Introduction

About 7,000 diseases are classified as rare genetic diseases, affecting the lives of about 5% of the world's population. Genetic therapies are commonly pointed out as the future of treatment for rare genetic diseases, promising to reduce symptoms and even cure patients. This global survey aims to collect the opinions of researchers with knowledge of genetic therapies for the treatment of rare genetic diseases to inform scientists, physicians, policymakers, and interested communities on the future perspectives on this topic.

Your contribution

We are asking you to share your perceptions regarding genetic therapies for the treatment of rare genetic diseases. The questionnaire will take only 4 minutes of your time to complete. The questionnaire will be available for completion for the next eight days.

Confidentiality and Privacy

This survey is for research purposes only, and your participation is voluntary. We will not ask you to provide any personal or sensitive data, and your individual responses will not be identified. By answering the questionnaire you give us your informed consent for the use of any data you provide. If you have any queries about this survey, please email cee.foresight@fiocruz.br.

The survey results will be submitted to a peer-reviewed journal, and we will send you the article once published.
* 1. Please indicate your knowledge level on genetic therapies for the treatment of rare genetic diseases

- I have high knowledge
- I have good knowledge
- I have some knowledge
- I have no knowledge
* 2. Considering the ORPHANET classification of rare diseases, which of the following groups best fits your research expertise?
3. Genetic therapies will be the standard of care for rare genetic diseases.

- Likely before 15 years
- Likely after 15 years
- Unlikely
- Unknown
Future of genetic therapies for rare genetic diseases

4. Considering the next 15 years, which of the following vectors is most likely to be successful in fixing or replacing defective genes?

- Viral vectors (e.g. Adeno-associated virus, Lentivirus, Retrovirus)
- Non-viral vectors (e.g. Liposomes, Plasmids, Peptides, mRNA therapies)
5. Please rank the following approaches from most likely to least likely to be successful in fixing or replacing defective genes in the next 15 years (4-point scale, where 1 is most likely and 4 is least likely)

- [ ] Transcription activator-like effector nucleases (TALENs)
- [ ] Zinc finger nucleases (ZFNs)
- [ ] CRISPR-Cas9
- [ ] Meganucleases (MNs)
6. Genetic therapies will have long-lasting effects, not requiring repeated interventions.

- Likely before 15 years
- Likely after 15 years
- Unlikely
- Unknown
7. Genetic therapies will lead to a cure for rare genetic diseases.

- Likely before 15 years
- Likely after 15 years
- Unlikely
- Unknown
8. Would you like to share something else with us?
Thank you for your answers. Now, please let us know a little about you.

9. Which rare genetic disease are you most familiar with?

10. What is the highest degree of education you have completed?
   - Associate’s degree
   - Bachelor’s degree
   - Master’s degree
   - Doctoral degree

11. Which occupation type best applies to you?
   - Masters, Ph.D. Student
   - Professor, Researcher
   - Physician, Clinician
   - Public Health, Healthcare Professional
   - Manager, Executive
   - Policymaker
   - Other

12. What type of institution do you work in?
   - University, Research Organization
   - Hospital or similar organizations
   - Government
   - Industry
13. How many years of experience you have in your field?
- Less than 5 years
- Between 5 and 10 years
- Between 10 and 20 years
- More than 20 years

14. In which region do you live?
- Asia (including the Middle East)
- Australasia or Pacific Islands
- Africa
- Europe
- North America (including Central America and the Caribbean)
- South America