Genetic Alliance UK
Supporting. Campaigning. Uniting.

National Assembly for Wales

Health and Social Care Committee

Access to medical technologies in Wales

Evidence from Genetic Alliance UK – MT 21

# Consultation response

## Inquiry into access to medical technologies in Wales

Response from Genetic Alliance UK, 18th October 2013

### Introduction

1. Genetic Alliance UK is the national charity 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 is available to all who need them. Our membership represents more than 160 voluntary organisations working for a wide range of conditions, many of which pose complex health and social care needs.

2. Genetic Alliance UK operates through project and policy work. One of our projects, Syndromes Without A Name (SWAN UK) supports families of children with undiagnosed genetic conditions. It is estimated that around half of all children who attend genetics clinics in Wales do not get a diagnosis for their condition – they may be affected by novel genetic mutations or chromosome rearrangements. Due to lack of a diagnosis, many families experience difficulties in accessing help and support from various services including health, education and social services.

3. In 2008 Genetic Alliance UK launched Rare Disease UK (RDUK), the national multi-stakeholder alliance for people with rare diseases and all who support them. RDUK is campaigning for a National Strategy for Rare Diseases in the UK, to ensure that patients and families living with rare conditions have equitable access to effective services.

4. We welcome the opportunity to respond to this inquiry.

### The value of new or alternative medical technologies

5. There are many thousands of genetic conditions which affect patients and families in Wales, however, for those conditions, there are many fewer that have effective cures or treatments. Many of those that do exist are generally risky, expensive and/or bring significant adverse effects. 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.

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6. Early diagnosis of genetic conditions through utilising new, advancing medical technologies can provide significant benefits, both practically and psychologically for parents of children who have a genetic condition. A diagnosis offers the patients' clinician and family a greater understanding of health difficulties and a clear genetic diagnosis can enable more focussed treatment choices and therapeutic planning and better access to information about their prognosis and future needs.

7. A technology that has been revolutionary in providing many more children with an early diagnosis is microarray comparative genomic hybridization (array-CGH). This advanced technique for genetic testing detects copy number changes in a person's chromosomes. This means it looks for deletions or duplications in their DNA that would not be identified using conventional microscopy-based chromosome analysis (karyotyping).

8. The improvement in diagnostic power that is available using array-CGH over karyotyping is clear, and valuable to families. Many children with developmental delay who have had a 'normal' result from a microscopy-based chromosome analysis in the past have, after consultation with their parents, been retested using genomic micro array analysis. A number of these children have been found to have a microdeletion or microduplication.
9. The diagnoses that array-CGH can provide allows parents to better plan their child's life. The diagnosis can be given to the school system to enable the child to gain access to special services. It also provides an opportunity for parents to gain further insight and help from support groups where they can meet parents facing similar challenges. One parent commented: 'It gave closure and we are very grateful for the test. We, and anyone involved in her lifelong care, will be better equipped for the future'.

10. It is also significant that when a specific chromosome imbalance is diagnosed, the parents (and other family members) can be tested to find out whether they are carriers of changes in their DNA that put them at risk of having more children with a chromosome change. As a result, these new, advancing technologies are empowering parents to have more informed choice in planning for future pregnancies. Similar benefits should be considered as part of the assessment criteria for new and alternative medical technologies that help provide patients with a clear genetic diagnosis.

## Assessment of potential benefits of new or alternative medical technologies

11. The patient perspective is vital in assessing the potential benefits of new or alternative medical technologies. The importance of including the patient perspective during the early stages of assessment is demonstrated in the example of array-CGH. The parent/family member will be best placed to value the real-life impact of early diagnosis and subsequent planning for services that can be realised as a result of the timely uptake of new medical technologies. Genetic Alliance UK supports the introduction of a mechanism that enables patients to participate in the early stages of NHS assessment of technologies.

12. It is vital that the NHS introduce a robust, transparent mechanism for assessing the potential benefits of new and alternative medical technologies which engages patients. Patients in Wales have been unable to gain access to advanced medical technologies that have been introduced in other nations of the UK for the purpose of genetic testing for some years. Array-CGH was introduced by genetics centres in England between 2009-2010; however, the service was only launched in Wales this year as a result of local service reconfiguration. Patients in Wales continue to be disadvantaged because of the lack of investment in clinical care which impedes service delivery, further research opportunities and weakens the impact of the Welsh Government's Science for Wales strategy.

13. As part of NHS assessment, Genetic Alliance UK supports the introduction of a process which aids the timely uptake and continued availability of new medical technologies for the benefit of patients in Wales by ensuring that appropriate funding is made available. Technologies such as Array-CGH and Next Generation Sequencing are revolutionising genetics services by allowing

testing to be undertaken concurrently, at a faster pace and with greater accuracy producing better results.

## Capital Investment

14. Work undertaken by the SWAN UK project has highlighted the extent to which further investment in research and new technologies is vital to advance our knowledge of genetic conditions. Members of SWAN UK are involved in research studies which aim to associate the symptoms that children affected by a syndrome without a name present 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.

15. These new technologies bring greater potential for a broader and more rapid search for patients with a rare disease; however, this service 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.

