

GENE PEOPLE

the genetic conditions support network

Response to Statutory Scheme Consultation

Who this response is from

This response has been written by Gene People, a registered charity that provides information and support to individuals and families affected by genetic conditions, and the condition-specific support groups that work with their communities.

Gene People was formerly known as Genetic Disorders UK and was founded in 2011. Our genetic counsellor-led helpline supports approximately 200 callers each year. Our free to join Partnership Network has 141 condition-specific support groups and charities as members, representing thousands of people impacted by genetic conditions and their families.

Gene People has sent this narrative response to the consultation rather than completing the survey questions as the questions are not relevant to the situation of Gene People and our Partnership Network, rather they are intended for the medicines industry. However, Gene People firmly believes that the voice of the patient should be heard in this consultation.

The rare disease context

There are approximately 3.5 million people affected by rare conditions in the UKⁱ, which means many more people from their wider families are involved in caring for and supporting them.

Estimates of the numbers of rare diseases have, until recently, suggested that there are around 7,000 conditions. Rare-X have published a report that posits that the number of rare diseases exceeds 10,000ⁱⁱ, suggesting a considerable amount of mis- and under-diagnosis.

Approximately 80% of rare conditions have a genetic cause. Very few conditions are screened for currently, although projects like the 100,000 Genomes Project have been seeking to advance diagnosis for patients.

Most rare conditions begin to present symptoms in childhood, with a significant proportion of those conditions (more than 30%ⁱⁱⁱ) resulting in the death of a child before their fifth birthday.

There are treatments for approximately 5% of rare conditions and access is defined by local arrangements. For clarity, this means that 95% of rare conditions have no treatment.

Many rare diseases need high levels of health and social care interventions and can have a high impact on family life, including increased financial pressures and poor mental health^{iv v vi vii}.

Response to the Statutory Scheme

Gene People is deeply concerned about the impact the proposed rise in rate for the Statutory Scheme will have on the development of and access to innovative medicines and treatments for those with genetic conditions.

We are uncertain as to what extent the specific situation of individuals affected by genetic conditions as set out above have been taken into full consideration when the proposed scheme was developed .

We believe that the proposal will have a negative impact on our community as we do not believe that it will help the UK achieve positive life science competitiveness indicators^{viii}. It is unclear as to how the proposal will help achieve the Life Sciences Vision or the Genomics Strategy.

There are two major areas of concern: decreasing clinical trials and ensuring timely access to treatments for patients with rare diseases.

Decreasing clinical trials

Following the publication of the ABPI report on industry clinical trials^{ix} that showed a 41% decrease in new trials over a four-year period, the BMJ published an article including this statement from the chief executive of the Association of Medical Research Charities, Nicola Perrin: "...urgent action is needed to ensure that the UK remains an attractive place for funders to support clinical trials."^x Gene People agrees with this statement.

For many people with genetic conditions, participating in a clinical trial is the fastest way to access innovative – and perhaps the only – treatment for their condition.

Trials also creates an element of hope for families that fosters a greater ability to cope; they are more in control of their fate and have actively decided to take part in a trial. Given the huge mental strain having a loved one with a genetic condition can cause a family being able to exercise agency is crucial. If companies take the view that there is little prospect of a return on their investment in the UK then it is likely they will locate trials elsewhere, making participation difficult or impossible for UK-based patients.

It has already been shown that very few rare diseases have treatments available. Decreases in clinical trials are likely to disproportionately impact the rare disease community as there are fewer treatments in the R&D pipeline in comparison to the non-rare disease pipeline.

Access to treatments

While clinical trials are one route to accessing innovative treatments, trials are only open to a specific number of patients. Once the trial has been completed, the treatment will need to go through an appraisal process for the devolved nations. These often take a long time, particularly in England^{xi}, and restrict access to the treatment while the appraisal is conducted. This can lead to poorer health

outcomes for patients and allow potentially avoidable adverse health consequences to occur.

Gene People is concerned that the uncertainty of the appraisal processes is already proving a disincentive to industry to launch new medicines in the UK^{xii} and the combination of difficult and long appraisal processes and a high rebate regime could create an operating environment that discourages drug launches.

Gene People is in favour of a life sciences ecosystem that incentivises research and creates a likelihood of a fair return for the investment made. This will mean that people with rare genetic conditions in the UK will be more likely to get access to the treatments that they so desperately need.

Creating a downward spiral?

Gene People is concerned that the proposal contributes to a downward spiral for life sciences in the UK.

The consultation document states:

We consider that the proposed approach, which will reduce statutory scheme member company revenues compared to the counterfactual where no update is made, may lead to some reduction in research and development investment of which a proportion would be felt in the UK. The department considers that research and development investments lead to 'spillover' effects – for example, through the generation of knowledge and human capital – which generate net societal benefits, compared to other companies spending their capital on other things. In addition, research and development investment could lead to improved medicines in the future that would be of benefit to patients and the health service.

There is uncertainty as to how long the process of recovering from that reduction in research and development, loss of intellectual property and skills and expertise, and the disbenefits to patients and the NHS would take. It is unclear how such a recovery would be stimulated.

It is also clear that the UK's exclusion from EU Horizon funding is acting as a disincentive to academics coming to UK universities, reducing the talent pool available to support innovative therapy development here. This may have a disproportionately adverse impact on therapies for rare diseases where expertise is already scarce,

The community Gene People serves does not receive the same level of attention and investment in life sciences as conditions that are population health issues. This means this downward spiral would disproportionately impact those with rare and genetic conditions.

Gene People, therefore, seeks reassurance that the special situation of those with rare diseases, especially those that are genetic, has been given full and proper consideration and analysis. We ask that the proposal is reconsidered.

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- ⁱ <https://www.gov.uk/government/publications/uk-rare-diseases-framework/the-uk-rare-diseases-framework>
- ⁱⁱ <https://rare-x.org/blog/2022/06/07/rare-x-releases-new-report-that-uncovers-large-number-of-previously-uncounted-rare-diseases/>
- ⁱⁱⁱ <https://www.gov.uk/government/publications/uk-rare-diseases-framework/the-uk-rare-diseases-framework>
- ^{iv} <https://bmchealthservres.biomedcentral.com/articles/10.1186/s12913-022-08060-9>
- ^v <https://contact.org.uk/help-for-families/campaigns-and-research/counting-the-costs/>
- ^{vi} O'Dwyer et al. <https://doi.org/10.1016/j.comppsy.2021.152261>
- ^{vii} [https://www.thelancet.com/journals/eclinm/article/PIIS2589-5370\(21\)00514-9/fulltext](https://www.thelancet.com/journals/eclinm/article/PIIS2589-5370(21)00514-9/fulltext)
- ^{viii} <https://www.gov.uk/government/publications/life-science-sector-data-2022/life-science-competitiveness-indicators-2022-life-science-ecosystem#life-science-competitiveness-indicators>
- ^{ix} <https://www.abpi.org.uk/publications/rescuing-the-uk-industry-clinical-trials/>
- ^x <https://www.bmj.com/content/379/bmj.o2540>
- ^{xi} <https://www.sciencedirect.com/science/article/pii/S0168851022000239>
- ^{xii} <https://pharmaphorum.com/news/gene-therapy-specialist-bluebird-exits-untenable-european-market/>