Genetic Alliance UK
Supporting. Campaigning. Uniting.

Consultation Response

Genome Editing

Open call for evidence – Nuffield Council of Bioethics

Response from Genetic Alliance UK, 5th February 2016

Introduction

1. Genetic Alliance UK is the national charity working to improve the lives of patients and families affected by all types of genetic conditions. We are an alliance of over 180 patient organisations. Our aim is to ensure that high quality services, information and support are provided to all.

2. Rare Disease UK (RDUK) is a campaign run by Genetic Alliance UK. It is the national alliance for people with rare diseases and all who support them. RDUK is a stakeholder coalition brought together to work with Government to develop a UK Strategy for Rare Diseases, which was published by the Department of Health in November 2013. RDUK continues to campaign for the implementation of the Strategy across all four home nations of the UK.

3. SWAN UK (Syndromes Without A Name) is a patient and family support service run by Genetic Alliance UK. It is a UK-wide network providing information and support to families of children without a diagnosis. It works to support the development of high quality information and services for families of children affected by undiagnosed genetic conditions and raise public and professional awareness of undiagnosed genetic conditions and the unique challenges faced by affected families.

Unmet health need as a background to the rare, genetic and undiagnosed communities' views

4. Genetic Alliance UK represents all those affected by genetic conditions. This is a growing community as our understanding of the influence of genetics on our health grows. We have an ever increasing number of rare single gene conditions, that currently number more than 6,000. As our understanding of the human genome grows, more of these are identified every month.

5. Our work with SWAN UK community has shown us that there are a myriad of poorly understood 'syndromes without a name' that affect children in serious unpredictable ways. Genome sequencing is bringing a better understanding of the cause of these conditions to a very small number of families, but this is very much the tip of an iceberg of a group of children for whom the diagnostic odyssey is prematurely stopped at global developmental delay or another symptom based false diagnosis. We estimate that approximately 6,000 children a year are born who are affected by a condition that geneticists will be unable to diagnose.

6. There are very few effective cures or treatments for single gene conditions, or for other conditions with a genetic origin. We have hematopoietic stem cell (bone marrow) transplantation, which is effective in a broad minority of genetic conditions but is not without its risks and challenges. We have a few small classes of innovative medicines, the majority of which focus on replacing missing

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steps in our metabolic pathways. The affordability of these medicines is increasingly being brought into question. And there are a handful of other inherited conditions that can be treated with clotting factors and dietary adjustments and supplements. For now, most genetic conditions have no cure or effective treatment. Palliation and mitigation are the most patients with genetic conditions can hope for, and for affected families, prevention is the strongest and most developed tool that might allow them to take control of the condition's impact on their family.

7. There is a tremendous amount of unmet health need in the rare, genetic and undiagnosed communities. Our members look towards research as the major potential source of progress in all of these areas. With thousands of conditions that are still relatively poorly understood, we know there is a tremendous amount of work that needs to be done. Patients affected by serious genetic conditions would like to see all promising avenues of research fully investigated.

8. Genome editing appears to be a powerful tool that may allow researchers to take new approaches to understand the fundamental biology that causes the genetic conditions that affect our members, and our members would like to see that potential realised.

Genome editing from the perspective of patients and families with unmet health need

9. Genome editing holds a few major potential strands of benefit to patients and families affected by genetic conditions: i) as a research tool, allowing us to better understand the cause of genetic conditions, and potentially inform the development of treatments; ii) as a technique in the manufacturing process of treatments for individuals or populations; and iii) as a potential addition to the range of reproductive choice techniques available to avoid the birth of children affected by genetic conditions.

10. These uses of genome editing certainly overlap, in that we may need the former before we can have either of the latter, but as we work to better understand the technique we should work to keep the uses separate. Not least because they are at different stages of feasibility and implementation.

11. As a tool for reproductive choice, we will be interested to see how the technology develops and where genome editing is found to be most useful. Preimplantation genetic diagnosis (PGD) has proved to be tremendously valuable to couples at risk of having a child affected by a genetic condition and is widely applicable to single gene disorders. Providing the family has the potential to conceive an unaffected embryo, PGD can be used to select an unaffected embryo and ensure a positive outcome. (Though of course the success rate is not 100%.) Though PGD is a viable technique that meets the needs of many families who seek to avoid the birth of children with serious genetic conditions, we should not rule out the eventual replacement of PGD with a new technique, which may be based on genome editing.

