alliance of over 180 patient organisations. Our aim is to ensure that high quality services, information and support are provided to all who need them. We actively support research and innovation across the field of genetic medicine.

2. For patients affected by rare, genetic and undiagnosed conditions there are numerous advantages to the UK, including Wales, being closely aligned with other European Member States. The UK Strategy for Rare Diseases, Orphan Medicinal Products Regulation, European Reference Networks and the International Rare Disease Research Collaboration are only a few examples of the added value that has been generated through transnational collaboration made possible as a consequence of our membership of the EU.

3. Genetic Alliance UK was unequivocal in advocating its support to remain in the EU. Being part of a community of 500 million has pushed rare and genetic diseases to prominence in the research and health care agenda. We were therefore disappointed by the referendum result. Our response outlines the potential implications of Brexit for Wales in relation to science, research and innovation in the field of rare diseases.

Funding

4. Though the majority of international research collaboration occurs outside the context of EU specific structures, it is important to recognise those EU initiatives that do facilitate cross border collaboration. One of these is research funding. A significant source of funds for health research in the rare disease field comes from European sources such as the Innovative Medicines Initiative and Horizon 2020. These are not solely a source of funding, but also a significant driver in the formation of partnerships across the EU.

5. Horizon 2020 has provided numerous opportunities for health, care and third sector organisations in Wales to access significant European and other international funding to support policy development and implementation, research and innovation, leading to better health and wellbeing and enhanced service sustainability. Wales was in a position to increase its involvement in EU funding due to the close fit and coherence between policies, providing a clear opportunity to exploit EU funding streams to add value in Wales.

6. The European Patients' Academy for Therapeutic Innovation (EUPATI) is working to train patient advocates across Europe (many of them based in the UK) to enable expert patient voice to be incorporated in decisions along the treatment development pathway. The Accelerated Access Review's interim report has recently identified greater patient voice along the innovation pathway as a key aim for the UK. EUPATI is funded by the Innovative Medicines

Initiative (IMI) which was part of the seventh Framework Programme for Research and Technological Development of the European Union (FP7) and will continue as part of the Horizon 2020 (the successor to FP7) programme.

7. We are performing psycho-social research, gathering parents' perspectives, as part of a clinical trial gathering evidence for the repurposing of an off-patent medicine for congenital adrenal hyperplasia as part of the Treating Adrenal Insufficiency in Neonates (TAIN) project. TAIN is funded by FP7.

8. EU funding programmes are necessary to facilitate research in rare diseases. Continued access to EU funding programmes, including Horizon 2020 and its successor, should be sought during negotiations.

Collaboration and Regulation

The importance of collaboration in the rare disease sector

9. Many rare diseases are severe and life-limiting. For individuals or families affected by most rare diseases, the day-to-day challenges of managing a severe condition are made worse by the absence of an effective treatment or cure. These patients look to research as the source of new therapies to address their unmet health need. In order for progress to be made, patients recognise that the rarity of their conditions means that research relies on the effective sharing and use of their medical data, nationally and internationally.

10. Unlike common conditions, patient populations of individual rare diseases are low, and sometimes very low. There may be too few patients with any particular rare disease in a single Member State to be able to advance treatment and research. National and international research collaborations are invaluable: by collating and analysing large amounts of patient data from across the world is it possible to make meaningful progress with understanding a condition or the effectiveness of a new treatment.

11. Regulations within the European Union provide a framework for this collaboration to take place. The Clinical Trial Regulation and the Data Protection Directive (soon to be updated by the incoming Data Protection Regulation) are major examples relevant to our community.

12. The Clinical Trial Regulation (which is still being implemented) represents a major improvement on the previous Clinical Trials Directive, improving harmonisation and reducing a great deal of regulatory burden restricting the scope to deliver low volume international multi-centre clinical trials.

13. Sponsors will have to consider the UK's relationship with the EU when considering the planning of trials. This, in addition to the lack of up to date comparator treatment use in the NHS, and the decreasing possibility of reimbursement for the eventual product of innovation in the UK are already cited as factors counting against the UK as a clinical trial host.

14. Clinical trials are an important source of treatments for Wales's rare disease patient population. There are numerous examples of patients that have accessed life-saving treatments through this route. These include the newest innovations in therapies for tuberous sclerosis and metabolic disease. It is therefore important that patients are not disadvantaged in terms of accessing clinical trials and treatments as a result of the UK leaving the EU.

15. In March 2016, the Welsh Government published a Statement of Intent outlining the key principles that would underpin the development of a Genomics for Precision Medicine Strategy. In the same year Wales joined the 100,000 genomes project and will be establishing a strategic

partnership with Genomics England, having successfully obtained £1m funding from the Medical Research Council and £2.4m funding from the Welsh Government. The funding will increase genomics capacity in Wales and lead to the establishment of a Genomic Medicine Centre in Cardiff. It will provide an opportunity for Welsh patients with rare diseases to take part in the 100,000 genomes research programme, with the potential to receive more accurate and quicker diagnoses. The UK is a world leader in genome sequencing research. The UK's major peer in Europe in this field is the Netherlands. The initiatives in these two countries benefit greatly from sharing information internationally, as it is not possible to validate a genetic sequence for a suspected impact on health without further examples of its occurrence. The UK, including Wales, must continue to have access to collaborative opportunities.

16. The sheer numbers of individual rare diseases mean that experts cannot be in every Member State and travel may be necessary for patients to access effective treatment. Patient communities may be too small in individual Member States, and benefit from making contact and collaborating across borders. During negotiations consideration must be given to ensuring health professionals and patients continue to be able to travel and work across the EU.

