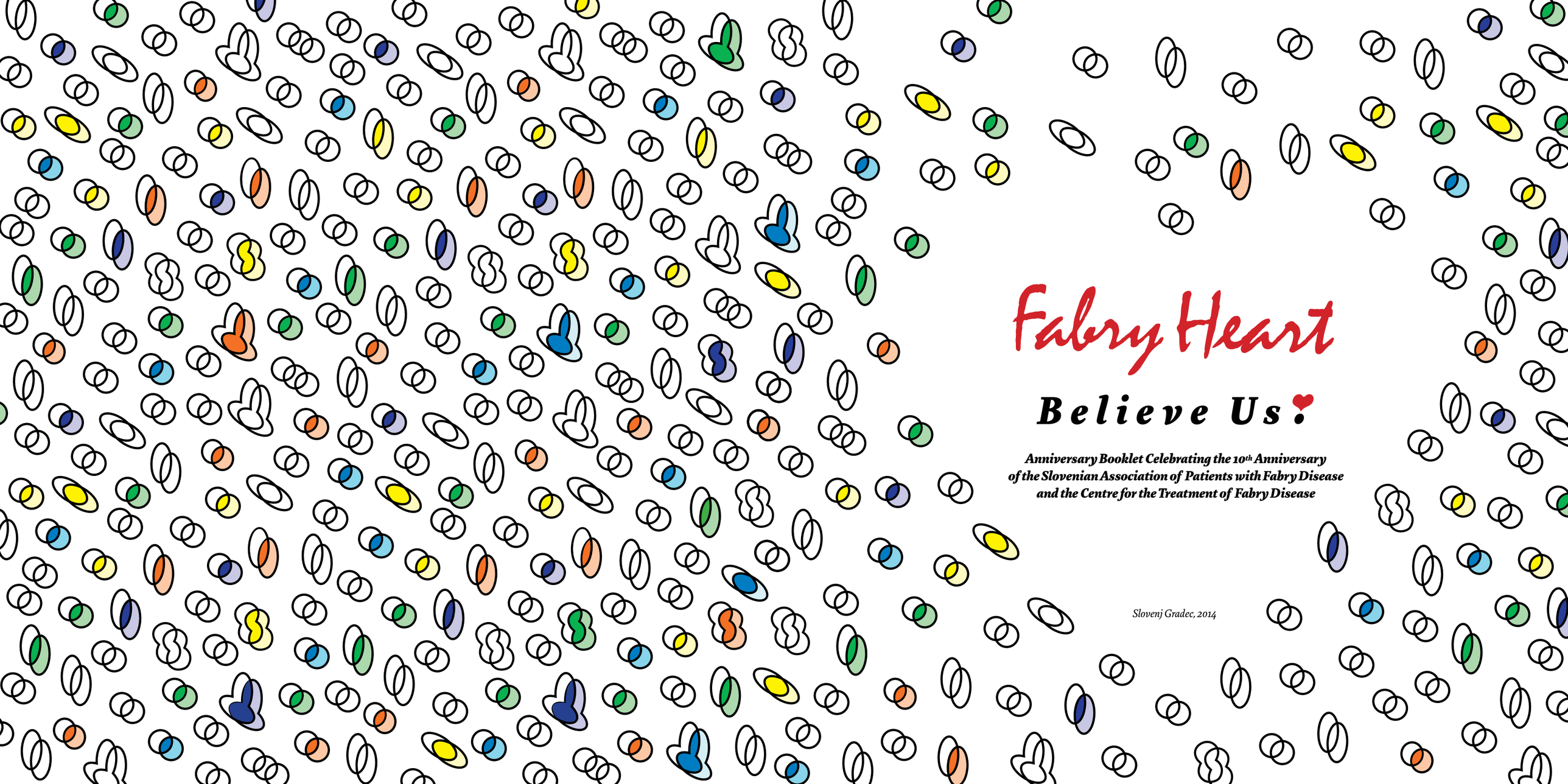
Fabry Heart



Fabry Heart

Believe Us!

Anniversary Booklet Celebrating the 10th Anniversary of the Slovenian Association of Patients with Fabry Disease and the Centre for the Treatment of Fabry Disease

Slovenj Gradec, 2014

1. Thanks Go to...

All Who Believe Us that Fabry disease exists
with many painful and sometimes suffering faces:

... Bojan Vujkovac, M. D., the heart of the Centre for the Treatment of Fabry Disease
and selfless member of the Slovenian Association of Patients with Fabry Disease.

... Andreja Cokan Vujkovac, M. D.,
for her idea to prepare this booklet, for her compassion and for her professional help extended to the medical team at the Center for the Treatment of Fabry Disease.

... Vesna Korat, R. N., a caring nurse at the Centre for the Treatment of Fabry Disease,
for help, compassionate kindness and warmth that she provides to the patients with Fabry disease.

... all members of the medical team at the Centre for the Treatment of Fabry Disease,
who selflessly provide help to all patients with Fabry disease.

... all members of Slovenian Association of Patients with Fabry Disease,
who share their sincere life stories with us.

