
LIFE
with
CHALLENGES

Supported by

PAL AWARDS Patient Advocacy Leadership Awards

genzyme
A SANOFI COMPANY

Supported by

UNIVERSITY PEDIATRIC CLINIC

FACULTY of MEDICINE

CONCEPT
marketing communications

**The Association of Citizens with a Rare Disease "Life with Challenges" was founded in 2009 by parents and patients with rare diseases.**

## Objectives and Activities of the Association

- Raising public awareness of rare diseases, diagnosis, treatment and rehabilitation;
- Human rights protection;
- Help and support for development of rare disease research;
- Helping, supporting, unifying and informing people on rare diseases and their adaptation in society;
- Contacting and cooperating with similar organizations in the country and abroad;
- Contacting and maintaining contacts between the Association and institutions in Macedonia (Ministry of Health, Health Insurance Fund and Ministry of Labor and Social Policy);
- Taking measures for coping with emotional consequences of the disease;
- Raising funds for treatment and support of patients with rare diseases

# RARE DISEASE GUIDE IN 5 STEPS

## 1 • What is a rare disease?

A rare disease is a condition affecting only a small per cent of the world population.

## 2 Rare diseases in numbers

**1**

person in every 2,000 affected in the world.

**350.000.000**

people in the world affected by a rare disease

**7.000**

various types of rare diseases in the world

**5%**

of existing rare diseases are adequately treated

## 3 • How to discover a rare disease?

It is always difficult to diagnose a rare disease although an early diagnosis is very important in order to start an early treatment.

## 4 • Are there symptoms and when do they occur?

Rare diseases have several common properties although they significantly differ from one another. In large number of rare diseases diagnosed

- first symptoms occur at an early age or immediately after birth

- diseases are mainly genetic

- diseases cause permanent disorders

## 5 • What if I have a rare disease? Is there any medication?

Although the percentage of existing rare diseases that can be cured, i.e. treated, is very low, there are, however, certain types of such diseases that can be fully eradicated. Even in Macedonia there are cases of patients who, having been treated, lead completely normal lives, and even have their own families.

The first symptoms of rare diseases occur in childhood

**Message from patients:**

One donation has changed our life! Fight: no matter how rare we are, nobody is alone in this world! The day shall come when life will lend you a helping hand!

**Vesna Stojmirova,**
**Bitola**
(President of the Rare Disease Association)

- a patient suffering from Gaucher, a rare disease

# When a hug "gives" you a broken rib...

Living with a rare disease means living with limitations, although you do not want to admit that. I was diagnosed with the rare disease Gaucher when I was a child at the age of 7 and I had literally no idea what that meant. I discovered gradually, through daily occurrence of strong stomach pains and bruises all over my body. Then, anemia began, loss of energy, weak immune system, easily-breakable bones and frequent visits to a doctor. Fortunately, I have parents who supported me and are still by my side every time I need that.

It all began with an occurrence of the first pains, which became more intense over the years. I started to adapt, thinking that it was simply my life and that nothing else could

be done. When I was a teenager I felt like an old woman.

The loss of energy reduced my going and hanging out, but even then, I always tried to hide the fact that there I was, the sick one. I was ashamed of that and did not want any pity. I only wanted to be just like my peers. When I told my friends for the first time that I suffered from Gaucher, they thought that I was joking...Still, they have not treated me any differently until today, they make me feel normal, which I actually am, aren't I?
One day I woke up and told myself that it was time I did something for myself. There can be no victory without a fight! Within 4 years I had 4 cracked ribs and excruciating pain.

I began to contact associations in Europe, Israel and America. I met plenty of people and eventually got in touch with Genzyme, the company that produces the medication. The company provided a donation for my treatment and has been sending medications for three years now at the pediatric Clinic in Skopje, where I have the liquid that changed my life administered. It is as if I got a second chance. I no longer have any physical difficulties and I am truly a lot happier. It seems to me that I can do whatever I want to. I am 28 and - having the complete support from my friends and my future husband - I feel healthy and filled with life. I recommend to all patients and parents facing a rare disease not to lose hope. It is the best piece of advice I can give you. If you become depressed and think you can do nothing to improve your life, it is the same as if you are giving up hope. Don't be afraid; ask for help, as nobody is alone in this world. Sometimes, you might find support where you least expect it. Life is not only about survival, but about making something special, and we are all special in our own way!