12. There are some family circumstances in which PGD cannot, or is unlikely to, allow the implantation of an affected embryo. These situations include:

    - Situations in which the couple are at risk of having a child with more than one condition and the chance of producing an unaffected embryo is therefore very low. This situation may be more likely in certain ethnic groups where the prevalence of particular genetic conditions is increased.

    - Situations in which the couple are both affected by the same recessive condition and cannot produce an unaffected embryo. Examples include sickle cell disease, some forms of thalassaemia, alkaptonuria and Fanconi anaemia.

    - Situations in which the male member of the couple has a dominant X-linked condition and the couple cannot conceive an unaffected female child.

13. It should be noted that a significant number of these conditions affect ethnic minority populations disproportionately.

14. It is feasible that there are those who might reject PGD on ethical grounds due to the discarding of human embryos, but might consider using a reproductive choice technique based on genome editing if no human embryos need be destroyed.

15. We have seen that mitochondrial donation has the potential to solve a reproductive choice issue that faces those at risk of conceiving embryos affected by conditions caused by mutations in mitochondrial DNA: PGD is in some cases an imperfect solution and in other cases does not work. Without mitochondrial donation, couples in this situation do not have a route to be sure that a pregnancy is unaffected. Genome editing has the potential to cover other similar gaps in our portfolio of reproductive choice, and to allow more members of the genetic condition community to be able to have pregnancies that are certainly unaffected by the condition they are at risk of passing on to their offspring. This would bring greater equity of access to reproductive choice.

16. The UK political and regulatory systems have in the past dealt effectively with controversial new research tools and reproductive choice techniques. Examples include human admixed embryos, mitochondrial donation, and the regulation of PGD and research on human embryos. Though there are new topics to discuss in the case of genome editing, such as the potential for germ line alteration, there is no reason to suppose that the approaches that have been successful in the past: good quality engagement, open debate, ethical consideration of the pros and cons of the activity weighed against the pros and cons of not permitting the activity, alongside prudent regulatory and political decisions timed appropriately; should be successful again.

Our members' views

17. As part the European Commission funded NERRI project (Neuroenehancement: Responsible Research and Innovation), Genetic Alliance UK surveyed patients and families affected by genetic conditions to examine their views on genome editing as a potential tool for treatment, and as a potential tool for enhancement. The survey had a significant focus on the patient perspective on ethical use and regulation of genome editing technologies. In line with the work of the NERRI project, we questioned respondents on the ethical use of such technologies to improve cognitive abilities in individuals living with a genetic condition, and in healthy individuals.

18. 163 respondents completed our survey from beginning to end. 152 of these completed responses came from patients, or their families or carers. Our key findings are:

19. Patients are interested in genome editing technologies, and would like to learn more about them. Two thirds of patient respondents had thought about the implications of genome editing technologies, and over 80% were interested in finding out more about these technologies. We received over 200 responses to the survey in the five weeks that it was live, suggesting that genome editing technologies are a topic of great interest to those living with genetic conditions. Patients and families are open to engaging in conversations that will enable them to learn more about the potential of these technologies.

20. Patients welcome the use of genome editing technologies in research and clinical settings, but are clear that such uses should be limited to treating medical conditions and not for the enhancement or alteration of physical or cognitive attributes of healthy people. Respondents overwhelmingly supported the use of genome editing technologies in research, where that research is focused on treating medical conditions. Respondents were equally welcoming of the use of genome editing technologies in a clinical setting, but again, a clear distinction was drawn between acceptable uses in a medical context, and the use of technologies to enhance physical attributes in healthy people, which was deemed unacceptable by most.

21. Patients support a multiple stakeholder approach to regulatory decisions to ensure ethical use and applications of genome editing technologies. This is compatible with the regulatory approach that is taken by Human Fertilisation and Embryology Authority.

22. Overall, we found that patients feel the future of genome editing technologies offer more potential benefits than risks, if regulated appropriately and used in the treatment of medical conditions.

23. There is significant discussion and debate as to the where the distinction between treatment and enhancement lies, and as to whether the distinction is valid. In the case of the membership of Genetic Alliance UK, it is perhaps an easier distinction to make given the profound unmet need that our members face and the type of treatment that they might imagine.

24. Genetic Alliance UK will be publishing the full findings of our survey in the coming weeks. We would be happy to share as much of our research as is helpful with the Nuffield Council of Bioethics review team.

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Director