... Andrej Porenta, Tomi Trilar *and* **David Valič ,**
for publishing their photos.

... Karmen Bizjak Merzel,
for editing the booklet.

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CENTRE FOR THE TREATMENT
OF FABRY DISEASE

DRUŠTVO BOLNIKOV
S FABRYEVO BOLEZNIJO
Dobja vas 153, 2390 Ravne na Koroškem

SLOVENIAN ASSOCIATION
OF PATIENTS WITH FABRY DISEASE

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2. A Word From the Editor

And could you keep your heart in wonder at the daily miracles of your life,
your pain would not seem less wondrous than your joy.

K. Gibran

All the Best and Have a Safe Journey, New-born Booklet!

We have it – a booklet, which will be gently held by the members of the Society for Fabry disease who will live next to their stories. We realized the idea and initiative to verbalize the experiences, feelings, fears, problems, obstacles… that inherited Fabry disease links, in one family.

»Fabry disease is a rare flower in an unexplored jungle,« was written by a specialist in internal medicine, Andreja Cokan Vujkovac, who together with her husband sowed the enthusiasm between the members of the Society on publishing the booklet. The booklet is a gift to mark the 10th anniversary of the Slovenian Association of Patients with Fabry disease in Slovenia and the Centre for the treatment of Fabry disease, which provide support, assistance and professionalism to patients with Fabry disease.

The gift knits together very different, but also in a way, similar life stories – often painful, but also optimistic ones. It is like a mountain, to which each patient climbs differently: some conquer the summit, as the disease does not hinder them at all, other follow a different path – with many stops, shortcuts… while some will have optimism and conquer the disease in their minds. Then there are fourth, fifth… each with their own breath and step of life.

How many opportunities can people take advantage of, how can you outsmart your unwanted companion, how can hope revive our bodies… all of this is collected within the pages of this booklet. Each person lives with their disease and must accept it as an unavoidable part of life. Often, quietly, unnoticed, hidden, with pain, they suffer – alone, with the disease…

The colours of life constantly change in the eyes of Fabry sufferers. The painters are they themselves – with their patience and, gratitude for sympathetic friends, family and compassionate doctors, with joyous moments without pain, learning and transmission of pain and wisdom. They donate these coulours now, to us, the readers, to look through their eyes to better comprehend the darker, mysterious side of life and together change it in to a rainbow.

Karmen Bizjak Merzel

3. Initial Medical Findings

... Who are you then?—
I am part of that power which eternally wills evil and eternally works good.

Goethe, Faust

The Beginnings of Our Diagnosis and Treatment of Fabry Disease

In 1991, Slovenj Gradec General Hospital's Internal Medicine Department came across Fabry disease quite coincidentally.

Actually, it all began a year earlier, in November 1990, when a 46-year-old patient was admitted for cardiac arrhythmia. During the tests, results of an EKG test caught our attention. In addition to irregular heartbeat due to atrial fibrillation, it recorded excess thickening of the left ventricle. This surprised us greatly, because this type of EKG abnormality is usually characteristic in high blood pressure patients. Our patient's blood pressure, on the other hand, had always been normal and until this occurrence of abnormal heart rhythm, he never had a heart condition. Echocardiogram revealed a uniform left ventricular hypertrophy, especially the interventricular septum showing unusually grainy structure. Lab research showed slightly increased urine creatinine values and protein in urine.

In May 1991, the patient's one year younger brother was admitted for additional tests after protein was found in his urine and it was suspected that he had a cardiovascular condition. Prior to his admission, he was examined by an internist, who found unusual skin changes, which took the form of tiny bleeding papules on his buttocks, groin, penis, armpits and oral mucosa. The patient said he had been noticing these changes since his childhood. His EKG also showed a thickening of the left ventricle. However, he denied having any heart problems and his blood pressure was normal. His echocardiogram results were similar to his brother's: A uniform thickening of the left ventricle, especially of the interventricular septum which had an unusually looking grainy structure. Other tests excluded any serious blood and kidney disorders. Due to the unusual skin condition, he saw Jožica Kotnik, M. D., specialist in dermatology and venereology at our hospital, who suspected Fabry disease, based on the clinical picture and family medical history. She suggested that the patient's other brothers be tested as well. Based on her advice, the patient underwent an abdominal skin biopsy. Microscopic histological examination of the sample revealed changes indicating Fabry disease.

Since the brothers had other siblings, we arranged to examine their whole families, their living mother, seven brothers, and two sisters. We asked them about any troubles associated with Fabry disease and gave them a thorough physical examination. This included EKG, echocardiogram and basic laboratory tests. They saw a dermatologist to examine skin changes and an ophthalmologist to find if they had any striped abnormalities (cornea verticillata) typical of Fabry disease. Based on all the results we obtained, we were able to conclude with great certainty that five brothers had signs and symptoms indicating Fabry disease, which were not found with other family members. All patients said they used to feel severe pain in their arms and legs or joints when they were younger. None of the five patients would sweat when hot. Three of them experienced frequent diarrhea regardless of the type of food they ate. Three of them had skin changes in the form of capillaries in typical places on their body. These lesions are called angiokeratomas and are characteristic of Fabry disease. No such changes were found with two of the brothers. EKG and echocardiogram tests showed similar abnormalities in all of them.

