VII. Objectives of the Federation of Rare Diseases Patients in Central and Eastern Europe

The Federation of Rare Diseases Patients in Central and Eastern Europe was established on 25 July 2010 in Poland.

The organisation objectives are as follows:

- to integrate families of the MPS and rare diseases patients in Central and Eastern Europe,
- to draw attention of the public to problems of the MPS and rare diseases patients,
- to establish funds in order to provide assistance to all persons affected by a rare disease.

JAK?

- through establishment of contacts by families with children suffering from MPS and rare diseases,
- through annual conferences during which parents and doctors can meet and obtain necessary information,
- through publishing, in the press, radio, television and websites, information addressed to all interested in problems of the MPS and rare diseases patients,
- through facilitating dissemination of medical knowledge and techniques in the area of MPS and other rare diseases all over the world,
- through attracting sponsors and donors whose donations make it possible to help children suffering from MPS and rare diseases and their families,
- through improving quality of life of the MPS and other rare disease patients in all countries of Central and Eastern Europe,
- through common organisation of meetings and events aimed at development and promotion of new methods of rare diseases treatment,
- through putting pressure on a given state to ensure treatment and specialist medical care to rare diseases patients,
- through stimulating scientific researches and cooperation of experts in the area of rare disease in individual countries and all over the world,
- through development of an international network of cooperation with MPS associations all over the world.

Don't wait – You can help us, too
– support those who need it.

MPS VI Lithuania

MPS VI Lithuania

Where can you obtain medical advice concerning mucopolysaccharidosis and other rare diseases?

You can get information on diagnostics, treatment and medical care from the members of the Board of the Federation of Rare Diseases Patients' in Central and Eastern Europe

Founding Committee for the federation of patients from Central and Eastern Europe (Belarus, Lithuania, Russia, Poland and Ukraine)

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Contact us and tell how You may contribute!

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Organisation of Rare Diseases Patients in Central and Eastern Europe registered in the Piaseczno county office in Piaseczno, Poland

Faith, hope and love today
– care, help and concern tomorrow.

Federation of Rare Diseases Patients' in Central and Eastern Europe

Federation of Rare Diseases Patients' in Central and Eastern Europe

I. Why do MPS and ultra rare diseases patients are unable to grow in a normal way?

The reason for that is **mucopolysaccharidoses (MPS)** and other rare diseases posing threats to their lives, since they result in a development of a large number of both physical and psychical defects in the patient bodies!

These diseases unfold and develop – help us stop them!

II. What are the reasons underlying those diseases?

Diseases called mucopolysaccharidoses entail inborn errors of metabolism. „Muco" means mucus, „poly" means many, whereas saccharides are sugar molecules.

Mucopolysaccharidoses are long chains of sugar carbohydrates which take part in the formation of connective tissue in the body (cartilages, sinews, skin etc). They are broken down by certain enzymes into parts and then produced again. There are at least six organic compounds, out of those produced by a living cell, necessary to achieve this. Ultra rare diseases patients lack certain enzyme, which means that the whole metabolism chain gets interrupted. As a consequence, sugars or lipids accumulate in all types of the connective tissue which in turn gets gradually destroyed.

A single person is unable to help gravely ill children – together we can save their lives!

III. Why does it happen?

Ultra rare diseases are inherited from healthy parents who are unaware of their genetic disorder. If both parents have gene errors, then it is probable that their children will be affected by this congenital disorder. It is only MPS II – Hunter syndrome – where solely mother is the disease carrier.

MPS is short for the disease, as lives of those children is significantly shortened.

MPS II Russia

MPS III Poland

MPS VI Belarus

MPS IV A Ukraine

MPS IV
Sakha Republic, Russia

MPS VI Lithuania

IV. What happens next?

The tragedy of rare disease patients lies in their regress, that is a reverse development in both physical and psychic terms. Such persons are born as normal, healthy children, however, some time later the disease manifests itself causing extremely dramatic changes which make beloved children hard to be accepted by others as they grow. In most cases, they die before coming of age.

Faith, hope and love today – care, help and concern tomorrow.

V. How do we recognise MPS and ultra rare diseases?

There are 7 major MPS types in Eastern Europe:

MPS type I – Hurler-Scheie syndrome
MPS type II – Hunter syndrome
MPS type III – Sanfilippo syndrome
MPS type IV – Morquio syndrome
MPS type VI – Maroteaux-Lamy syndrome
MPS type VII – Sly syndrome

The organisation takes also care of the ultra rare metabolic diseases patients, **inter alia lysosomal storage diseases: Mannosidosis, Sialidosis, Fucosidosis, Pompe disease, Nieman-Pick disease, Gaucher disease, Fabry disease, Epidermolysis Bullosa (EB), Krabbe syndrome, Gangliosidosis, Ceroidolipofuscynosis, Pallister-Kilian syndrome, Homocystinuria, Hallervorden – Spatz NBIA HSS syndrome, Metachromatic Leukodystrophy (sulphaditosis) etc.** MPS and ultra rare diseases are identified and diagnosed on the basis of detailed clinical tests. The most characteristic MPS symptoms include: thickened face features, joint contractures, bone changes, hepatosplenomegaly. Some disease types involve also corneal opacification, hearing impairment, heart changes, lower height and developing mental disability.

Treatment, rehabilitation and specialist medical care are extremely expensive, but human lives, children's ones in particular, are invaluable!!!

VI. Can the above mentioned diseases be cured?

Drugs registered in the European Union for individual ultra rare diseases

Disease name	Deficient enzyme	Drug name	Date of registration in the EU
Gaucher	beta-glucocerebrosidase	Cerezyme®	08.07.2002
	beta-glucocerebrosidase	Vpriv®	08.2010
Fabry	alpha-galactoside A	Fabrazyme®	10.01.2003
	alpha-galactoside B	Replagal®	2003
Hurler type (MPS I)	alpha-L-iduronidases	Aldurazyme®	10.06.2003
Pompe syndrome (glycogenosis II)	alpha-glycosidasis	Myozyme®	29.03.2006
Maroteaux-Lamy syndrome (MPS VI)	arylsulfatase B	Naglazyme®	01.2006
Hunter syndrome (MPS II)	liduronidase-sulfatase	Elaprase®	08.01.2007
Mannosidosis	alpha-mannosidosis		clinical trials
Morquio syndrome (MPS IV A)	galactose-6-sulfatase		clinical trials

Also in the case of other diseases, there are intense clinical trials taking place all over the world with the aim of discovering an effective method for introduction into the patient's body of the enzyme he or she is unable to produce. Every family having an ill child should be able to use health services provided by metabolic disease clinics where it could get genetic advice, use pre-natal tests, allowing to identify the disease already in the first months of the pregnancy, as well as to determine which members of the family are carriers.

Treatment is possible provided we succeed in collecting funds.

Only together are we able to save their lives!
We can help those without treatment to live in dignity, with no pain and suffering!