**Ljuljzim Sali,**
**Tetovo**

- a patient suffering from Gaucher, a rare disease

## It is hard to be a child who can easily hurt himself while playing a simple child's play!

It may sound ironic, but laughter is not a sin: I am 36 years old and I still visit the Pediatric Clinic to be treated, just like when I was a child, and I argue with nurses about what type of needle and which arm to use for the infusion ...
This is the story that my life told me and I am telling it to you now.
I suffer from a rare disease, Gaucher, diagnosed when I was 10. It was then that my family and I realized why a simple child's play had ended with broken bones and bad health when I was younger. Almost no year would pass by without me having a fracture. The most frequent symptoms of the rare disease I suffer from are precisely the weak bones that result from loss of their thickness; frequent fatigue, anemia...it is hard to be a kid

that can easily get hurt from a simple kid's joy!

At the time when I was diagnosed by Nikola Zisovski, MD, along with Sofijanka Glamochanin, MD and Olivera Muratovska, MD, there was no treatment or medication and I only went to hospital to be examined, each year witnessing the merciless progress of the disease. After a while, my spleen enlarged to the extent that I had to be operated, having the spleen completely removed. Due to the lack of treatment, I have lived without a spleen for a second decade now.

In spite of all the hardships I suffered ever since the earliest childhood, I never said "I cannot take this anymore"! One fine day life gave me a helping hand. Through a charity donation of Genzyme, a manufacturer of the Cerezyme medication, I got a new chance! I was able to be treated! I had the right to feel joyful about life! I have been taking the medication for several years now, once in two weeks.

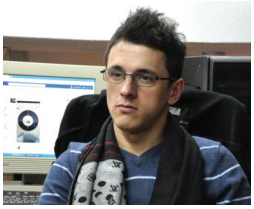
I felt better and stronger right after the first year of therapy. Now, I no longer have problem with broken bones and, what is more important, I have a wife and children, a family and life! I sometimes "catch" myself worrying too much over the children, fearing that they might get injured or sick. I keep forgetting that they are healthy and do not suffer from the Gaucher disease like their father.

Now, from this perspective I can say that once you have the medication, Gaucher is not even a threat! I hope that the time will come when all patients are able to receive adequate treatment and feel what it is like to be healthy!

When the only solution is to seek the medication abroadood

**Message from patients:**

Symptoms slowly give way to the power of therapy. If necessary, turn the world over to get the medication!



**Jetmir Ceka,**
**Tetovo**

- a patient suffering from Gaucher, a rare disease

## Apart from health issues, a rare disease brings along other problems as well

After a long roaming about hospitals, hours and days spent in hospital halls and at reception desks, seeking for answers to the question what my illness was and what it is that differs me from others, one day I finally received the answer. However, together with the ruthless fact that I suffer from a rare disease, I received an additional explanation that there was no treatment for it. It was very difficult for me; I only wondered how I would live with the excruciating pain...how must my parents feel?

Even when I was a 2-month-old baby, I had a huge stomach resulting from the disease that had developed very early. During my entire childhood I differed a lot from my peers. My stomach was growing

enormous, my bones grew weak, a leg problem occurred and I could hardly move. Symptoms of enlarged spleen recurred, a lot more than usual. It is not very easy to live with a rare disease. At times I even thought that my parents grew tired of me, as I was always complaining that I was in pain and suffering.

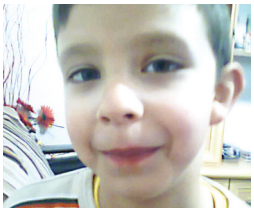
It all changed a year and a half ago, when I was informed at the Pediatric Hospital in Skopje about a project that was under way in Bulgaria. I am being treated there now under a test project for new medication, which is not administered through infusion but in a form of pills and is developed by Genzyme. At the beginning, I travelled to Bulgaria every two weeks and now I do that every three months. Having started receiving the therapy, I feel much better. I know that if I continue with the treatment, I will be just like the others - I will have a normal life.
In addition to health issues, having a rare disease caused other types of problems. I could not do any physical or hard work, and had no energy. It is difficult to watch the others work and be forced to stand out from the daily routine...
Now I only think about the future...

What does it hold?

When food is "forbidden fruit" instead of fuel of life

**Message from patients:**

Children with a rare disease also have a right to normal life!



**Viktor Dimitrijoski,**

- a child suffering from Phenylketonuria

Story told by:
**Dragan Dimitrijoski,**
Viktor's father

# My child is condemned to a different life

Have you ever come across these three letters, written next to one another – PKU?

Do you know what difficulties families face when a child with this genetic disease is born? How to help them raise a child with normal mental and physical capabilities instead of a sick one?

