

Novartis Position on Medicines for Patients with Rare Diseases

At Novartis our mission is to reimagine medicine to improve and extend patients' lives. Using science-based innovation, we strive to deliver better outcomes for patients and address the evolving healthcare needs of society.

Rare diseases are defined as life-threatening or chronically debilitating conditions that affect only a small number of patients¹. While individually rare, the large number of 6000-8000² rare diseases means that up to 6%³ of the World population may be affected by a rare disease at some point in their lives.

Rare diseases pose unique challenges to patients, their families, society, healthcare providers, and healthcare systems. People are often unaware of individual rare diseases, physicians may be inexperienced in diagnosing and treating them, and patients may not always be able to get the treatments they need.

Medicines to treat rare diseases are called orphan medicines. Developing these products is challenging due to the severity of the diseases and the small number of patients available for clinical trials. When orphan medicines make it to market, they are often not commercially viable with such small patient populations. The added value of orphan medicines is often not adequately recognized, leading to reimbursement challenges. To address these obstacles and to foster development of orphan medicines, some countries have introduced specific regulatory and market incentives.

Since their inception, orphan incentives have helped bring many new products to market. Despite this progress most rare diseases still do not have adequate treatments. Many countries still do not have tailored regulatory and market access frameworks for orphan medicines, while others, such as the EU, are considering tightening the eligibility criteria for orphan incentives.

Novartis position

Novartis recognizes the high unmet need and is committed to bringing new medicines to rare disease patients. Based on a research approach that follows the science, we have developed many rare disease products, which has earned us the recognition of leading patient organizations.⁴ Novartis believes in four key principles to help address the specific needs of patients with rare diseases and to tackle the challenges of developing and providing treatments:

1. Creating a supportive policy framework for research, development, and commercialization
2. Partnering with patients and patient organizations to understand their needs and to incorporate their perspectives into the development process
3. Leveraging collaborations between the pharmaceutical industry and the academic, medical and patient communities
4. Providing access to treatments through tailored approaches that meet the needs of healthcare systems and patients

1. Creating supportive policy frameworks to address challenges in research, development, and commercialization

While science guides our research, Novartis believes that a favorable policy environment can support development and access to orphan medicines. An agile regulatory system that considers the challenges of small patient populations, such as difficulties in recruiting enough patients for clinical trials, can result in faster authorization of medicines. Specific orphan committees and regulatory fast track models have proven to be effective tools for bringing orphan medicines to approval. Market incentives, such as orphan exclusivity, help compensate for the small number of patients with a given rare disease. Receiving adequate reimbursement in line with the value a new medicine provides to patients, the healthcare system and society ensures healthcare innovation is sustainable in the long-term.

2. Partnering with patients and patient organizations to understand their needs and incorporate their perspectives into the development process

We are committed to actively listening to the patient community to inform decisions throughout the development and commercialization process of our medicines⁵. This collaboration enables us to design clinical programs that reflect patient-focused risk-benefit evaluations and the most relevant outcomes for patients, including through patient reported outcome measures. It also helps us to provide the most impactful therapies and design accompanying support services.

3. Leveraging collaborations between the pharmaceutical industry and the academic, medical and patient communities

Novartis recognizes the importance of working with multidisciplinary clinical expert teams to build on existing knowledge on rare diseases to accelerate research and development of new medicines and to facilitate optimal patient care. We believe in collaboration and partnership between for-profit and not-for-profit rare disease researchers in all phases of research and development and support enabling initiatives such as public-private-partnerships. To facilitate this exchange, teams across Novartis have established strong relations with external researchers, disease and technology experts, and patient communities around the world.

4. Providing access to treatments through tailored approaches that meet the needs of healthcare systems and patients

Novartis works with stakeholders to provide access to rare disease treatments. We believe that health technology assessment and re-imburement decisions should be based on a holistic approach that encompasses clinical, patient, healthcare system and societal value. Comprehensive stakeholder engagement with authorities, patients, healthcare providers and industry from early development is important to gain alignment on priorities and to understand the impact new orphan medicines will have on healthcare systems. Funding and delivery of rare disease treatments should be patient-centric, adequate for a country's health care system and allow for tailored solutions that reflect the value of these highly specialized products. It should also provide patients with rare diseases best chances of receiving the appropriate treatment for their disease.

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¹ The EU Orphan Regulation defines rare diseases as those affecting fewer than 5 in 10'000 Europeans. The US Orphan Drug Act defines a rare disease as a disease or condition affecting less than 200'000 people in the United States, or ~6 in 10'000.

² [Orphanet](#), a European rare disease resource contains information on 6172 unique rare diseases; the [US FDA](#) counts over 7000 rare diseases. (Both websites last accessed July 2022.)

³ Estimating cumulative point prevalence of rare diseases, Wakap, SN et al., Eur. J. Human Genetics, Vol 28; 2020

⁴ In recognition of our work for rare disease patients, Novartis received the 2018 EURORDIS Black Pearl Company Award for Innovation

⁵ [The Novartis Commitment to Patients and Caregivers](#)