Examining the role of genetic counselors to promote patient centricity in cell and gene therapy clinical trials

Contributor

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As the list of FDA-approved cellular and gene therapy products continues to grow,[1] oncology and rare diseases remain the most commonly targeted indication groups among the gene, cell and RNA therapeutic pipelines. Given that an estimated 87% of rare diseases have a known- or suspected-genetic basis,[2] the role of genetic counseling in clinical trials is expanding not only from the perspective of eligibility confirmation but also to enhance patient centricity.

This article explores the role that genetic counselors can play in a clinical trial and shares the results of a pilot study that used genetic counseling services. It also discusses strategies for optimizing the implementation of complex cell and gene therapy studies by incorporating genetic counselors to ultimately help ease participation for patients and their families.

Enhancing patient centricity through genetic counseling

At a high level, genetic counselors can serve as partners to a clinical trial's site staff by interacting with clinical trial participants, educating them about the study, answering questions, providing resources and easing any anxieties. Whether meeting in person or through telemedicine, these specialists can help patients feel empowered to make study-related decisions, ensure the consequences for the patient's broader family are understood and build trust within the patient community.

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Importantly, many services provided by genetic counselors in a patient care setting can be easily transferred to clinical trials, while others are aspirational and potentially achievable with additional training. These services include:

Promoting study design and awareness	Genetic counselors are well-positioned to provide early insights on the targeted patient population and potentially participate in collecting family feedback on the study design. They can represent an additional resource to help promote clinical trial awareness as they educate their patient community about the availability of clinical trials.
Providing pre- and post-testing support	In the pre-testing phase, genetic counselors discuss the role of genetic testing in a clinical trial and explain risk assessment and informed consent. After genetic testing, the counselor discusses the patients' results, provides referral recommendations and explains the meaning of variants of uncertain significance (VUS), if applicable. Genetic counselors can also help explain to patients and families why they are not eligible for a specific clinical trial, provide them with access to other potential options and direct them to other available resources (e.g., patient advocacy groups, clinicaltrials.gov).
Enhancing patient identification	To support patient identification in a clinical trial, genetic counselors would need to be equipped with a strategy for addressing undiagnosed or misdiagnosed patients and have the ability to analyze proprietary genetic testing data. The identification process can also include identifying relevant physicians and may also involve identifying additional affected or at-risk individuals and family members.
Supporting patient recruitment and pre-qualification	Patient recruitment represents another key area where genetic counseling services could add value to a clinical trial. Assuming previously tested patients have opted-in to be contacted about current or future studies, a genetic counselor is able to provide direct-to-patient and family outreach, in an appropriate manner that adheres to patient privacy policies and can support pre-qualification for a study based on genetic test results and available medical history. Of note, regulatory authorities are increasingly requiring a robust process for confirmation of genetic results.
Contributing to study education and endpoint collection	Genetic counselors could also play a role in delivering study education and even potentially administering interview-based endpoints for a trial. As trained healthcare professionals, these counselors could answer questions about clinical research, provide support for patient questions, interpret genetic test results for any variant-related risks and facilitate informed decision-making about clinical trial participation. With training, they could also administer study questionnaires and collect data on patient experience during the trial, as well as potentially assisting conduct long-term follow-up (LTFU) required for cell and gene therapy studies to promote integration and continuity for patients and their families.

Case study: Supporting a qualitative telemedicine interview study with a genetic counseling service

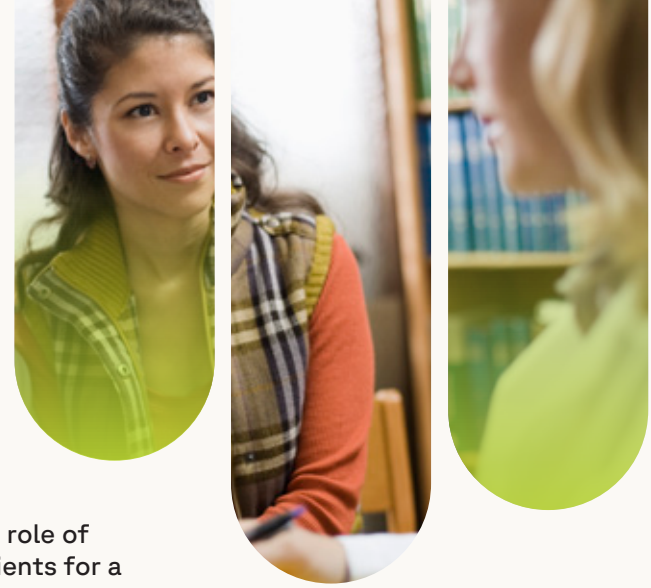
To understand patient and caregiver perceptions of their experience with Limb-Girdle Muscular Dystrophy (LGMD), a cross-sectional, qualitative interview study was conducted in the U.S. Using a genetic counseling database to identify 227 people living with specific LGMD subtypes, 138 people were initially identified as eligible and had consented to receive contact.