Phenylketonuria is a rare metabolism disorder preventing the organism to dissolve protein-rich food in a normal manner. This results in a large concentration of phenylalanine in the blood, which then accumulates in the brain and damages it permanently. This in turn leads to mental and physical developmental disabilities in the child. Imagine a child

unable to eat meat, eggs, fish, tree nuts, corn, wheat...That is practically 90% of the food that others consume regularly!
An additional difficulty is that there are no screening tests for early detection of the disease in Macedonia and there are neither specialist doctors in this area nor special staff needed to control the level of phenylalanine in the blood.

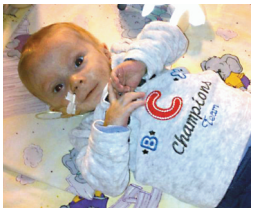
Being a father of a child facing this disease, all I want is for him to grow into a healthy human being and have an equal opportunity for normal life, just like any other child. Phenylketonuria treatment is possible only in the form of a low-protein diet, i.e. by reducing the natural protein intake. The vitamins and minerals needed should, on the other hand, be taken in a form of a special protein product and special flour for bread. If the child does not take this product, it ceases to grow, the creation of immunoprotein in the organism is reduced and a muscular fatigue occurs, intellectual capacity is affected, proteins in muscles begin to decompose and phenylalanine concentration increases, causing a severe retardation. The product is rather expensive and the whole therapy costs 400 – 500 euro a month. Considering our living standard, this medication that means life equals luxury. I hope that all competent institutions we addressed in this country will have understanding for the situation we are coping with, as we are now forced to ask for help neighboring countries such as Serbia and Bulgaria.

My child is left on its own! I am wondering why are we less important than other patients and citizens?

When childhood is everything but carefree!

**Question from patients:**

How much does a child's life cost?!




**Hristijan Todorovski - Kiko,**

- a child with a rare disease not diagnosed yet

Story told by:
**Dragan Todorovski**
Hristijan's father

# What does future hold for our child?

It is very hard to describe and even harder to write about the pain of a parent when he finds out that his child suffers from a rare disease. This past year cannot be described in short, but here is the story of greater problems we faced recently.

My wife had a perfect pregnancy: all 9 months all tests were excellent. But still, it turned out not to be so. Last year, Hristijan was born on January 4. From the very beginning, he was subjected to treatment, i.e. infusion. Upon our initiative, we took him to the Pediatric Clinic in Skopje. Until his first departure to Slovenia for diagnosis, Hristijan had received injections several times a day. Every single neonatologist took blood samples, in an attempt to

determine the type of the rare disease.

It is not certain that even bone marrow transplantation would put an end to the problems he faces. Doctors in Germany also do not know what his problem is, but they promised to test the entire genome.

The prognosis were terrible in Macedonia: "if he vomits blood one more time, he is finished... ", "there is no medication, it is what it is, he is alive until he is..."! In such periods a support from your family is very important and we are happy that at least our closest ones understand and help us. Without their support, we would not have been able to endure this.

Receiving documents for treatment abroad was not an easy process at all. We had to beg the doctors to write e-mails to certain health institutions, as hospitals do not contact parents only; in addition, they ask for an expert opinion of the patient's condition. We found a hospital, arranged a treatment and appeared before a committee. We were refused at the First Instance Committee as the amount was too high: 350,000 euro. It was then that my wife and I asked ourselves how much is our child's or any other child's life worth...

The last desperate attempt was out appearance in the media. Does it have to be so dramatic to get what we are entitled to? Don't we have health insurance? Babies are 100% insured until their third year. Why are committees and heaps of papers needed for their treatment?

Here, in Germany, a thought is troubling me, the parent, on how Hristijan will receive further medical treatment when we return home. His future most probably holds frequent visits to doctors and hospitals. What can we do when back home we do not have any program for people with rare diseases? What does future hold for our child?

**Dejan Angeleski,**
**Skopje**

- a rare disease child
Hereditary Angioedema

Story told by:
**Natasha Angjeleska**
Dejan's mother

# The courage of my child is the best lesson of life

I am Dejan's mother. I often introduce myself with those words and it makes me feel good. I will tell you why. I adopted Dejan as a 3-month-old baby and was the world's happiest mother of a lovely child with angelic blue eyes that absorbed my love from the very first day. Starting with the first months, there was need for frequent visits to the doctor's, but we ascribed that to his weak immunity, as he had been deprived of mother's care and had not been breast-fed. The visits to and talks with the good nice doctor helped me learn a lot. Being an experienced doctor and having a strong medical intuition, she advised us to make tests hardly any doctor would refer patients to. Then, in 2008 Dejan had his diagnosis made at the Pediatric Clinic in Skopje. He has a rare genetic disease – Hereditary Angioedema (HAE). It is estimated that 1 in 10,000 to 50,000 people are affected by HAE. This condition is manifested in swellings on various types of the body (legs, hands, face, lips, throat, and genitals) and internal organs as well. It is impossible to predict the occurrence of a swelling, which can even be deadly if it occurs on the throat or respiratory tract. Having informed me of the diagnosis, the doctor gripped my hand firmly and soothingly said:

"All you need to do now, mummy, is to accept reality and become informed about the condition in order to prepare the child to cope with it..."

