

Novartis Position on Medicines for Patients with Rare Diseases

Novartis' mission is to discover new ways to improve and extend people's lives. Using science-based innovation, Novartis strives to deliver better outcomes for patients and to 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 patients in each countryⁱ. While individually rare, the large number of rare diseases, an estimated 6000-8000ⁱⁱ, means that up to 6–8%ⁱⁱⁱ of the world population may be affected.

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

Medicines to treat patients with rare diseases are called orphan medicines. Developing these medicines is particularly challenging due to the small number of patients and the severity of the diseases. Market mechanisms also fail due to the low number of patients. To address these challenges and to foster development of orphan medicines, some markets have introduced specific regulatory and market access frameworks.

Despite the progress that has been made in recent years, the vast majority of people living with rare diseases still have unmet medical needs today and many markets do not yet have tailored regulatory and market access frameworks for orphan medicines.

Novartis position

Based on a research approach that follows the science, Novartis has a long standing track record in developing medicines for rare diseases. Novartis believes in four key principles to address the specific needs of patients with rare diseases and to tackle the challenges of developing and providing treatments:

1. Creating supportive policy frameworks for research and development
2. Partnering with patients and patient organizations to understand patient needs and incorporate their perspectives in the development process
3. Leveraging research collaborations between the pharmaceutical industry and the scientific, medical and patient communities
4. Providing access to treatments for people living with rare diseases through tailored approaches that meet the specific needs of the patient population

1. Creating supportive policy frameworks for research and development

Novartis believes that the creation of favorable policy environments is essential to address the many challenges related to rare diseases. We closely collaborate with governments, policy makers, regulators and patient representatives to create policy frameworks that foster research and development of orphan medicines, and compensate for failure of common market incentives. We also work towards regulatory systems that take into account the unique challenges of small patient populations such as difficulties to recruit sufficient patients for clinical trials. Specific orphan committees, regulatory fast track models and market exclusivity have proven to be effective tools to bring orphan medicines to market and compensate for the small number of patients with a given rare disease.

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

We are committed to actively listen to the patient community to inform decisions throughout the development and commercialization process of our medicines^{iv}. This collaboration enables us to design clinical programs that reflect patient-focused benefit- risk 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 research collaborations between the pharmaceutical industry and the scientific, medical and patient communities

Novartis recognizes the importance of working with multidisciplinary clinical expert teams to build on existing knowledge of rare diseases in order 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 support enabling initiatives such as public-private-partnerships. To facilitate this exchange, the Novartis Institute for Biomedical Research (NIBR) has established strong relations with external researchers and disease and technology experts around the world.

4. Providing access to treatments for people living with rare diseases through tailored approaches that meet the specific needs of the patient population

Novartis works with stakeholders to provide access to rare disease treatments. We believe that health technology assessment and reimbursement decisions should be based on a holistic approach that encompasses clinical, patient, healthcare system and societal value. Comprehensive dialogue between authorities, patients, healthcare providers and industry from early development onwards is important to gain alignment on priorities and to understand the impact of introducing new orphan medicines to the healthcare system. Funding and delivery of rare disease treatments should be patient-centric as well as adequate for a country's health care system. It should also provide patients with rare diseases best chances of receiving appropriate therapy for their disease.

Last updated September 2017

ⁱ In the EU, rare diseases are defined as those affecting fewer than 1 in 2.000. In the US, a disease is defined as rare if it affects fewer than 200.000 Americans at any given time ([Rare Disease Day 2018 - Article](#)).

ⁱⁱ [Rare diseases – IFPMA](#).

ⁱⁱⁱ Orphan drugs: the regulatory environment, Franco, P. *Drug Discovery Today*, Vol 18; 2013.

^{iv} [The Novartis Declaration for Patients](#).