After three contact attempts, 74 people were deemed "potentially eligible" to enroll and 33 were contacted to participate. **Of the 33 people contacted, 26 (79%) consented to receive additional information and completed structured interviews via video conference.** Pediatric participants were interviewed with their caregivers while adult participants were interviewed individually. Participants provided positive feedback about their role in helping advance LGMD research and the engagement of a genetic counseling service helped facilitate the successful completion of the study.

Optimizing participation of genetic counselors in clinical trials

Incorporating genetic counselors into a clinical trial requires evaluating study complexity, especially from the perspective of the patient population, and then clearly defining how genetic counselors can best help address key challenges. Based on our experience at Fortrea with delivering comprehensive genetic counseling and personalized genetic risk assessment for clinical trials, our team has gathered several key considerations for drug development sponsors.

- **Clinical research design and feasibility**
  Beyond complying with all genetic testing guidelines, sponsors should understand the validity and any potential limitations of genetic testing and reporting. In this phase, process development is essential to discuss and define several factors, such as who orders genetic testing, how results are reviewed, which results are disclosed (and to whom) and how genetic counseling activities are tracked.
- **Approvals and regulatory requirements**
  The use of sponsor-initiated genetic testing requires approval from Institutional Review Boards (IRBs)/ Ethics Committees (ECs) with informed consent and assent forms as well as privacy protections in place. In terms of regulatory requirements, direct patient and family engagement with genetic counseling is feasible within the U.S.; outside of the U.S., genetic counseling may need to be delivered via investigators and site staff. However, new centralized genetic counseling services that can support clinical trials across European countries are emerging.



Awareness and patient recruitment
To promote patient awareness and recruitment, sponsors can consider the role of genetic counselors to help identify patients for a clinical development program or to direct patients to potential studies for clinical trial participation through clinicaltrials.gov and reputable patient advocacy websites.

Participant screening and education
If genetic counselors are involved in the screening process, sponsors must establish how the counseling will be provided and who delivers patient education regarding the study. This process requires careful coordination with the clinical site(s) to ensure consistent messaging enhances trial engagement and commitment. Sponsors may also consider the value of expanded family education (e.g., outreach to a patient's sibling(s) with age-appropriate, long-term updates).

Long-term follow-up (LTFU)
Sponsors should be aware that genetic counselors represent one method for offering high-touch, accessible solutions in LTFU studies. They can support test interpretation and communication of results as well as offer counseling and coordinate remote care with study physicians, as needed. They can also help inform the refinement of the predicted path vs. the altered path post-treatment.

Genetic counselors can make significant contributions to a clinical trial—beyond confirming participant eligibility—and their services can be tailored to meet the specific needs of a study and its participants. Whether helping expand patient identification, enhancing patient education or promoting family engagement, genetic counselors can help make clinical trials more accessible and create a pathway for lifelong patient support.

Learn more about our patient-centric approaches to optimize the development of cell and gene therapy products here: https://www.fortrea.com/scientific-expertise/specialty-areas/cell-and-gene-therapies.html

References

1. U.S. FDA. Approved Cellular and Gene Therapy Products. https://www.fda.gov/vaccines-blood-biologics/cellular-gene-therapy-products/approved-cellular-and-gene-therapy-products Accessed Apr 17, 2024.
2. Lamoreauz, K, et al. The Power of Being Counted. *RARE-X* publication. https://rare-x.org/wp-content/uploads/2022/05/be-counted-052722-WEB.pdf

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