



RARE AID: A PLATFORM TO SUPPORT PEOPLE WITH RARE DISEASES

About the project and rare disease trends in Russia

Rare Aid is a platform that supports people with rare (orphan) diseases. It aggregates useful information about diseases and treatments and promotes services for healthcare professionals (HCPs), patients, charities, and pharmaceutical manufacturers. The project was **launched in 2021**.



Educational events and a growing library of video lectures

4 online conferences
and over 100 videos for HCPs

Clear information on diseases

300+ articles and interviews
for patients

7000+

HCPs registered for 2.5 years

Help with finding and accessing care

Catalogues of charitable foundations, HCPs, medical institutions, interactive legal guides

Developing a knowledge base

Collecting scientific papers, translating key articles, developing a diagnostic reference book on rare diseases

Any questions or ideas about working together?



E-mail us at

info@rare-aid.com

One of the portal's goals is to aggregate major rare disease events and to share this information with a wider audience

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What diseases are considered rare (orphan) in Russia?



In Russia, rare diseases are defined as diseases with a prevalence of ≤ 10 cases per 100,000 population.¹

How many patients with rare diseases could be in Russia?



2+ mln people² representing 1.4% of the population

1,300+ patients

with spinal muscular atrophy (according to the SMA Family Foundation for 2023)

3,500+ patients

with cystic fibrosis according to the Research Institute of Pulmonology of the Federal Medical-Biological Agency of Russia and the Circle of Kindness Foundation

677 patients

with acromegaly and pituitary gigantism treated using budgeted funds in 2021

22 patients

with Pompe disease (glycogen storage disease type II) treated at the Circle of Kindness Foundation's expense in 2022

2,500 patients

with epidermolysis bullosa according to the Butterfly Children Charity

4,600+ patients

with primary immunodeficiencies according to the Registry of Patients with PIDs of the National Association of Primary Immunodeficiency Experts

291 patients

with Duchenne muscular dystrophy treated at the Circle of Kindness Foundation's expense in 2022

32 patients

with hypophosphatasia treated at the Circle of Kindness Foundation's expense in 2022

1. Federal law "On the Fundamentals of Protecting the Health of Citizens in the Russian Federation" No. 323-FZ, Part 1, Article 44.

2. Natalya S. Volkova, Elvina Aksu. Rare (Orphan) Diseases: Legal Regulation in Russia and Abroad [Redkiye (orfannyye) zabolevaniya: pravovoye regulirovaniye v Rossii i za rubezhom] // Journal of Foreign Legislation and Comparative Law [Zhurnal zarubezhnogo zakonodatel'stva i sravnitel'nogo pravovedeniya]. 2018. No. 4 (71). URL: <https://cyberleninka.ru/article/n/redkie-orfannye-zabolevaniya-pravovoe-regulirovanie-v-rossii-i-za-rubezhom> (Accessed 31 October 2023).

How are rare diseases diagnosed in Russia?

Newborn children are screened for many rare diseases as part of [a national screening program](#). Since 2006, every newborn in Russia has been screened for 5 diseases, and by 2023, dozens of additional diseases have been included in this program.¹ The newborn screening program has been expanded because many rare diseases have effective therapies and therefore require early detection, including at pre-symptomatic stages. All children born in the country are screened for free of charge.

2006 through 2023

5 conditions

- + Phenylketonuria
- + Congenital hypothyroidism
- + Galactosemia
- + Adrenogenital syndrome
- + Cystic fibrosis

Since 1 January 2023

40+ conditions

- + Classic phenylketonuria Phenylketonuria B
- + Congenital hypothyroidism
- + Cystic fibrosis
- + Galactose metabolism disorder (galactosemia)
- + Adrenogenital syndrome
- + Adrenogenital disorders associated with enzyme deficiency
- + Biotinidase deficiency
- + Other types of hyperphenylalaninemia (biopterin synthesis deficiency, tetrahydrobiopterin deficiency)
- + Tyrosinemia
- + Maple syrup urine disease
- + Propionic acidemia

1. Order of the Ministry of Health of the Russian Federation N 274n "On Approval of the Procedure for Providing Medical Care to Patients with Congenital and/or Hereditary Diseases" dated 21 April 2022.

How are rare diseases diagnosed in Russia?

Russia has several **selective screening programs** in addition to the mass newborn screening program for rare diseases. They are medically indicated for high-risk patients.

The following diseases are detected by such programs:

- + Hypophosphatasia
- + Fabry disease (alpha-galactosidase-A deficiency)
- + Gaucher disease (cerebroside lipidosis syndrom)
- + Mucopolysaccharidoses
- + and other rare conditions

What national programs support people with rare diseases?



