SCIENCE AND TECHNOLOGY

https://daily27.info/2026/09/10/health-and-research-equity-a-matter-of-principles-and-rights/

Health and Research Equity: A Matter of Principles and Rights

By Antonio Liras

SEP 10, 2026

#Economic cost, #Healthcare, #Human Rights, #Innovation, #Knowledge, #Medicine, #Rare diseases, #Research

Image generated with Artificial Intelligence

It is well known that the objective of basic research (R&D) —research and development— is to generate new knowledge, whereas applied research (R&D&I) —research, development and innovation— translates this knowledge into practice in ways that improve people's quality of life. In both cases, the economic cost is high, and even more so in applied research, which is why companies (spin-offs, start-ups, or large multinational pharmaceutical companies) are often the ones that bear these costs. In healthcare, this translates into the development of new diagnostic methods or treatments through clinical trials involving patients.

The impact of this translational medicine is social, as it seeks to improve people's lives, but it is also economic, since substantial investment is required to develop healthcare innovations. This is where the so-called **commercial portfolio objectives of a company** towards its customers or investors come into play. This is the key point of discussion and controversy when we talk about health *vs.* disease, because there will be a medical and social benefit as long as there is economic profitability for pharmaceutical companies. This may sound "harsh" when referring to any type of disease, but even more so in the case of a rare or ultra-rare disease.

A rare disease (affecting 1 in 2,000 individuals) or an ultra-rare disease (affecting 1 in 50,000) [1,2] is not particularly profitable for **a company's commercial portfolio**. Herein lies the conflict: if investment is not made, health benefits cannot be generated, but such investment is ultimately based on profitability (**economic considerations based on the higher or lower incidence of a disease guide priorities**). On the other hand, there is the ethics of principles, which establishes four pillars on which current translational medicine should be based: **non-maleficence** (excessive testing, malpractice, etc.); **beneficence** (doing good for individuals and society); **autonomy** (people's freedom to make decisions and confidentiality); and **justice or equity** (equality in healthcare provision) [3].

Furthermore, Article 25 of the Universal Declaration of Human Rights [4] must be taken into account: "*everyone has the right to a standard of living adequate for the health and well-being of himself and of his family*"; as well as the principles of the World Health Organization [5]: "*Health care is a legal and ethical obligation of the State, which must ensure the availability of accessible, high-quality and sustainable health systems*"; and Article 14 of the Spanish Constitution [6], which establishes the principle of equality and equity before the law and prohibits discrimination.

The objective reality is that the development of an **orphan drug** for a **rare disease is not profitable for the pharmaceutical industry**. For this reason, governments offer economic incentives to promote research and commercialization in this field [2]. **Ninety-five percent of these diseases still lack an approved specific treatment, and globally, fewer than 10% of ultra-rare diseases have any active drug research program**, compared with common diseases and rare diseases with higher prevalence [7].

The reasons for this investment gap in these medicines may include investment based on market size—the number of patients [8]—which is not spread **across** millions of patients [9], as well as research **portfolios** that focus on a limited number of specific conditions—cancer, cardiovascular diseases, obesity, etc. [10].

In the absence of sufficient public and private funding, research groups working on rare diseases must seek interdisciplinary collaboration with other related groups in order to share resources. They can also turn to patient associations, which are increasingly concerned with securing funding for research into their own diseases [11–13].

In this context, the Advanced, Gene and Cell Therapies Group for the treatment of congenital coagulopathies, led by Dr. Antonio Liras, is pursuing therapeutic protocols for the treatment of coagulation factor V deficiency, an ultra-rare disease [14]. Cross-disciplinary and interdisciplinary collaboration with other research groups is essential, but the group also receives support and funding from the Association for Research and Cure of Factor V Deficiency [15]. Despite the many difficulties and the lack of public administration support, all of this has made it possible to conduct high-quality research, establishing cell therapy protocols [16]; studying mutations in patients [17]; establishing cell models with factor V deficiency through gene editing (CRISPR) and correcting this deficiency [18]; and developing factor V-deficient mouse models [19,20]. A proof-of-concept study of a gene therapy protocol using lentiviral vectors, which has significant potential for clinical translation, is currently undergoing editorial review for publication.

References

[1] Consorcio Internacional de Enfermedades Raras (IRDiRC). Disponible en: https://irdirc.org/.

[2] Orphanet. Disponible en: https://www.orpha.net/es/other-information/about_orphanet.

[3] Principios de bioética de Beauchamp y Childress (1979): Beneficencia, no maleficencia, justicia y autonomía. Disponible en: https://abimad.org/principios-de-etica-biomedica/.