Since there were no enzyme tests performed in Slovenia in 1992, we agreed to have them done at the Chemical and Biochemical Institute at the Medical Faculty in Zagreb. During the tests, the activity of the ά-Gal A (ά-galactosidase) enzyme in leukocytes is measured. Fabry disease is characterized by reduced ά-Gal A enzyme leukocyte activity. The younger of the two brothers diagnosed with Fabry disease took the test together with his son and daughter. The father's enzyme activity was not measurable, while his son's results was almost normal and his daughter's was slightly lowered. We agreed to perform similar tests with other family members, at least those showing clinical signs of Fabry disease, but the outbreak of the war in Croatia prevented us from carrying them out.

Motivated by all our findings, we decided to proceed with our Fabry disease research and systematically examine the patients' family members across multiple generations. Four generations included a total of 45 people. Since we could not analyze the enzyme activity or perform genetic research in Slovenia, we turned to the experts from the Academic Medical Center in Amsterdam for assistance. Gabor E. Lindhorst, M. D., and his colleagues kindly offered their help. They performed analyses of the ά-Gal A activity and a genetic analysis for 31 of 45 family members including all members of the first three generations and several members of the fourth generation over 10 years of age. The results revealed that the mother was the carrier of the disease, having passed it on to seven male family members. Apart from the grandmother, 10 other women carriers were discovered across all generations.

During our research and testing of Fabry disease patients, similar research was going on elsewhere in Slovenia. In 1980, military doctors Nedeljko Akšam, M. D., and Risto Angleski, M. D., of the Military

Hospital Ljubljana diagnosed Fabry disease with a 23-year-old soldier and described the case in a journal for doctors and pharmacists published in the then Yugoslav People's Army, called *Vojnosanitetski pregled* (Ed. 39, No. 2, p. 118–120, April/May 1982).

The first description of two Fabry disease patients was revealed to the professional public at the Slovenian Nephrology Congress in Portorož in 1996. Marija Čeh, M. D., internal medicine specialist form the Novo Mesto Hospital, described two families of Fabry disease patients. In the first family, the son was the patient and the mother was the carrier. In the second, the deceased father was the patient and his two daughters were carriers. Fabry disease was proven based on histological testing of skin abnormalities and an analysis of the ά-Gal A enzyme in plasma and leukocytes performed in London. We found later on that another family was discovered in northwest Slovenia with the son as the patient and the mother as the carrier. With both, Fabry disease was confirmed through kidney biopsy.

The number of newly discovered Fabry disease patients in Slovenia grew year after year. Having gained a lot of experience, we decided to start performing examinations of all Slovenian patients with diagnosed or suspected Fabry disease. In order to examine them systematically, as well as advise appropriate treatment and monitor their medical condition through regular periodic examinations, a group of experts was formed at our hospital in 2000 consisting of specialists from various fields. Since 2001 we have been able to treat all Fabry disease patients in Slovenia at our hospital. By 2003, five families with Fabry disease were discovered. Sadly, three of our patients first diagnosed with Fabry disease passed on that year.

Until 2003, treating Fabry disease patients meant alleviating their symptoms and severe accompanying diseases with medications and other treatments. Elsewhere across Europe and in the US, hospitals have already begun using treatments that substituted the missing enzyme with an artificial one. In implementing this treatment in our institution and overcoming other difficulties associated with Fabry disease we were kindly assisted for a number of years by Diana Rovers, representative of Genzyme, a pharmaceutical company producing the replacement enzyme called Fabrazyme. In September 2003, we joined others worldwide in implementing a new treatment method. It was administered to seven male patients and one female patient. This increased survival prospects of Fabry disease patients substantially.

Over the years, we gained a lot of experience in recognizing Fabry disease and applying the new treatment method, which led us to the idea of establishing a special unit dedicated to treating these patients in a truly holistic manner. On 8th October 2004, the Center for the treatment of Fabry disease was established officially in our hospital, which is currently the only organization in Slovenia that is successfully treating Fabry disease patients.

Franc Verovnik, M. D.

They Are Heroes for Me…

I first heard of Fabry disease in medical school. It was considered an exotic disease and I did not think that career I would meet a patient with this disease. However fate had a different plan. I started to participate in the team guided by doctor Verovnik, who after they discovered the first patient with Fabry disease, made a family tree and this way discovered 19 sufferers. All the time I ask myself, what would happen to these people if there wasn't so much enthusiasm from my former superior and teacher? Probably these patients would wander from doctor to doctor with variety of problems. Unfortunately this is the fate of many Fabry sufferers.