PROGRAM	14 HIGH COST CONDITIONS	LIST OF RARE LIFE-THREATENING CONDITIONS	THE CIRCLE OF KINDNESS FOUNDATION	DISABILITY BENEFITS*
ORPHAN DISEASES OR THEIR GROUPS	11 conditions hemophilia, cystic fibrosis, pituitary dwarfism, Gaucher disease, malignant neoplasms of lymphoid, hematopoietic, and related tissues, juvenile arthritis with systemic onset, mucopolysaccharidosis etc.	17 conditions Paroxysmal nocturnal hemoglobinuria, idiopathic thrombocytopenic purpura, tyrosinemia, homocystinuria, galactosemia, Fabry disease, Niemann-Pick disease (acid sphingomyelinase deficiency), osteogenesis imperfecta etc.	88 conditions Spinal muscular atrophy, Duchenne and Becker muscular dystrophy, hypophosphatasia, neuroblastoma, tuberous sclerosis, neurofibromatosis, epidermolysis bullosa etc., as well as children aged <19 years with diseases included into the 14 High Cost Conditions program.	10+ conditions Hepatocerebral dystrophy, phenylketonuria, acute intermittent porphyria, cancer, hematological diseases, cytopenia, Addison's disease (primary adrenal insufficiency), precocious puberty etc.
FUNDING	Federal Budget of the Russian Federation	Regional Budget of the Russian Federation	Federal Budget of the Russian Federation	Regional Budget of the Russian Federation
NUMBER OF PATIENTS RECEIVING CARE	26,000 patients	15,000 patients	23,000 pediatric patients	Over 80,000 patients**
AGE GROUP	Children and adults	Children and adults	Children and patients aged <19 years	Children and adults

* Decree of the Government of the Russian Federation N 890 dated 30 July 1994
** Estimated number of patients with rare disorders (1.4%) of the number of people with established group I & II disability according to Federal State Statistics Service of Russia (Rosstat) for 2023 (5.8 mln people).

How do charities help people with rare diseases?

In Russia, there are more than **10,000** charitable foundations. Hundreds of them help people with rare diseases. They typically provide targeted financial aid to families in need, such as covering the cost of diagnostic procedures not covered by the state budget and providing funds for travel to the treatment location. Many foundations develop systematic programs that help patients find doctors and receive psychological and legal assistance. Many foundations have programs for HCPs who are interested to improve their skills in diagnosing and treating a specific rare disease.

The Circle of Kindness

In January 2021, to support patients with rare diseases, the President of the Russian Federation issued an order creating the Circle of Kindness Foundation to support children with severe and rare (orphan) diseases. The Circle of Kindness is funded by increased income tax for Russian citizens with an annual income exceeding **5 mln rubles**.

23,000+

pediatric patients who have received or will receive care based on approved applications (as of end of 2023).

210 bln rubles

This is the amount of contracts and agreements concluded to provide medical care and to purchase drug products and medical devices from 2021 to 2023.

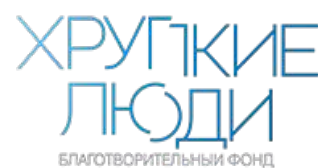


circleofkindness.ru

This is only a small part of the Russian charitable organizations and patient associations that provide support to people with rare diseases:



Genodermatoses



Osteogenesis imperfecta



Amyotrophic lateral sclerosis



Severe diseases



Primary immunodeficiencies



Leukemia



Duchenne muscular dystrophy



Spinal muscular atrophy



Palliative care



Rare liver diseases



Cystic fibrosis



Hemophilia, bleeding disorders



Childhood cancer



Childhood leukemia



Severe diseases



Duchenne muscular dystrophy



Severe diseases



Childhood leukemia, childhood cancer



Cancer in children and adults



Neuroblastoma



Rett syndrome (cerebroatrophic hyperammonemia), neuronal ceroid lipod



Short bowel syndrome



Deafblindness



Severe diseases



Cystic fibrosis



Hypophosphatemic rickets



Spina bifida



Huntington's disease (chronic progressive chorea)



Acromegaly



Mucopolysaccharidosis

+ >100 rare disease foundations

Initiators of the Rare Aid portal and information partners of the project

The Rare Aid information and education project was developed with the support of Pharmimex.



- + Pharmimex JSC is one of Russia's largest suppliers of pharmaceuticals and medical devices.
- + Pharmimex was the first Russian pharmaceutical company to supply drugs for the treatment of orphan diseases including hemophilia, cystic fibrosis, Gaucher disease, Fabry disease, spinal muscular atrophy, and Duchenne muscular dystrophy. The company also supplies drugs for the treatment of various oncological, hematological, and other diseases.
- + Pharmimex, together with other participants, has initiated the creation of a modern production of human blood plasma drugs in Russia by signing a special investment agreement to build a plant, a plasma storage facility, and 10 modern plasma centers for obtaining plasma for fractionation as a raw material for manufacturing human blood products.

Skopinpharm is an information partner of the project.



- + Skopinpharm LLC is one of the leading distributors of orphan drugs in Russia.
- + The company participates actively in the national program '14 High Cost Conditions,' which offers free medication to patients with the most serious and rare diseases.
- + In 2021-2022, Skopinpharm LLC, in collaboration with the Academician N.P. Bochkov Medical and Genetic Scientific Center and Novartis Pharma LLC, implemented a Neonatal Screening for Early Detection of Patients with Spinal Muscular Atrophy and Primary Immunodeficiencies in the Russian Federation project.
- + In 2023, Skopinpharm supported launching a social "+Ya" (Eng.: And Me) project aimed at donating blood and plasma.

Rare Aid Information and Education Portal



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