THE JOURNAL OF GENE MEDICINE
J Gene Med 2007; **9**: 922–923.
Published online in Wiley InterScience (www.interscience.wiley.com) **DOI:** 10.1002/jgm.1097

RESEARCH ARTICLE

A patient and family perspective on gene therapy for rare diseases

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Received: 15 June 2007
Revised: 30 July 2007
Accepted: 31 July 2007

Abstract

The authors describe the view of patients and patient organisations on gene therapy research and gene therapy regulations. In particular, the added value of partnership between scientists and patient organisations, and patient involvement in the gene therapy field, are addressed. Copyright © 2007 John Wiley & Sons, Ltd.

Keywords gene therapy; patient perspective; regulation; clinical trials

Introduction

For decades gene therapy has been held up as the great promise for treatment of otherwise incurable inherited conditions. In particular, it has been seen as the road to treatment of rare, single gene disorders. Despite initial optimism and some promising early results (e.g., in cystic fibrosis (CF) and severe combined immune deficiencies (SCID)), gene therapy so far has not delivered a range of clinically available interventions that will treat, prevent or cure the growing number of identified rare diseases. Yet despite this apparent lack of progress, patients and families continue to be amongst the keenest advocates for research and development in gene therapy.

Why is this? Why do patients and families continue to endorse gene therapy as a topic for investigation and in some cases raise substantial funding to support targeted gene therapy research programmes (in the UK the CF Trust has generated millions of pounds to underpin the work of the UK CF Gene Therapy collaboration)?

The context for families

Most rare genetic conditions are complex, multi-system disorders. Any effective therapy would need to intervene across the board if it were to treat or prevent the impact of the disease in an affected individual. Given that little is known about many of the rare single gene disorders, and the limited likelihood of a comprehensive programme of R&D being set up to develop specific curative intervention, it seems to many families that gene therapy offers a route to tackling the fundamental biological cause of their disorder, and eventually would eliminate the need for complex services of integrated symptomatic treatment (much of which needs to be lifelong with increasing intensity). Even where palliative symptomatic interventions currently exist these are often demanding and of limited benefit to the affected person (e.g. the daily treatment regime for children with CF).

Optimism or realism?

The commitment shown by patients (and by many patient support groups) to support gene therapy R&D might be a consequence of the early hype surrounding this issue, especially the 'miracle cure' stories that have appeared from time to time in some sections of the media. If this view is correct it is also necessary to assume that patients and families are naive and gullible. Whilst this may be true of some, the vast majority of families are too firmly grounded in the reality of the situation to be taken in by the prospect of an instant quick fix. They know, much as they might wish otherwise, that the reality is a complex biology of their disease and that genotype-phenotype interactions will be difficult to unravel. They also know that miracle cures rarely (if ever!) happen, and are wary of claimed breakthroughs. Thus the support from patient groups for gene therapy is not blind or unconditional. Rather it is rational and evidence based.

Patients and families who belong to support groups for rare genetic disorders often have a very close relationship with interested/expert clinicians and the research community, as much of the diagnosis and treatment is provided by specifically assigned centres at a national level. This creates a mutually beneficial, sustainable partnership that helps to create a framework for rational decisions on the best way forward. Patient organisations are not blindly committed to gene therapy. If another route were to become more promising their focus of attention would shift. Their interest is the outcome, not the process.

From research to clinical practice

Any gene therapy that looks like it is going to work will have to be licensed by the relevant competent authority. The European Commission has recently introduced regulations (the Advanced Therapies and Tissue Engineered Products Regulations). These have just (May 2007) been adopted [1] in the face of substantial opposition from religious and environmental factions and will provide a common pan-EU framework for evaluating quality, safety and efficacy of novel therapies including gene and stem cell therapies introduced to the market. Patient advocacy groups played a key role in persuading the European Parliament and the Council of Ministers of the importance of these legislative proposals and of the need to avoid confusion across the different Member States, and to avoid any delay.

As the number of clinical trials for diseases amenable to gene therapy rapidly increases, and as the trials move from phase 1/2 to phase 3, the prospect of applications for marketing authorisation becomes a reality, so a clear, well-defined regulatory framework is ever more necessary. Whilst most of these (potential) products are for cancers, the most successful ones are in the area of inherited disorders, which provides substantiated hope for future progress in other rare genetic disorders. For this hope to be translated into reality it will need a sustained commitment to high-quality medical research. Public understanding and support is essential if the necessary investment is to be provided, and the rational advocacy of patient organisations (who are disinterested, but not uninterested, in the outcome) is a key component in creating this public support of the continuance of this research. Therefore, the European patient umbrella organisation EGAN (European Genetic Alliances' Network [2]) actively participates in major European research projects on gene therapy, such as Consert [3] and Clinigene [4], among others, in contributing to the ethical debate from the patient perspective, especially on items related to clinical trials, and by communication to the public.

Conclusions

Gene therapy offers significant promises for patients with inherited genetic diseases. Trust based on mutual respect, transparency and commitment to collaboration should nourish the progress on which the development of effective treatment aiming at cure flourishes.

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DOI: 10.1002/jgm