

GENE PEOPLE

the genetic conditions support network

Medicines and Healthcare products
Regulatory Agency (MHRA)

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Dear Sir or Madam

Gene People response to the MHRA consultation on the future regulation of medical devices in the UK

This letter forms the Gene People response to the above consultation.

Introduction

Gene People (registered charity no 1141583) improves the lives of those affected by genetic conditions, their families, and carers by providing information and support through our unique genetic counsellor-led helpline and web resources, strengthening the members of our free Partnership Network for condition-specific organisations and groups, and responding to national policy initiatives such as this. Our helpline receives around 500 calls each year with many more queries raised by email. There are currently over 130 organisations in the Partnership Network.

Gene People is governed by a Board of Trustees, chaired by Alastair Kent OBE FRSA, who has over 25 years' experience in the field of patient advocacy.

Many of the groups we support are very small, helping those with extremely rare conditions often affecting fewer than 1 in 500k families in the UK. Some operate purely as informal peer to peer support or Facebook groups with no formal legal structure and no paid staff. All the organisations in the Network are experts in the conditions their communities face. All rely on donations to function.

People with rare genetic conditions frequently struggle to access diagnosis and expert support. The diagnostic odyssey can last many years, increasing frustration, fear and stress for the person and whole family. The vast majority of rare genetic conditions are complex multi-system disorders. 80% of genetic conditions are paediatric in origin, with many being progressive and life-limiting. Fewer than 10% have a disease modifying therapy available.

Importance of IVDs.

For those with a genetic condition, IVDs, in the form of genetic tests, are the route to a diagnosis, enabling families to understand their situation, access help and treatment. Families can often wait years for a diagnosis, sometimes never receiving a definitive answer. Their quest for a diagnosis can involve seeing numerous doctors, potential misdiagnoses and having to travel to specific centres. Delays to diagnosis can mean that patients do not receive treatment, support and other interventions that could have improved their health outcomes. Misdiagnosis can lead to potentially receiving



interventions which are unlikely to be helpful. Both scenarios can increase stress and strain on families.

The genetic conditions community was heartened by the 100,000 Genomes Project and its recent expansion. The proposals about genomic sequencing for diagnosis have been largely welcomed as a means to reduce the number of people with no diagnosis. These plans rely on the availability of reliable, affordable genetic tests. The diagnosis yield with genome sequencing is now in excess of 30% and rising, indicating that there is a substantial potential for innovative technologies to change the situation for patients and their families.

In addition, the genetic conditions community has a high utilisation of devices with variable degrees of complexity and invasiveness. Some with a genetic condition might need to use asthma inhalers, whereas others will need implantable defibrillators. For some, IVDs that were previously used in clinical settings are now being used in the home, with the increase of homecare. There is also a rise in the use of software to monitor patients with genetic conditions. Some of this monitoring is conducted remotely, minimizing disruption to the lives of patients and their families while enabling the delivery of improved healthcare. It goes without saying that the pandemic has demonstrated how important the ability to maintain health supervision from a distance can be to patients.

Given the importance of IVDs to this community, the lack of recognition of the specific needs of this population in the consultation is puzzling. Therefore, the rest of this response outlines the issues that should be considered for those affected by rare and genetic diseases.

Issues for Rare and Genetic Disease affected families.

Families and the community want and need confidence that the regulatory framework is appropriate, fit for purpose and proportionate.

Regulation is not simply a matter of invasiveness for those with genetic conditions but also the consequence from using the device. A simple blood test is minimally invasive, but the information revealed by genomic analysis may be devastating for an individual and their whole family. Gene People has concerns about the role of recreational genomics, such as well-known direct to consumer tests. The regulation of private sector companies has significant implications and clarity about the reliability and claims made for results is desirable. The lack of support available to people who have used direct to consumer tests is worrying. There is the potential for misuse of personal information should a genome sequencing company go bankrupt or be taken over by other companies that could be based in less well-regulated jurisdictions with greater latitude to data protection. This is also a concern.