16. Investment in new technologies will in many cases lead to savings over the previous generation of technology. Better and quicker diagnosis will allow for quicker treatments and fewer events of wrong treatments being delivered to patients. Earlier intervention can prevent the need for more expensive 'end stage' interventions. As a result, health and social care budgets will benefit from this initial outlay on new technologies. Ultimately, capital investment will lead to cost savings and better health.

## The need for sustainability in supporting the uptake of new technologies

17. The availability of funding to aid on-going development costs for new technologies such as next generation sequencing is essential to allow the NHS to keep pace with this constantly evolving field of development. A process to access funding which fills the gap in terms of on-going costs associated with reagents, staff time for developing protocols and validating clinical services locally is essential in allowing Wales to be part of a progressive health service that embraces new medical technologies which are fundamental to genetics research and providing diagnoses for patients.

18. Investment in expertise to analyse the genomic data that is generated as a result of using next generation sequencing technology is essential. There is currently a plethora of data which is being produced as a result of these highly advanced technologies which leads to a disparity between the amount of data being produced and the speed at which analysis can take place. This disparity has been attributed to the lack of sufficient expertise in bioinformatics and sequencing knowledge in Wales which results in inadequate support for conducting analyses of data. This additional support would bring real benefits to patients who currently experience a number of difficulties in accessing a diagnosis. Rare Disease UK's Experiences of Rare Disease: Patients and Families in Wales found that over 23% of patients surveyed had to wait more than 2 years for a diagnosis, with more than 13% waiting over 10 years. 33% of patients attended more than 10 GP appointments before receiving a diagnosis.

19. Genetic Alliance UK endorses the introduction of a process to support the training of specialist informaticians so that they are equipped to analyse the collection of genomic data that is produced as a result of next generation sequencing capabilities. This investment would result in quicker analysis of results leading to faster diagnosis time for patients and greater benefit for the health service in terms of focussed treatment choices and therapeutic planning at an earlier stage in the clinical pathway.

## Embracing medical technologies

20. Genetic Alliance UK accepts that medicines are outside the scope of this inquiry; however, we believe that the NHS in Wales should be embracing diagnostic technologies that are associated with stratified medicine and are currently revolutionising the development process. Companion diagnostics are used to assess a unique trait of a patient's condition (biomarker), the outcome of a diagnostic test will determine whether that patient may respond to the associated treatment.

21. An example of how this technology works for patients can be seen in a treatment for breast cancer. A subset of patients will have lesions that express HER2 (growth-promoting protein). HER2 is also a biomarker for aggressive disease. Herceptin is a drug that has been designed to interfere with how this protein functions so a patient who tests positive for HER2 expression will be expected to benefit from this medicine. Conversely, a patient that does not have a presentation of this biomarker will probably not benefit from this treatment.

22. This method of targeted medicine ensures that patients are treated based on their response profile so that those patient populations who are known to respond to the treatment will receive the right treatment at the right time. This results in improved patient outcomes and a reduction in the number of patients receiving unnecessary treatments avoiding the risk of side-effects. Ultimately the health service will save on resources and costs as medicines will only be commissioned when they work for a specific patient population.

## Joined up approach to commissioning

23. It is crucial that the NHS recognises that there needs to be a joined up approach to commissioning for clinical genetics services. At present the lack of process impacts negatively on patients who are already disadvantaged by the fact that they do not have a diagnosis for their condition and cannot access information or plan for the future as they have no prognosis of how their condition may develop. Genetic Alliance UK calls upon the Welsh Government to introduce and implement a commissioning process for genetics services in Wales.

24. Development of a commissioning process would require input from commissioners, clinicians, geneticists, researchers, Public Health experts and patient representatives who have experience of the service. This group would need to examine the way that genetics services are currently delivered in Wales. It would provide advice about opportunities to improve productivity and efficiency through a process that streamlines services to deliver care that is cost effective and has greater impact on improved outcomes and improved quality of care for patients. This process would include the development of a national procurement policy for medical technologies and the continued development of these services in Wales.

25. Array-CGH and next generation sequencing capabilities are advancing at a rapid speed. It is vital for patients that these technologies are made available so that it is possible to get early diagnoses so that parents/patients can make informed choices and plan for the future. Investment in the early stages of the process will incur further cost benefits later in the treatment pathway as treatments may be tailored to the particular condition. It will also provide benefits for the future planning of services in both the health and social care setting.

    Genetic Alliance UK believes that the introduction of a process for commissioning genetics services is essential to enable NHS Wales to overcome the current barriers in accessing medical technologies. Ultimately, it is patients who will be disadvantaged by the lack of a robust, transparent process for assessing the potential benefits of new or alternative medical technologies and ensuring that they are made available in a timely way.

26. It is a transformational period for many patients with genetic conditions. Medical technology is advancing at a rapid pace and new developments have enabled genetic testing techniques to become more sophisticated, undertaken concurrently and at a faster rate, improving the rate and breadth of the search for a diagnosis. Early and accurate diagnosis will aid understanding which

will allow patients to better manage their condition and plan for the future. It is imperative that the right mechanisms and funding streams are developed to ensure that patients in Wales can take advantage of these latest medical technologies.

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