[4] Declaración Universal de los Derechos Humanos. Art. 25. Disponible en:

https://www.unidosporlosderechoshumanos.es/course/lesson/articles-19-25/read-article-25.html

[5] Organización Mundial de la Salud. Disponible en: https://www.who.int/news-room/fact-sheets/detail/human-rights-and-health

[6] Constitución Española. Art. 14. Disponible en: https://www.boe.es/buscar/act.php?id=BOE-A-1978-31229

[7]. The Next Generation of Rare Disease Drug Policy: Ensuring Both Innovation and Affordability. Institute for Clinical and Economic Review. Disponible en: https://icer.org/wp-content/uploads/2022/04/ICER-White-Paper_The-Next-Generation-of-Rare-Disease-Drug-Policy_040722.pdf.

[8] Unlocking the full potential of rare disease drug development: exploring the not-for-profit sector's contributions to drug development and access. Front Pharmacol. 2024;15:1441807. doi: https://doi.org/10.3389/fphar.2024.1441807.

[9] Drug development for neglected ultra-rare diseases of no commercial interest: Challenges and opportunities. Drug Discov Today 2025;30(4):104346. doi: https://doi.org/10.1016/j.drudis.2025.104346.

[10] The Hard Truth About Rare Disease and Gene Therapy Drug Development. Applied Clinical Trials 2025;34(2).Disponible en: https://www.appliedclinicaltrialsonline.com/journals/applied-clinical-trials/applied-clinical-trials-04-01-2025.

11. Duchenne Parent Project España. Disponible en: https://www.duchenne-spain.org/investigacion-actual/proyectos-financiados/.

12. Fundación ELA (FUNDELA). Disponible en: https://www.somospacientes.com/noticias/al-dia/asociaciones/ayudas-investigacin-ela-fundela/.

13. Fundación Anemia de Fanconi. Disponible en: https://anemiadefanconi.org/.

14. Déficit de factor V de la coagulación. De la mutación a la "curación" mediante terapias avanzadas. An. R. Acad. Farm. 2025,91(1):17-44. Disponible en: https://analesranf.com/wp-content/uploads/2025/91_01/91_01.pdf.

15. Asociación para la Investigación y Cura del Déficit de Factor V (ASDEFAV). Disponible en: https://unaesperanzaparacelia.org/.

16. Cell therapy for factor V deficiency: An approach based on human decidua mesenchymal stem cells. Biomed. Pharmacother. 2021,142:112059. doi: https://doi.org/10.1016/j.biopha.2021.112059.

17. High Mutational Heterogeneity and New Mutations in the Human Coagulation Factor V Gene. Future Perspectives for Factor V Deficiency Using Recombinant and Advanced Therapies. Int. J. Mol. Sci. 2021,22:9705. doi: https://doi.org/10.3390/ijms22189705.

18. Development and Characterization of a Factor V-Deficient CRISPR Cell Model for the Correction of Mutations. Int. J. Mol. Sci. 2022,23:5802. doi: https://doi.org/10.3390/ijms23105802.

19. Development of a novel and viable knock-in factor V deficiency murine model: Utility for an ultra-rare disease. PLoS One 2025,20(6): e0321864. doi: https://doi.org/10.1371/journal.pone.0321864.

20. Effect of a truncated mutant factor V on hemostatic function and embryonic development in mice. Sci Rep. 2026,16(1):8460. doi: https://doi.org/10.1038/s41598-026-38387-w.

Antonio Liras

Professor of Biochemistry and Molecular Biology at the Complutense University of Madrid. He holds a degree in Chemical Sciences and a PhD in Pharmacy from the same university, and is a European Union Specialist in Clinical Analysis. He is a member of the Advisory Committee of the Spanish Federation of Rare Diseases (FEDER) and Director of the Research Group on Advanced, Gene and Cell Therapies for the Treatment of Congenital Coagulopathies. He also serves as Scientific Coordinator of the Association for Research and Cure of Factor V Deficiency.

He is a member of the editorial boards of several biomedical journals and an Academician of the Royal National Academy of Pharmacy of Spain. He has been principal investigator of numerous research projects in biomedical research and rare diseases and is the author of more than one hundred national and international publications, as well as several scientific and educational books and book chapters. He has been a speaker at numerous national and international conferences.

His extensive work in technology and knowledge transfer includes the exploitation of research results, patents, and contracts with pharmaceutical companies. He has also served as a reviewer and evaluator of research projects at both national and international levels. Among his achievements is the patent for the first viable mouse model of coagulation Factor V deficiency. He has received several research awards, including the ConfiHe Baxter Award, the Spanish Society of Thrombosis and Haemostasis Award, the Victoria Eugenia Royal Foundation Award, and the Royal National Academy of Pharmacy of Spain Award.