Although an increasing number of diagnoses will be through genome sequencing this is not a complete answer for those with rare conditions. Non-heritable and chromosomal anomalies will rely on a variety of different technologies. In many cases the numbers used each year will be very low so imposing quality assurance frameworks designed for commonly used tests could impose unrealistic barriers to access.

While consent is crucial Gene People suggests the scope of consent – for clinical use, research and for quality control - ought not to be too narrow and restrictive. Most patients in the rare and genetic disease community are keen and willing to support research. Broad consent at time of taking the sample may be further refined when

specific projects arise at some future date. The normal boundaries to research use should apply, such as right to withdraw. NB consent would apply to identifiable samples and data only.

Gene People would not support measures that restricted research by health institutions either directly or indirectly. The proposals in the consultation are insufficiently specific to be able to form a judgement on whether these would impede the progress of knowledge on these complex conditions.

For anonymised samples, no consent is needed or possible once identifiers removed. Patients need to be told if their samples will be retained, why, and for how long, and for what range of potential uses the samples may be used at the time of collection.

For some families affected by rare disease, there is anxiety about unintended consequences, in particularly around genome sequencing for both clinical and non-clinical uses. These anxieties tend to be about being able access to new information as knowledge advances, and fear of discrimination or stigma. Such anxieties can be managed through consistent and clear consenting. We would suggest that those collecting samples and consents from patients with rare and genetic conditions have knowledge of rare conditions and the impact that they can have on the patient and wider family as it is a different experience for those giving samples than for those trialing a general device.

It is important that patient and family engagement can act as a guarantor of probity, as well as adding value to studies through their lived experience, and being a source of additional expertise and additional supplementary evidence. Gene People would ask that the MHRA develops a methodology to capture the benefits from patient and family engagement and that the means to be able to share this with the wider technologies system is included.

The patient cohorts available for some rare and genetic conditions are very small indeed. Some conditions represented by member organisations in our Network have no more than 200 patients worldwide. The avoidance of unnecessary parochialism in the case of rare conditions is key as there may not be enough patients in UK to generate sufficient data.

Gene People welcomes the proposal for summary information about devices to be published. These need to be accessible lay summaries for the average reading age of the UK population (currently 9 years old) that are easily available in a central repository. The public should be informed of this and awareness of this properly and regularly disseminated.

The clinical utility should be explicit. A CE marking that merely says confirms that a device behaves in the way that it was intended is not as rich in information as it could be for commissioners and patients. As the whole health system is under pressure, documenting whether a device is helpful or not would be useful additional evidence that might speed decisions or save time. Speed of access is often a matter of life and death for those with rare diseases.

One of the promises of genome sequencing is increasingly personalized, precision treatments. It is unclear from the consultation how companion diagnostics will be regulated in the context of precision medicine.

Technical consultation.

The consultation is unwieldy and difficult for a person not immersed in the current system to be able to respond meaningfully. It assumes a working knowledge of the system and where the limits to regulation currently are placed. It is also very difficult to work out how the answers will interact in building a regulatory framework, for example, will saying yes to a proposed change in one area have unintended consequences in another. Therefore, we took the unusual step of not responding to the consultation via the website, rather, responding by letter.

We would suggest that a direct programme of engagement by the MHRA with the rare and genetic condition patient community to explore their understanding of and expectation from the revised regulations is undertaken. Gene People would be willing to work with the MHRA on such a programme.

Conclusion.

Gene People welcomes the efforts of the MHRA to provide additional clarity by revising the regulations. We also welcome the acknowledgement of and early thinking about innovative areas such as software medical devices.

We are concerned that:

- the needs of those with genetic conditions, especially those that are rare and extremely rare, are not recognized in the consultation as they have specific needs
- there is a lack of patient voice and feedback solicited during the evaluation process – whilst the emphasis is on safety it would be desirable to start the journey to market with some form of input from patients
- the technical nature of the consultation will have deterred rare and genetic condition organisations from responding.

We look forward to the outcome of the consultation being published in due course.

Yours sincerely



Samantha Barber FRSA, Chief Executive