The UK Strategy for Rare Diseases and the Welsh Rare Disease Implementation Plan

17. In response to the European Commission Communication on Rare Diseases: Europe's Challenges, and the following European Council's recommendation on an action in the field of rare diseases, the ministers for health from all four nations of the UK published the UK Strategy for Rare Diseases in November 2013. It is the first time since the establishment of the NHS that patients and families affected by rare conditions have a clear and strong commitment from Government that their healthcare needs will be met. It is a shared vision for improving the lives of all those affected by rare conditions to ensure "no one gets left behind just because they have a rare disease" (UK Government, 2013).

18. The European Commission's and the European Council's work in this area has raised the profile of rare disease within the UK, to the benefit of the whole rare disease community, which includes patients, families, carers, clinicians, researchers, industry, and healthcare commissioners.

19. Effective implementation of the UK Strategy for Rare Diseases will improve the diagnosis and treatment of all patients affected by rare conditions. It will help to ensure that patients who are affected by rare conditions receive the care and treatment they require.

20. The strategy recognises, that through specialist clinical centres, the "UK wants to support the sharing of information, data, knowledge and best practice in treatment nationally, across Europe and further afield". And in order to deliver this and improve the healthcare that patients receive, the strategy notes that "Centres should have connections to others across the UK and in Europe" (UK Government, 2013).

21. In February 2014, the Welsh Government published The Welsh Rare Disease Implementation Plan. The Welsh Plan underpins delivery of the UK Strategy, which in turn supports the drive in Europe to improve rare disease services.

22. The Welsh Plan acknowledges that, where relevant, there must be inter-country and EU wide collaboration for opportunities for patients to become involved in rare disease research. The Welsh Plan relies on patients being able to access and participate in high quality research outside of Wales as it is only possible to sustain high quality research in a few disease areas. For example, the Welsh Congenital Anomaly Register and Information Service (CARIS) is a member of EUROCAT, the European network of congenital anomaly registers.

23. In the interests of patients with rare diseases, it is necessary for the Welsh Government to maintain its commitment to the UK Strategy for Rare Disease and its commitment to inter-country and EU wide collaboration.

Medicines

24. The European Union's regulation of medicine in Europe, overseen by the European Medicines Agency (EMA – based in London), creates the largest single regulatory environment for developed nations' populations, with a population of 500 million. This infrastructure is attractive to pharmaceutical companies wishing to bring medicines to a significant market. The European Union can leverage this critical mass to provide incentives for the development of orphan medicines and for advanced therapy medicinal products.

25. States outside of the EU (such as Norway and Iceland) may still benefit from the EMA's regulatory environment, but they cannot have any influence in decisions made by EMA.

26. The UK's participation in the EU's centralised procedure for the evaluation of medicines gives a benefit at both ends of the product development pathway. For patients in the UK, we are part of the same market, which is usually either first or second (after USA) on the list of markets that an innovator would seek to launch their products in.

27. The orphan medicinal product regulation provides incentives and support for the development of treatments with indications with a prevalence of fewer than 1 in 2,000. To date this has supported the development of 114 treatments (European Commission, 2016) for patients affected by rare diseases.

28. It is vital for patients that there is continued alignment with EU regulatory frameworks for medicines and medical devices. This will ensure patients continue to have timely access to new treatments. Continued alignment will also help to support the life sciences industry in Wales.

Organ, blood, tissues and cell donation

29. The UK's membership of the EU Tissue and Cells regulatory system increases the potential pool of donors for haemopoietic stem cell transplantation (HSCT) – one of very few effective treatments for genetic conditions – for UK patients and is therefore a significant benefit to our patient community. European regulation allows cross-border transfer of cells for clinical use, which is highly beneficial to the search for a matching donor. It is vital for patients that this collaboration continues.

European Reference Networks and the Expert Group on Rare Diseases

30. The EU directive on cross-border healthcare (European Union, 2011) provides incentives to Member States to develop European Reference Networks (ERN). ERNs seek to identify already established centres of expertise and to encourage voluntary participation in a Europe wide collaboration with other centres of expertise. ERNs for rare diseases will serve as research and knowledge centres, updating and contributing to the latest scientific findings, treating patients from other Member States and ensuring the availability of subsequent treatment facilities where necessary.

31. ERNs will be ideally placed to facilitate improvements in access to diagnosis and delivery of high-quality, accessible and cost-effective healthcare especially in the case of patients that require a particular concentration of expertise or resources including patients affected by rare conditions.

32. The UK is well represented on the Expert Group on Rare Diseases which advises the EC on issues relating to rare diseases. This membership demonstrates the expertise within the UK on

rare disease issues. Involvement at this level furthers relationships in the research and treatment spheres, where the UK is one of a few nations taking the lead in innovation in this area.

33. Patients in Wales must continue to have access to the most up-to-date care and advice. During negotiations the position of the UK in relation to European Reference Networks must be established.

Conclusion

34. Exiting the EU has the potential to significantly and negatively affect rare disease patients in Wales. At present there are a number of advantages to the UK being closely aligned with other European Member States. To maintain these advantages, the Welsh Government must consider the following in their negotiations:

- Ensuring that there is continued access to EU funding programmes including Horizon 2020 and Framework Programme 9.
- Ensuring that patients continue to be able to access healthcare across the EU.
- Maintaining a commitment to inter-country and EU-wide collaboration.
- Continued alignment with EU regulatory frameworks for medicines and medical devices will ensure patients continue to have timely access to new treatments and support the life sciences industry in Wales.

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