I remember our first patients. They were brothers. During check-ups they told me about their childhoods. Some have forgotten the terrible pain they had in their hands and feet and they had to try and remember when we asked directly. I was told that they had to cool their hands and feet in the snow to ease the terrible pain, which sometimes made them unable to walk. One of the brothers had to be in a wheelchair for a few years. They had been jumping on each other's foot just to drive away the unbearable pain that could not treated with household painkillers or even stronger analgesics. Later, other sufferers told me about their sad childhood. One had skin lesions removed by liquid nitrogen. He was in a lot of pain and was forced to hide from doctors and lie to his mother. Due to the severe pain he was stealing the medicine, Tramal. The pain was caused by stress and heat. When he hid in school because he did not want to do PE, they found him and as punishment he had to dig potatoes in hot areas. He was considered a simulant and this still hurts him. The mother, who is also a sufferer, said that her son could not walk because of the pain. They visited the doctor, who simply did not believe their description of the pain. When they left, she looked at them through the window and saw how the mother had to carry the child, so she finally called them back. I think the most painful thing for sufferers is that no one was believed to have problems. And what makes it worse is, - not even relatives believed. Several times the mother complained that she feels guilty because she did not believe her son.

People with Fabry disease have repeatedly surprised me with optimism, a strong will, and determination. The kind of tasks they do and how successful they are at them! One certain sufferer – a postman got frozen feet on his motor bike in winter. But also this did not reduce his will to work and live – moreover – he became a miner! I can't imagine how he could manage his pain with such efforts.

Young sufferers have told me they spend all day the summer in a tub with cold water, because they get so overheated when they are exposed to external heat. Sufferers with Fabry disease do not sweat. That makes them unable to regulate their body heat. The temperature changes cause this unbearable pain.

At first, sufferers were treated only symptomatically, therefore we only eased the symptoms. Then we got the medicine in Slovenia. For this, most of the credit belongs to doctor Vujkovac. This medicine is very expensive and we are happy to have it in Slovenia for which it is provided for all Slovenian Fabry patients who need it. However, across our southern border sadly it's not available. 10 years ago we founded the Slovenian Association of Patients with Fabry disease, which functions well and includes sufferers, families, medical staff and friends. Almost everey year we attend gathering together, these are really special because they are enriched with guidance by our biologists. Such meetings solidify our cooperation and connections even more. They are important for the exchange of experiences and mutual support.

During my participation in the medical team staff, I attended many professional courses and met a lot of experts who deal with this disease. On several occasions we exchanged experiences and advice. We obtained further details which would help patients. We also participated in registers, which are really important for this kind of rare disease, because they inform us of many new discoveries and illusions about the disease.

I wish that each Fabry patient would get the required medicine on time. This can stop the progress of the disease of time. Often I am sad when I feel helpless, faced with the many problems of sufferers. When dealing with Fabry sufferers I am surprised when they express their optimism and will to live, and calmness when they deal with serious problems, this characteristic pervades every individual. They are heroes to me and have taught me the best lessons life can give you.

Fabry disease is a rare flower in an unexplored jungle. With this booklet we help to uncover its features and secrets. I believe that in the future a drug will be developed which will cure the disease completely.

Andreja Cokan Vujkovac, M. D.

Our Story

We all like listening to nice stories and our story is one of these. There are good and bad characters, the story has its complications, but it seems to have a happy ending.

It all started a long time ago. At our department we accepted someone who suffered from heart and renal failure and had unusual rashes on the skin. In searching for a possible diagnosis we consulted our dermatologist doctor Kotnik, mainly because of the skin rash. She surprisingly diagnosed the person with Fabry disease already at the entrance to her office. We were excited and respectful of her knowledge. But the patient's problems were not solved, because back in 1991 there was no known medicine for Fabry disease. But we had good fortune as our department head, doctor Verovnik, found a great challenge in this case. He initiated a systematic examination of other family members. He was looking for possibilities across Europe to carry out genetic analysis in order to definitively confirm the disease. At that time there were no established routes and many of these investigations were carried out more as a favour on the basis of personal contacts. Back then the technology itself was such that we were waiting for the results, sometimes for almost a year. Nowadays such a situation is hard to imagine as we can determine the results ourselves within a few days. However, doctor Verovnik was extremely patient and within a few years he managed to build the first disease family tree of the affected family. Based on his experience he came to an important conclusion very early, namely that women are not just disease carriers, but also sufferers, which he systematically emphasised. He generously shared his findings and knowledge with his colleagues through numerous lectures and paper contributions. It was also due to Fabry disease that our hospital became widely known. In the nineties individual patients were also diagnosed in other hospitals but due to the absence of proper drugs only the symptoms and certain complications were able to be treated. When we decided to systematically process and manage our patients in the year 2000, colleagues from other hospitals also handed over the treatment of their patients.

From the very beginning we have implemented a consistent family testing regime which subsequently enabled us to discover new sufferers, many of whom, especially younger men and women who did not show signs of the progressive disease. Gradually the number of patients increased from year to year. Unfortunately, we were not able to offer more than monitoring and alleviation of the symptoms to our patients at that time. Everything changed in 2001.

