Rare Diseases at FDA

Over 7,000 rare diseases affect more than 30 million people in the United States. Many rare conditions are life-threatening and most do not have treatments. Drug, biologic, and device development in rare diseases is challenging for many reasons, including the complex biology and the lack of understanding of the natural history of many rare diseases. The inherently small population of patients with a rare disease can also make conducting clinical trials difficult. Since the Orphan Drug Act was signed into law in 1983, the FDA has approved hundreds of drugs for rare diseases, but most rare diseases do not have FDA-approved treatments. The FDA works with many people and groups, such as patients, caregivers, and drug and device manufactures, to support rare disease product development.

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What is a rare disease?

The Orphan Drug Act (/industry/designating-orphan-product-drugs-and-biological-products/orphan-drug-act-relevant-excerpts) defines a rare disease as a disease or condition that affects less than 200,000 people in the United States.

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What is the Orphan Drug Act?

The Orphan Drug Act is a law passed by Congress in 1983 that incentivizes the development of drugs to treat rare diseases.
Companies and other drug developers can request orphan drug designation and FDA will grant such designation if the drug meets specific criteria. Orphan designation qualifies sponsors for various incentives, including:

- Tax credits for qualified clinical (in humans) testing
- Waiver of the Prescription Drug User Fee (currently at almost $3 million for a new drug)
- Potential 7 years of market exclusivity after approval

In addition, the Orphan Drug Act established the Orphan Product Grants Program (https://www.fda.gov/industry/developing-products-rare-diseases-conditions/orphan-products-grants-program) to provide funding for developing products for rare diseases or conditions.

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What is an orphan drug?

An orphan drug is a drug for a rare disease or condition. Some rare disease treatments have been "orphaned" or discontinued because there was not enough financial incentive to continue development or production. The Orphan Drug Act incentivizes drug development for rare diseases.

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How does the FDA encourage the development of medical products to diagnose and treat rare diseases?

The FDA works with stakeholders, including patients, patient advocates, product developers, and researchers, to support the development of safe and effective drugs, biologics, and devices for rare diseases.

The FDA is one part of the many parts involved in finding and developing treatments for rare diseases. Specifically, the FDA:

Administers Laws and Regulations

- Carries out the Orphan Drug Act (/about-fda/page-not-found) and related laws and regulations

Reviews and Grants Designations to Rare Disease Drugs, Rare Pediatric Diseases, and Devices

- Evaluates whether a drug or device qualifies for designation, based on whether both the product and the disease must meet certain criteria specified in the relevant laws. Relevant designation programs for rare diseases include the orphan drug (/industry/medical-products-rare-diseases-and-conditions/designating-orphan-product-drugs-and-biological-products), rare pediatric disease (/industry/medical-products-rare-diseases-and-conditions/rare-pediatric-disease-rpd-designation-and-voucher-programs), and humanitarian use device (/industry/medical-products-rare-diseases-and-conditions/humanitarian-use-device-hud-designation-program) designation programs. Also, FDA reviews and awards Rare Pediatric Disease Priority Review Vouchers (/industry/medical-products-rare-diseases-and-conditions/rare-pediatric-disease-rpd-designation-and-voucher-programs).

Regulates Drugs and Devices

- Protects the public health by ensuring the safety, efficacy, and security of drugs, biological products, and medical devices

Gives Research Grants

- Awards grants for research on rare diseases, including grants for clinical trials, natural history studies, and pediatric device consortium

Conducts Outreach

- Communicates with professional organizations, patients, and rare disease advocacy groups about rare disease issues

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What does the FDA Office of Orphan Products Development do?

The FDA's Office of Orphan Products Development (/about-fda/office-clinical-policy-and-programs/office-orphan-products-development):

- Coordinates FDA activities for rare diseases
- Administers the orphan drug, rare pediatric disease, and humanitarian use device designation programs
- Funds grants and cooperative agreements for rare disease research
- Is a central point-of-contact for patients, caregivers, and advocacy groups who have questions about the FDA's work in rare diseases

Learn more about the FDA Office of Orphan Products Development (/about-fda/office-clinical-policy-and-programs/office-orphan-products-development).

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How do the FDA medical product centers support rare disease product development?

The FDA Center for Drug Research and Evaluation (CDER), Center for Biologics Evaluation and Research (CBER), and Center for Device and Radiological Health (CDRH) all support the development and approval of safe and effective drugs, biologics, and devices to treat rare diseases.

To address specific considerations in developing and approving medical products for rare diseases, the FDA centers:

- Conduct specialized training for FDA staff on rare disease topics
- Issue guidances for industry to encourage medical product development in rare diseases
- Host and participate in patient-focused meetings and listening sessions on rare disease topics
- Award research grants, cooperative agreements, and contracts in addition to conducting pilot programs and special data analyses to advance the regulatory science for rare diseases
- Administer the expanded access or compassionate use program for investigational medical products in their specific area
- Participate in FDA's Rare Disease Council to discuss rare disease issues
- Collaborate on FDA's annual Rare Disease Day event

Read more about CDER's Rare Diseases Program (/about-fda/center-drug-evaluation-and-research-cder/rare-diseases-team) and Accelerating Rare disease Cures (ARC) Program (https://www.fda.gov/about-fda/center-drug-evaluation-and-research-cder/cders-arc-program)

Read more about CBER's Rare Disease Program (/vaccines-blood-biologics/resources-you-biologics/select-cber-projects-and-activities).

Read more about CDRH's Breakthrough Devices Program (/medical-devices/how-study-and-market-your-device/breakthrough-devices-program) and the Humanitarian Use Device (/industry/medical-products-rare-diseases-and-conditions/humanitarian-use-device-hud-designation-program) and Humanitarian Device Exemption (/medical-devices/premarket-submissions-selecting-and-preparing-correct-submission/humanitarian-device-exemption) programs.

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How can patients become involved in FDA's work on rare diseases?

The FDA's Patient Affairs Staff partners with the National Organization for Rare Disorders (NORD) to host Patient Listening Sessions (/patients/learn-about-fda-patient-engagement/fda-patient-listening-sessions) that focus on rare diseases. Patient Listening Sessions are one of many ways the patient community can share their experience with a disease or condition by talking directly with FDA staff.

We encourage rare disease patients and caregivers to contact (mailto:PatientAffairs@fda.hhs.gov?subject=Rare%20Disease%20Patient%20Affairs%20question)our Patient Affairs Staff (/about-fda/office-clinical-policy-and-programs/patient-affairs-staff) with any questions they might

have about the FDA, treatments for their disease, and how they can participate in FDA processes. Patients or caregivers may also request a meeting by using the Patient Portal at www.fda.gov/PatientsAskFDA (http://www.fda.gov/PatientsAskFDA).

Learn more about the FDA's Patient Engagement work (/patients/learn-about-fda-patient-engagement).

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