**January 2017**

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# Implementation of the UK Strategy for Rare Diseases in England: Response from PHG Foundation

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The PHG Foundation is non-profit health policy think tank. We work to achieve the prompt, effective and responsible application of biomedical and digital technologies within health systems

The PHG Foundation is a non-profit policy organisation focused on the implementation of genomic medicine and other new biomedical technologies for the improvement of health and healthcare. Active since 1997, we have been closely involved in policies through which the practical implementation of rapidly evolving new genetic technologies can take place. Our Director, Dr Hilary Burton led the subgroup on research for the original UK Rare Disease Strategy and has been involved in the Rare Disease Forum since that time.

Advances in genetic technologies are enabling more rapid and detailed diagnoses and recently new treatments based on better understanding of the molecular pathology of disease have become available. Although in the future the relevance of genomics in common complex disease may become clear, many of the opportunities for immediate application of genetic technologies thus far have been for rare diseases. If these technologies are properly applied patients with poorly differentiated sets of clinical symptoms or rare combinations that could not easily be recognised by the non-specialist can receive a more rapid diagnosis and, thereafter, treatment and management by experts in that rare disease.

Such management will clearly include an important element of information giving, tailored clinical management and often lifelong support which may require dealing with chronic multisystem disabilities and provision of advice to family members who may also have inherited the condition or be at risk of passing it on.

The UK Strategy for Rare Disease published in November 2013 recognised that the phenomenon of rare disease presents a set of challenges that are not easily met within a health system. The needs of patients and their families could not be met by standard services. The Strategy made 51 commitments, and, crucially, recognised that each country would need to develop its own implementation plan in order to make changes on the ground that would help families. It is lamentable that 3 years later there is no implementation plan in England: the organisations with responsibilities for patients and healthcare (the Department of Health (DH), NHS England (NHSE)) have provided neither leadership nor coordination for the different elements of the strategy. These omissions are letting down patients and families, as well as failing to capitalise on the opportunities for better and more cost-effective NHS management offered by accurate diagnostic testing capacity such as that provided within the context of the 100,000 Genomes Project. The beneficial impact of improved testing technologies cannot be fully realised without implementation of the rare disease strategy.

This approach singularly fails to recognise the core message of the rare disease strategy, which is that 'standard' NHS systems to meet the needs of more common diseases and health challenges cannot meet the needs of this patient group, and that specific focused initiatives are required to ensure they are not subject to iniquitously poor care.

We noted in their response to the APPG Inquiry that DH explicitly ruled out responsibility for implementation and accepted responsibility only for 'facilitating its various arm's length bodies to implement it'. NHS England has, at least, acknowledged the Strategy by issuing its Statement of Intent and its work, for example with UKGTN to make high quality diagnostics more accessible through agreed patient pathways or to work with NICE in technology appraisal, is to be commended. However, the NHSE responses to many of the other commitments is lacking in actions targeted specifically at addressing the needs of rare disease patients and instead implies that the commitments made in the strategy can be met without any additional effort beyond that already being expended on NHS-wide programmes of improvement in care quality and efficiency.

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Moreover, the NHSE Statement of Intent relies heavily on the Rare Disease Advisory Group, which provides advice on the development of services and the newly constituted UK Rare Disease Stakeholder Forum although both of these groups have a UK wide remit.

As part of the Strategy, Public Health England has also played an important role in developing an expanded congenital anomaly and rare disease registry for the whole of England, which will contribute to disease surveillance and research. The 100,000 Genomes Project is also recognised as being a vital component of the strategy. Whilst this initiative should, in the longer term contribute to research into genomics and disease, the extent to which this Project will ultimately contribute to the everyday needs of a large number of patients for quick and accurate diagnosis and personalised care, will depend on how well NHS England is able to embed genomic testing within a wide range of clinical pathways and its ability to address many outstanding challenges. These include ensuring sustainable and comprehensive funding for genetic testing and the development of a new cohort of doctors and other health professionals (such as biomedical informaticians and scientists) who together are able to identify patients with potential rare disease, order, analyse and interpret test data, and take appropriate clinical action.

Overall, whilst it is undeniable that existing projects and policy developments may benefit patients with rare disease in England, in our view the fact that there is no implementation plan and an absence of overall leadership or coordination for its delivery represents a significant gap. As long as no single organisation is tasked with the responsibility for doing this, an important opportunity is being missed to make significant and systemic improvements to the care in the NHS care of those with rare diseases.

We feel that, in parallel with the assignment of responsibilities in the devolved nations, responsibility for implementation in England should lie with the DH. In saying so we recognise that DH capacity has been considerably affected by recent staff reductions in the various relevant departments. However, we believe that this should be explicitly recognised; if necessary a more limited scope or derogation of authority either for the development of the implementation plan or for some elements of it should be agreed with other organisations.

To do less than this will simply lead to frustration, disappointment and loss to the patients so closely concerned, to the health systems trying to provide high quality care and to the country as a whole as it seeks to lead the world in genomic medicine.