By accident I met Mrs. Rovers who was representing the Genzyme company at the Congress of Nephrology. At Genyzme they had just developed a completely new drug for the treatment of Fabry disease. At that time we already had five approved patients with the advanced stage of disease, which was, according to the European context, a very large number for such a small country, and even larger so for such a small hospital. Very soon after this event we were invited to participate in the first international clinical trial of the efficiency of this new drug. Despite the recognition and the challenge to be able to cooperate with one of the most reputable institutions in the world in this field, we refused the offer. We had the chance to include just two patients in the study in which one patient would receive a drug while the other one only a placebo. The decision was even more difficult because all five of our patients were brothers and they all needed treatment. Unfortunately, we were not able to include more patients simply because the drug was not available in such quantities. Even today our decision would most likely have been the same.

Then, we concentrated all our efforts on providing treatment as soon as possible for all our patients. In the following years, we made numerous applications and requests, wrote letters, and organized a series of meetings. We gradually got acquainted with Slovenian medical bureaucracy. As an example, let me mention that all the relevant authorities and officials in health care required a positive decision from the expert commission for internal medicine before taking any further steps, but the commission had no session for years. Even the chairman of this commission, who was then in charge of other important functions, explicitly required the same thing, but at the same time had not called any meetings. We were often told to act more responsibly because treatment of a disease for only a few patients is simply too expensive. However, there were also many exceptions, in particular Professor Rajko Kenda, who has always given us advice, helped us to overcome many bureaucratic obstacles. I still remember the meeting of the Health Council, in which as a response to our presentation the late Mr. Toth, M. Sc., took a very clear position: »If the treatment has been proven effective, then it is our duty to provide the funding for it.« With this statement the meeting was closed. A battle was won, but not yet the war...

I still remember many conversations with Metod, one of our patients, who came regularly to us for dialysis, because of Fabry disease leading to kidney failure. Metod was a very brave and courageous man, who was from the very first day, convinced that we would succeed. We were all waiting for the moment when the miracle would happen to him, but we were facing only further complications.

Then it happened. Suddenly Stefan died. We were shocked, then his brother Simon died also. We were still waiting for a decision from the authorities to initiate the treatment. We were repeatedly asking: »Do all the brothers have to die before this will happen?« Perhaps it was a blessing in disguise that the sad family story got noticed by journalists who immediately made a TV show. We can only speculate whether this was the critical moment that finally opened the door to the new treatment, but three months later in September 2003 Metod received the first infusion of enzyme replacement therapy. But as if there were not enough accidents, three days after the first infusion Metod suffered a stroke and later as a consequence of the disease he died. Fortunately, the treatment did not cause major complications with the other patients. Unfortunately, in the following years the treatment has not turned out to be a miraculous drug. So as a result of the disease Branko, Elizabeta, Tone and Marjan passed away in the coming years. Today we know that the treatment should be initiated as soon as possible in order to be efficient.

With the final approval of the enzyme replacement therapy, the treatment became available to all Slovenian patients who needed it. This important decision is still valid today. However, we soon realised that drug administration is only one of our tasks. We set up a new organisational framework and founded the Centre for the treatment of Fabry disease in October 2004. In its ten years of activity the Centre has found its place and role both in our country as well as abroad. Through our activity and results we managed to convince those responsible that patients suffering from rare diseases need to be treated differently which requires different approaches and organisational regimes. It is essential for us to develop a patient-centred approach in the Centre while the therapy and the treatment itself should be adjusted in a way as to best meet the patient's needs. However, there are still important tasks and challenges to be addressed in the future.

Once, I was asked how we managed to achieve all this. »Faith,« was my reply and we also believe in the future what we are striving for and we are striving for what we believe in.

Bojan Vujkovac, M. D.
Head of the Center for the Treatment of Fabry disease,
General Hospital Slovenj Gradec

Great People

Nine years ago I began to work as a fairly inexperienced nurse at the Slovenj Gradec General Hospital in the Internal Medicine Department. At first I was not assigned to a single department; instead I was circulating between different wards. From time to time I also heard the word Fabry. This word represented something unknown and unusual to me. During the circulation around the various parts of the hospital, I often worked in the F section, where the Centre for the Treatment of Fabry Disease was located. I remember how I couldn't bring myself to open room no. 119, the door of the Centre and check what was happening in there. The reason for this was probably my initial shyness and inexperience. However, since I felt that I needed new challenges, I applied for the vacant nurse's post in the Centre for the Treatment of Fabry Disease. When I was selected, I had mixed feelings of dear, because of my lack of knowledge, and excitement, because I could do something new. I immediately got some theoretical knowledge about Fabry disease through the Internet and from various articles, and quickly found out that to be a success i n this field is very hard. A colleague of mine who used to work in the Centre for the Treatment of Fabry Disease handed over the organisation and contents of work, and then it started.

When the first patients arrived, it was necessary to gain their trust, as some had been treated for several years and were used to the previous nurses. Soon, we became very friendly, we have established nice relationships based on respect and trust. Treatment is for life and because I meet sufferers every 14 days, I get attached to them. Because Fabry disease is complex and patients have a wide variety of problems, I am glad to be able to talk to them about what burdens them. During our sessions (therapies), sufferers have taught me a lot. Despite the extremely serious illness, they bravely continue to walk the path of life. Their life stories have touched me, and so I appreciate things, that I have previously taken for granted, even more. Because the disease is genetic, the treatment of sufferers includes their family members, who often need our support. Through the years of our sessions we have built not only professional but also friendly relationships. So I am really looking forward to the annual meetings of the Slovenian Association of Patients with Fabry Disease, when we chat about everyday things, relax, joke and deepen our relationships.

