CERTARA

The Tools You Need to Accelerate Rare Disease Drug Development

Patients with rare diseases have high unmet medical needs.

Over

7,000

Rare Diseases Identified

Only

5%

of Rare Diseases Have a Global Approved Drug

In 2024, CDER approved 50 novel drugs, but significant unmet needs persist worldwide.

What makes something a "rare or a neglected disease"? There is no universally accepted definition as the prevalence criteria varies by region/country.

Regulatory Agency	Prevalence Criteria	Reference
U.S. FDA	200,000	Orphan Drug Act, 1983
EMA	5/10,000	EC 141/2000
Japan PMDA	50,000	JPMA, 2008
Australia	2000	Therapeutic Goods Act, 1989

Rare Disease Affects More Than

300 M

People Worldwide

Countries worldwide have implemented various mechanisms to encourage the development of treatments for diseases

50%

of People Affected By Rare Diseases Are Children

APPROXIMATELY

30%

of Children With These Debilitating Diseases Will Not Live To See Their 5th Birthday

APPROXIMATELY

80%

Of Rare Diseases Are Genetic

While rare disease drug development is challenging, the experts at Certara can help!

Small patient pools

We use clinical pharmacology expertise in combination with MIDD tools to discern effects in small patient populations.

Sensitive patient populations

We model the impact of a new drug on other disease states or untested populations.

Unique regulatory landscape

We provide end-to-end regulatory support and medical writing services: from meetings with health authorities to preparation and submission of marketing applications.

Reimbursement challenges

We develop a quantitative multi-criteria decision analysis-based framework adapted to the very specific issues in rare disease development and access including quality of evidence, disease severity, ethical considerations, population-level factors, economic impact of the disease, and specific budgetary impact.

We have broad therapeutic expertise in rare disease drug development

Genetic & Hereditary

- Fabry Disease
- Pompe Disease
- Hunter's Disease
- Metachromatic Leukodystrophy Disease
- Transthyretin-mediated Amyloidosis
- Urea Cycle Disorder
- Hereditary Angiodema
- ACE-induced HAE
- Lennox-Gastaut Syndrome
- Familiar Mediterranean Fever
- Gaucher Disease
- Cystic Fibrosis
- Hypophosphatasia
- Chronic Liver Disease
- Huntington's Disease
- Dravet Syndrome
- Thrombocytopenia
- Hereditary Transthyretin-mediated Amyloidosis

Antinfectives/Antivirals

- Anthrax
- Orthopoxvirus/Smallpox
- Symptomatic Botulism
- Respiratory Syncytial Virus

Gastrointestinal

- Short Bowel Syndrome
- Pediatric Crohn's Disease
- Carcinoid Syndrome

Oncology

- Acute Lymphoblastic Leukemia
- Metastatic Myeloma
- Multiple Myeloma
- Acute Myeloid Leukemia
- Urinary Bladder Cancer (Photodynamic Therapy)
- BRAF Mutation
- Blood Cancer
- Ovarian Cancer

CNS

- Myasthenia Gravis
- Restless Leg Syndrome
- Spasticity due to Multiple Sclerosis
- PKAN (Pantothenate Kinase-associated Neurodegeneration)
- Friedreich's Ataxia

Blood Disorders

- Paroxysmal Nocturnal Hemoglobinuria
- Atypical Haemolytic Uremic Syndrome (aHUS)
- Beta-thalassemia (Sickle Cell Beta-thalassemia)

Bone & Muscle Disease

- X-linked Hypophosphatemia
- Duchenne Muscular Dystrophy
- Hypophosphatasia

Other

- Radiation Poisoning
- Hypoparathyroidism
- Molybdenum Deficiency
- Peyronie's Disease
- Hyperuricemia in Patients with Gout Refractory
- Hepatic Veno-occlusive Disease
- Onchocerciasis
- Neglected Tropical Diseases

We have partnered with biopharmaceutical companies of all sizes on drug development plans for a wide range of rare/orphan diseases.

Learn more about our unparalleled experience in Orphan Drug Development: certara.com/services/rare-disease-and-orphan-drug-development-resource-center/

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