CEP Clin Exp Pediatr Vol. 67, No. 7, 356–357, 2024
https://doi.org/10.3345/cep.2023.01109

Editorial

Advancing orphan drug development for rare diseases

Jung Min Ko, MD, PhD[1,2]

[1]Division of Clinical Genetics, Department of Pediatrics, Seoul National University College of Medicine, Seoul, Korea; [2]Rare Disease Center, Seoul National University Hospital, Seoul, Korea

Key message

- Rare diseases present unique challenges and unmet needs for which the development of orphan drugs tailored to them offers hope.
- Despite the hurdles posed by limited patient populations, orphan drug designations from regulatory agencies provide incentives, such as extended market exclusivity and tax credits, that ignite transformative advances.
- Scientific progress in genomics, personalized medicine, and analytics empowers precise interventions by decoding genetic anomalies and encouraging effective treatments.

Rare diseases, which affect millions of individuals worldwide, present unique challenges and unmet medical needs. The development of orphan drugs specifically tailored to treat these conditions offers a beacon of hope for patients and their families. This review article[1] delves into the significance of orphan drugs and the multifaceted efforts required to bring relief to those living with rare diseases.

Orphan drug development is a complex journey riddled with obstacles. The limited affected patient population poses a financial hurdle for pharmaceutical companies, deterring traditional research and development pathways. Regulatory agencies recognize this conundrum and have introduced orphan drug designations to incentivize progress in this area.[2,3]

Orphan drug designation bestows a range of benefits upon developers, including extended market exclusivity, tax credits, and fee waivers. These incentives catalyze research and development, transforming the landscape for rare disease treatments. Collaborative endeavors among researchers, patient advocacy groups, and industry leaders have been instrumental in garnering support and resources.[3]

Patient advocacy groups wield a remarkable influence, propelling the development of orphan drugs through awareness campaigns, fundraising initiatives, and policy advocacy.[4] Collaborative networks that connect patients, clinicians, researchers, and regulators are vital to the sharing of knowledge and expertise, fostering a holistic approach to rare disease treatment.[5]

Advancements in genomics, personalized medicine, and analytics have revolutionized the landscape of rare disease research. With the power to decode genetic anomalies and tailor interventions to an individual's unique molecular profile, these breakthroughs hold promise for treatment precision and efficacy.[6]

The journey to orphan drug development extends beyond national borders. International collaboration and harmonization of regulatory processes expedite access to life-changing treatments for patients across different nations. Policymakers worldwide are implementing strategies to encourage orphan drug research and streamline approval processes.[7]

As we stand at the crossroads of scientific innovation, the future of orphan drug development appears brighter than ever. Emerging technologies, including gene editing and cell therapies, hold the potential for groundbreaking treatments. By embracing collaboration, sharing data, and aligning efforts globally, we can unlock a future in which rare disease patients no longer suffer in the shadows.[6]

In conclusion, the quest for orphan drugs embodies a ray of hope for individuals grappling with rare diseases. The convergence of scientific advancements, collaborative initiatives, and regulatory incentives paints a promising picture of improved outcomes and a brighter future. Clinical geneticists at the forefront of this endeavor play a pivotal role in shaping the landscape of rare disease treatment. Let us continue to relentlessly champion the cause, advocate for patients, and collaborate to ensure that no rare disease goes unnoticed or untreated.

See the article "Development of orphan drugs for rare disease" via https://doi.org/10.3345/cep.2023.00535.

Corresponding author: Jung Min Ko, MD, PhD. Department of Pediatrics, Division of Clinical Genetics, Seoul National University College of Medicine, 103 Daehak-ro, Jongno-gu, Seoul 03080, Korea
✉ Email: jmko@snu.ac.kr, https://orcid.org/0000-0002-0407-7828
Received: 17 August 2023, Accepted: 19 September 2023

Footnote

Conflicts of interest: No potential conflict of interest relevant to this article was reported.

Funding: This study received no specific grant from any funding agency in the public, commercial, or not-for-profit sectors.

ORCID
Jung Min Ko https://orcid.org/0000-0002-0407-7828

References

1. Yoo HW. Development of orphan drugs for rare disease. Clin Exp Pediatr 2023 Jun 28. doi: 10.3345/cep.2023.00535. [Epub].
2. Ferreira CR. The burden of rare diseases. Am J Med Genet A 2019;179:885-92.
3. Kimmel L, Conti RM, Volerman A, Chua KP. Pediatric orphan drug indications: 2010-2018. Pediatrics 2020;145:e20193128.
4. Dunkle M, Pines W, Saltonstall PL. Advocacy groups and their role in rare diseases research. Adv Exp Med Biol 2010;686:515-25.
5. Litterman NK, Rhee M, Swinney DC, Ekins S. Collaboration for rare disease drug discovery research. F1000Res 2014;3:261.
6. Miller KL, Fernaglich LJ, Maynard J. Using four decades of FDA orphan drug designations to describe trends in rare disease drug development: substantial growth seen in development of drugs for rare oncologic, neurologic, and pediatric-onset diseases. Orphanet J Rare Dis 2021;16:265.
7. Chan AYL, Chan VKY, Olsson S, Fan M, Jit M, Gong M, et al. Access and unmet needs of orphan drugs in 194 countries and 6 areas: a global policy review with content analysis. Value Health 2020;23:1580-91.

How to cite this article: Ko JM. Advancing orphan drug development for rare diseases. Clin Exp Pediatr 2024;67:356-7.