Because of i ts specific forms of organisation, work in the the Centre for the Treatment of Fabry Disease differs significantly to the traditional health care system, as the coordination of the multidisciplinary team, is largely independent. Due to the variability of the system, which is updated frequently, it requires a person who is flexible. At the beginning, this is definitely a challenge for a nurse without an experience in this type of environment. Just like in any other profession, with experience and several years of training it gets better and easier. However, without the support from my team, doctors Bojan Vujkovac and Andreja Cokan Vujkovac, it would not work. Therefore, thank you for all the knowledge and support. Thanks also to my co-workers who have been, and are involved in our work. Above all, thank you, dear sufferers, for being extremely flexible and patient in the treatment schedule and thanks for still tolerating occasional mishaps during the therapy. I believe that the treatment of patients at the Centre for the treatment of Fabry disease is professional, the people are friendly and in particular, it provides a comfortable environment like home.

So I think that the patients, their families and of course the medical team which is involved, feel good, relaxed and satisfied. I'm glad I can be part of this treatment and that I have met a lot of »»great« people, from whom I learned a huge amount and surely will in the future too.

Vesna Korat, R. N.
Nurse at the Center for the Treatment of Fabry disease,
General Hospital Slovenj Gradec

4. Slovenian Association of Patients with Fabry Disease

Do what you can, with what you have, where you are.

Theodore Roosevelt

DRUŠTVO BOLNIKOV S FABRYEVO BOLEZNIJO
Dobja vas 153, 2390 Ravne na Koroškem

About the Association

The Slovenian Association of Patients with Fabry Disease was founded in 2003. It brings together patients, their family members and friends as well as the medical staff.

2003 was also the year when the treatment using the artificial enzyme was first implemented in the Centre for the Treatment of Fabry Disease in Slovenj Gradec General Hospital.

The Slovenian Association of Patients with Fabry Disease joined the Fabry International Network (FIN) and Eurordis. Some of the members attended various conferences, such as the ones in Warsaw, Amsterdam, Sarajevo, Dubrovnik and Tuhelj, where they could share their professional and personal experience in treating rare diseases (patient status, drug supply, healthcare system organization, raising awareness of rare diseases, etc.).

One of the important events for our members is the Rare Disease Day taking place each year on 28th February in Ljubljana. This day is crucial because it promotes public awareness and establishes a place for rare diseases. This awareness unfortunately does not have the staying power, as experience show.

Doctor Bojan Vujkovac, who is the heart of the Centre for the Treatment of Fabry Disease , is an active member of two work groups at the Ministry of Health, specialized in the field of rare diseases.

As a member of the Slovenian Association of Patients with Fabry Disease, he submitted the following propositions:

· To found a frame organization for the patients with rare diseases (Slovenia is the only EU country who does not have one). This would promote improved organization of all patients with rare diseases.
· To organize a national congress inviting all stakeholders (patients, association members, Ministry of Health representatives, Health Insurance Institute of Slovenia representatives, healthcare representatives) to help define concrete future projects and measures.
· To organize a work group at the Ministry of Health specialized in rare diseases and responsible for elaborating an action plan.
· To open a rare disease office responsible for gathering information. This information would serve as the basis for establishing a national registry (the Center for the Treatment of Fabry disease already has its own registry of Fabry disease patients and can submit the data for the joint registry immediately). The registry would provide a snapshot of the rare disease situation in Slovenia. The office would also keep supplier information that could be made available to the patients.

The Slovenian Association of Patients with Fabry Disease annually organizes a day trip for the patients and their families. These trips are also an occasion where doctor Bojan Vujkovac provides them with the latest information about Fabry disease.

Among the Association's projects is the publication of this booklet to mark its 10th anniversary. I would like to give my sincere thanks to all members and the medical staff for contributing their thoughts, feelings and stories and for helping make this booklet happen.

Marija Pintarič
President of the Slovenian Association of Patients with Fabry Disease

5. Living with Fabry Disease

A Father with a Fabry Heart

It's not that I would not want to write anything about the disease, it is just so damn hard. It's hard to recall all the beautiful and sad memories and stay cool, it is hard to browse through the brain without disturbing the heart. Despite tears and a headache, another Fabry story has been written.

Once upon a time there lived a man. He was strong and full of energy. Physical work has never been too difficult for him. In his job he had always been »the head«. He mowed the whole lawn by hand in the morning or in the midday heat (it did not matter to him) – it was easy for him. Walking in the mountains and skiing was nothing more than a pure pleasure. Once he told me how, as a young boy he went with a friend for a few days in the mountains. In those days it was not as easy as today, when we drive to the starting point or take the cableway and go to the top. No, it was different then. He rode by train as close as he could get to the mountains and then walked for several hours, slept in a cabin, and continued his path which led to the valley of the first station. But what if he missed the last train? No big deal, he just had to walk all the way home. Yes – these things this man was able to do.

