European and External Relations Committee

Inquiry into EU reform and the EU referendum: implications for Scotland

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 being closely aligned with other European Member States. Our response outlines the potential implications for Scotland in relation to science, research and innovation in the field of rare diseases.

3. The Scottish Government and Genetic Alliance UK are official partners of the 8th European Conference on Rare Diseases and Orphan Products (ECRD) which will take place in Edinburgh in May 2016. The ECRD is a forum across all rare diseases, all European counties, bringing together all stakeholders. The conference covers research, development of new treatments, healthcare, social care, information, public health and support at European, International, national and regional level.

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 Seventh Framework Programme 2007-2013 (FP7), 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. Genetic Alliance UK receives a significant portion of funding from European Union initiatives. In most cases, this is because the type of activity we are involved in can only happen at a continental level.

6. The European Patients’ Academy for Therapeutic Innovation (EUPATI) is working to train a 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 FP7 and will continue as part of the Horizon 2020 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 the FP7 programme.

8. We are also partners in two other currently active projects, and have been part of four others in the past five years. These projects have contributed to 7.1% of our income over this time.

9. We have contributed to Healthcare Improvement Scotland’s work on the development of a collection of good quality clinical guidelines and associated research recommendations for RARE-Bestpractice, which was initiated in 2013 with funding from the FP7 Programme. This project is intended to support access to diagnosis and provision of high quality healthcare for patients with rare diseases in line with EU Directive 2011/24/EU on the application of patient’s rights in cross border healthcare.

**Collaboration and Regulation**

**The importance of collaboration in the rare disease sector**

10. 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.

11. 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.

12. 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.

13. 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. It would be disingenuous to argue that leaving the EU would rule the UK out of participation in clinical trials for rare diseases, but it would be another negative aspect that sponsors would have to consider in the planning of trials. 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 the UK’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 muscular dystrophy and metabolic disease.

15. The UK is a world leader in genome sequencing research and in 2015 the Scottish Genomes Partnership was announced, representing an investment of £15 million in gene sequencing technology in Scotland. 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.

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.

The UK Strategy for Rare Diseases and The Implementation Plan for Rare Diseases in Scotland

17. In response to the EC Communication on Rare Diseases: Europe’s Challenges 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”.4

18. The EC Recommendation 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”.1

21. In July 2014, the Scottish Government published ‘It’s not rare to have a rare disease – the Implementation Plan for Rare Diseases in Scotland”. The Scottish Plan

underpins delivery of the UK Strategy, which in turn supports the drive in Europe to improve rare disease services.

22. The Scottish Plan acknowledges that, where relevant, there is existing inter-country and EU wide collaboration. The Scottish Plan envisages that more collaboration of this nature will develop as rare disease work progresses. For example, the Scottish Health Technologies Group (SHTG) provides clinical and cost-effectiveness advice on health related technologies to support planning and decision making in Scottish NHS Boards and also provides early information on health technologies in development. The SHTG currently links to European and international health technology networks.

Medicines

23. 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.

24. 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.

25. 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 innovators in the UK, we are part of a large market with a harmonised regulatory approach, that as we will argue in paragraphs 29-32 (Scientific Advice), we are able to influence. 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.

26. 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 for patients affected by rare diseases.

Organ, blood, tissues and cell donation

27. 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.

\[2\text{ ec.europa.eu/health/human-use/orphan-medicines/index_en.htm}\]
European Reference Networks and the Expert Group on Rare Diseases

28. The EU directive on cross-border healthcare\(^ 3 \) 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.

29. 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.

30. 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.

Conclusion

31. Some may argue that the points raised so far in this response would be, to some degree, possible without membership of the EU. We believe it would be to a small degree, but must concede this point to an extent. However, we should acknowledge that we are referring to an EU that the UK has been a member of for 42 years. The UK has had an enormous impact upon the EU’s outlook with respect to science, innovation and research.

32. The UK’s third sector’s influence on the Clinical Trial Regulation and the Data Protection Regulation (currently ongoing) has been seminal and powerful. Without our voices, the EU would undoubtedly be a worse place to do research, and cross border collaboration would certainly be much more difficult. The Medicines and Healthcare Regulatory Authority (MHRA) is a key opinion leader at the EMA and has played a significant role in creating the regulatory environment that we have in the EU today.

33. Leaving the EU would almost completely end our influence over policy development in the EU. The large consortium of countries on our doorstep with harmonised approaches to research and medicine regulation would begin to change significantly. The UK has a world leading approach to the regulation of innovative approaches in research and healthcare; this voice is crucial in the balance of attitudes on our continent.

34. An EU without the UK would eventually become an EU with which collaboration would have little value. Consequently, the argument that the UK can continue to benefit from the EU from outside will, at least in a health research and medical context, be shown to have been incorrect.