Now I realize that she was completely right. I spent many sleepless nights besides the hospital bed of my child, unable to help him, forced to be patient. Oh, how long are the minutes, how intense the ear buzz, how patience decreases to an extent at which you no longer wish to hear any comforting words or drink any water...or anything! You only wish for the child's pain to stop. And then you suddenly feel strong, a ray of hope shines on you, telling you that you need to be strong for tomorrow when your kid wakes up and you need to fake a smile and tell him a nice word and wipe his tears. I learned that temptations only seem to make us weaker, when in fact we become stronger. Dejan is now 10 and he is still too young to understand what is happening to him. He is frequently absent from school and I am trying to explain to him, to the school officials and to his friends and parents that he would feel much better if he spent his time at school together with his peers and that his condition is neither his nor his mother's choice. Dejan is still a happy and brave little kid. He encouraged me to learn to administer the intravenous therapy myself. He is not always in the mood to go to hospitals, particularly when we have to travel or visit unknown doctors. He grew accustomed to the friendly faces in "our" clinic.

Unfortunately, the therapy for Dejan's disease is not available in our country and the few patients diagnosed thus far do not always have the opportunity to obtain it themselves. Dejan and our friends whom we grew close with as a result of the disease have many difficulties to live "normally" and carry out their everyday obligations like everybody else. However, believe me, we support each other; and the fact that I have who to share experiences and consult with and that I have met people with big hearts, perseverance, fighting spirit and a wish to live just like everybody else, makes me happy.

## Rare Diseases within the Association

The Association of Rare Diseases - Life with Challenges was created by patients and parents facing Gaucher rare disease. Later on, parents of children with Phenylketonuria joined, along with Hereditary Angioedema (HAE) patients as well as a child with Alagille Syndrome, which is the only case in Macedonia.

Other rare diseases have been diagnosed in Macedonia as well, such as: multiple myeloma, myelodysplastic syndrome, gastrointestinal stromal tumor, acromegaly, gastroenteropancreatic neuroendocrine tumors, mucopolysaccharidosis, hereditary tyrosinemia, galactosemia, hereditary fructose intolerance...

---

• **Gaucher** is a disease caused by the lack of one enzyme in the organism that leads to problems such as enlarged spleen and liver, weak bones, frequent fractures, weak immune system, thrombocytopenia, low hemoglobin, dizziness resulting from the loss of energy, etc. Currently, 12 Gaucher patients have been diagnosed in Macedonia, 5 of whom are being treated with a donation from the company that manufactures the medication, which relieves all the symptoms and completely normalizes the patients' medical condition.

• **Phenylketonuria** is a rare disease manifested in a disorder of the organism, preventing it from normally dissolving protein-rich foods. It causes brain damages and mental retardation, accompanied by physical retardation. There are about 6 patients in Macedonia diagnosed with this disease, who themselves obtain the special protein products and special flour for bread, along with medications necessary to cope with complications, in order to ensure normal life.

• One child has been diagnosed with **Allagille Syndrome** in Macedonia. This genetic disease is characterized by a progressive loss of the liver function, causing cirrhosis in 30% to 50% of the cases, along with yellow skin, scratching and eye-problems. In order to improve the condition and ensure normal growth and development, a treatment with liver, skin and eye medications is necessary, as well as a special infant formula diet.

• **Hereditary Angioedema (HAE)** is a rare disease manifested in swellings (edema) on various parts of the body and internal organs. No medication has been discovered yet, but there are treatments to maintain the normal condition of the patients. In Macedonia around 10 HAE patients have been diagnosed thus far.
The Association is open to other patients and parents with rare diseases as well as supporters!

**The Association is open to other patients and parents with rare diseases as well as supporters!**

There is a low level of rare disease diagnostics, and diagnosis and treatment need to be improved. Currently, the Ministry of Health provides funds for treatment of only two mucopolysaccharidosis patients under the Rare Disease Program.

Association of Rare Diseases


Association of Rare Diseases
Life with Challenges – Bitola

Contact: 070 705 446

Gyro Account: 210 – 0649841801 – 71

e-mail: zivotsopredizvici@gmail.com

Find us on Facebook:
https://www.facebook.com/groups/312483895490987
https://www.facebook.com/LifeWithChallenges

www.challenges.mk