Then once, the same man, while his normal summer job, stopped. He was mowing the lawn, but he suddenly became exhausted, he had severe nausea and had to rest and cool down. He was almost 50 years old, and his heart did not allow him to finish the work.

His body had lost its freshness and pure energy and strength of about ten years ago. The problems at first were slight, but later, started escalating, but because he is a man of strong character, they have remained more or less unnoticeable.

My father's condition worsened so much that also to the outside world it became clear that something is wrong. Besides mowing the lawn, walking up the stairs also became an effort the cold and heat became harder to beat. Digestion developed into an everyday problem and his job became so strenuous that a switch to part-time work was necessary.

This was also the period when Fabry disease was diagnosed in the family. Sadly, he was not the only one. But that's just the way it is – that's nature and we can't do nothing about it as we are part of it. *In nature, there is nothing useless, not even uselessness itself* (Essays, Michel Eyquem de Montaigne).

At 53 years old, coronary disease had progressed, so operative intervention was necessary. That was a really difficult ordeal for him. Every operation on the heart in itself is very difficult and long-lasting, especially when they need to do more bypasses. When I saw him after surgery in an intensive care unit it was hard to say how he was feeling. He was pale and quiet, calm but not placid, the atmosphere in the room was tense, as there were ghosts in the room. But how should someone look after such a serious procedure? Certainly not rosy. Then I saw his wife, how she had tears on her face and then I was completely lost. Until a few days later he said: »A day after surgery I felt extremely well (smiling he was seen also by his wife) it is true that it was not easy to wake up from the anaesthesia and start to breathe independently, this is bad, but not the hardest thing. Then, after the second day something happened, the plot ... and I can still hear the sound of »irons«(defibrillator).« It must have been really bad, because he said that he will never have this type of surgery again... As we ourselves understand, he barely survived this crisis. Then he slowly recovered and it was never the same as before.

So it happened, that the man who had always been the fastest, became the slowest. He went into the mountains only as a companion to the starting point, an observer of the ascent, and as a driver for sightseeing tours. Only when he stepped on the skis was he transformed, for a while, into his old powerful image. Oh, he was a joy to watch! And not only was he faster than the others, he showed his childish and did a flip while skiing and skied down the icy part. Thank you Mother Nature for giving him the opportunity to do this, although to a very limited extent and almost to the end of life. This was one of the few activities in which he really enjoyed and even an outside observer could see his happiness.

Until then, the Centre for the Treatment of Fabry Disease was established, which regularly monitored the progress of the disease and integrated him into a treatment program. Treatment began when he was 52 years old and he was treated for seven years. At first, after he received the medicine he noticed improvement. Last year when he got the pacemaker, due to heart failure, he repeatedly complained that his treatment was not working and that his condition was worse. I however believed that attention from the Centre for treatment made him feel good and because of it he was feeling safe.

The Centre also enabled him to socialise with other patients and in particular, he could speak to the medical staff at any time who always helped him out. Therefore, even though every person is alone with their illness, he had someone to talk to.

The disease changes man, physically and mentally, and when he is no longer physically strong, the psyche begins to collapse. But, surprisingly it was not evident that this man's mental condition weakened, or was tormented with/by depression or anxiety, perhaps it was different at night, but during the day you never felt that the disease burdened him so much that it broke his will. It is true that otherwise he was not exactly a happy man. He was grim rather than smiley but it seemed that to him every day little things such as reading, television, music and walks satisfied him. I envy him for this, because it is a lot easier to simply go with the flow and collapse under the burdens of the world. I believe that a one of the main reasons for this was that he was in touch with nature. God knows what he had in mind when he went walking every day.
Later on, in addition to other problems, an infected wound appeared, which – to a healthy man would mostly not cause trouble, but to a 57-year-old Fabry sufferer, due to poor blood flow becomes persistent, long-lasting and intractable. Urgent surgery was necessary, but preparation for surgery itself was too much to handle for a wavering heart. He felt the upcoming end, but he just could not say goodbye on his last day with us. Soon afterwards another Fabry heart said goodbye.

Marija (52 years)

Optimism

Ever since I can remember, the disease which in my early years took my mum's life, was my life companion. Therefore, it was certainly a big shock to all of my family (husband, children, parents, brothers and sisters), when I was diagnosed for the inherited and incurable disease in the Slovenj Gradec General Hospital. There are no words, which can describe the intense feeling when I found out.

After thorough check-ups I was also »tagged« as a patient. I slowly settled with it and kept on living my life with a positive attitude. I did not have serious health problems only tiredness and digestion problems.

Together with my family I live according to a stable rhythm – job, home, holiday, leisure weekends that we spent in nature, hanging out with friends.

In short, everything a person could want – fulfilled life full of positive energy and happiness. But the time came when my doctor suggested that I start receiving a replacement enzyme.

For me it was a shock, but still I had no argument with the treatment. I had to adjust my life to »my new friend, Miss Fabry«.

