Genetic Alliance UK
Supporting. Campaigning. Uniting.

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Committee Clerk
Health and Social Care Committee
National Assembly for Wales
Cardiff Bay
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Dear Madam,

Health and Social Care Committee Inquiry: Access to medical technologies in Wales - Scope

Genetic Alliance UK is the national charity of over 150 patient organisations, supporting all those affected by genetic conditions. We aim to improve the lives of people affected by genetic conditions by ensuring that high quality services and information are available to all who need them. We welcome this opportunity to comment on the scope of the inquiry.

Introduction

There are many thousands of genetic conditions which affect patients and families in Wales. The number of cures and effective treatments for genetic conditions is much lower, and those that do exist, are in the main risky or expensive or both. The vast majority of patients with genetic conditions are left with palliation and mitigation, to limit the effect of the condition as much as possible to raise the quality and quantity of their lives as much as possible. There is an enormous burden of unmet need in the community of those affected by genetic conditions.

Genetic Alliance UK operates through project and policy work. One of our projects is SWAN UK. SWAN UK represents children with Syndromes Without A Name, i.e. undiagnosed children. It is estimated that around half of all children who attend a genetics clinic in Wales do not get a diagnosis for their condition. They are in the main affected by novel genetic mutations or chromosome rearrangements. Access to social care, education and healthcare is made significantly more difficult for these children and their families because of the lack of a diagnosis. They do not have a name of a condition to give their local authority, and lack of information about the prognosis of their child makes planning of services difficult.

Rare Disease UK (RDUK) is another project of Genetic Alliance UK. RDUK is a multi-stakeholder initiative which campaigns for the Health Departments across the UK to develop a strategy for rare diseases to ensure that patients and families living with these conditions have equitable access to effective services.

Our contribution to the scope

Genetic testing is an important component of the health service for those affected by genetic conditions. Genetic testing confirms the molecular cause of the condition affecting a patient. This allows the patient's clinician(s) a greater degree of understanding regarding the patient's prognosis, and influences treatment choice and therapeutic planning. Genetic counselling following a genetic test result can inform patients and their family members of their risks regarding the condition and can

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inform family planning. For those affected by conditions without a cure or a treatment, genetic tests are one of the very few innovations the health service has which allows them to take control of the way the condition affects them and their family.

We therefore strongly support the inclusion of genetic testing in the scope of this inquiry.

Our work with RDUK has shown the value of diagnosis, and the difficulties associated with accessing one. Patients and their families can be bounced around the NHS in a rudderless search for some expert knowledge which might be able to give them an indication of what they can expect in their future or where they can access answers. Rare Disease UK's Experiences of Rare Diseases: An Insight from Patients and Families1 found that of more than 400 patients, more than half have to wait more than a year for a diagnosis, and a fifth had to wait more than five years. A fifth of the patients surveyed saw more than five doctors before they were diagnosed.

Genetic testing technology is improving quickly. The costs associated with sequencing a patient's genetic information is dropping all the time. Advances such as Next Generation Sequencing put us on the threshold of great changes in the way that genetic testing is used by health service providers. The pace of development allows more testing to be done concurrently and at a faster rate, improving the rate and breadth of the search for a diagnosis.

Our work with SWAN UK has shown the need for further research and development in our understanding of genetic disease. Many of the members of SWAN UK are enrolled in research which aims to associate the symptoms children affected by a syndrome without a name are affected by with changes in their genetic code. This work has the potential to discover many more genetic conditions and bring the benefits associated with a diagnosis to families affected by these conditions. This research utilises the new technologies being developed to read our genetic information faster and more cheaply.

The technology that is being utilised by these research projects, and which is bringing greater potential for a more broad and more rapid search for patients with a rare disease comes at a price. Infrastructure investment will be necessary to ensure the ability of NHS Wales to keep pace with these developments and to participate in this kind of ground-breaking research.

We therefore strongly support the inclusion of capital investment in genetic laboratory infrastructure in the scope of this inquiry.

One of the most promising areas of development in the field of medicines currently is stratified medicine (personalised medicine). These are medicines which are best suited to a subset of the population defined by their expression or lack of expression of a biomarker. Many of these biomarkers are genetic. Before the medicine in question can be prescribed, a test using a companion diagnostic must be carried out for the relevant biomarker.

This method of delivering medicine ensures that patients do not take drugs that do not work for them, and therefore avoid the risk of side-effects when they are not necessary; and avoids the commissioning of drugs which will not work.

While we accept that medicines are outside of the scope of this inquiry, we believe that access to the companion diagnostics that make stratified medicines so powerful will become a pivotal issue in the future.

We therefore strongly support the inclusion of companion diagnostics for stratified medicine in the scope of this inquiry.

We are happy to comment further on the points outlined in this letter, and look forward to participating in this inquiry.

Yours faithfully,

Alastair Kent OBE
Director