It means a lot to me that my family and friends stand by my side in all aspects of my life, because in today's rapid tempo people walk past each other and do not have time for compassion, closeness and, conversation… I know that if I need them I can call anytime. **Now I am receiving the enzyme replacement for the ninth year. Changes are noticeable, not as I would have liked, but there are some. My quality of life has improved.**

However, I wish that medicine would invent a drug that you drink or eat because my veins do not like the stinging sensation and I can't administer the therapy by myself.

Marina (52 years)

Embrace of the Mountains

If I were asked for a key phrase to describe Fabry disease, I would not only answer, but blurt it out - uncontrollable neuropathic pain.

Ever since the lady, named Fabry, sneaked into our family and became a normal (although unwelcome) part of it, I know how it feels to see pain and suffering that comes with it. I am saying this not only as a Fabry patient, but as a mother of a wonderful teenager, who lives patiently and who has to breathe with his own pain.

Storžič – a mountain hike, planned by my husband with our nine-year old son. The mountain from which my husband and my son returned as different people. As they reached the top, they were still full of energy, but on their descent, my husband had to carry my son on his back, because his feet were in too much pain. Even today, Storžič remains a symbol of their perseverance and of their hope that they may still someday climb it, along with other Slovenian mountains. The photo taken on Storžič, which stands on my son's desk, shows immense pride in having conquered it. However, that same trip to the mountains left us wondering why this complication even happened in the first place. The explanation came soon afterwards.

The explanation for my son's worsened health situation and the difficulties he experienced when hiking showed up on my family's doorstep when I was five months pregnant with my fourth child. My son's sudden high fever, which would not let up for ten days, frightened us. We took him to the emergency room, because he was in severe pain, but the tests they took in the ward for infectious diseases did not show anything substantial. He had unusual red dots on his left thigh, so they referred us to a dermatologist as well. His fever started to go down in the next few days and his pain let up as well, while the check-up at the dermatologist, doctor Dragoš, left us with the suspicion that he had Fabry disease.

From that time on, we have been receiving strong support from the Centre for the Treatment of Fabry Disease in Slovenj Gradec. The blow we received when we had to face and accept the disease was softened by the professional and psychological approach of doctors Bojan Vujkovac and Andreja Cokan Vujkovac, for which we are extremely grateful to this day. They are honest, caring and highly professional; they were a strong pillar on which we leaned to get our breath back as we were being told about the disease. Of course, I had no clue at the time what kind of events our family would have to face in the future because of our son's disease.

As early as that fall, our ten-year-old son began enzyme replacement therapy, which inspired us with great hope that the problems would be lessened. But unfortunately, that wasn't the case. With each year, his neuropathic pain was getting worse and we had to begin dealing with them.

As a Fabry patient myself, I was only familiar with the burning pain in my feet and hands and the pain I felt as we were throwing snowballs at each other when I was a teenager. I also experienced strong »migraine« headaches now and then, which were in fact a symptom of Fabry disease. But despite all these issues, my life was normal: I was exercising, I loved to learn, to read, I ran a half marathon, I enjoyed learning new languages, I was happy with my job, and I still don't feel any limitations in performing it. My marriage and family life make me extremely happy.

The pain my son was facing now was new to me and I could not understand it at first. I felt that it was nothing he could not handle by himself and, later on, with our help. I have to admit that it was very difficult for me to accept it. For a long time I believed that the pain will subside and stop its horrible march. Unfortunately, that did not happen. We started looking for a solution: various analgesics, acupuncture, and alternative medicine … The result: his condition remained the same or got worse. We were utterly helpless. The pain always won, searing and defining our daily lives.

Our son was absent from school often, and I had trouble convincing his teachers that he was not a malingerer. It took them about three years to start believing me and stop reacting skeptically to his condition. It was the same story with his schoolmates, except they never changed their opinion; some of them even wished that they would be absent from school as often as my son was. They did not want to attend PE classes, because they did not feel like exercising, learning… My son told me many times how his schoolmates envy him for the concessions that needed to be made for him, for the absences from school, even for staying at the hospital! Sometimes it seems that the disease itself is not as bad as the cruelty of the environment reacting to it. Nevertheless, through perseverance, many talks, evidence and truthfulness, and human vulnerability, I entered into my son's world – open to all suspicions, but also to the humanity and support from some of the teachers and the principal, who understood our distress. Despite the pain he was in, my son had plans for his future and did much more to realize them than his healthy peers.

I remember some of his victories, which became the building blocks of his personality: writing his research paper (for which he later received an award) all night until early in the morning despite the pain – my husband and I were present at all times, of course; receiving all the reading badges (Slovenian, English, German); being an A student every year and also the only student who was praised for doing all his homework responsibly and with above average results despite his numerous absences. My son was able to attend his chosen high school without any fear or doubt.

When high school started, he did have some doubts whether he would be able to keep up with the curriculum because of his neuropathic pain. However, the initial doubts were laid to rest by the school psychologist, whom we talked to before the school year even began, and then throughout the year. The accommodations they made for my son were beyond anyone's expectations. My son could start high school with confidence. Doctor Bojan Vujkovac was kind enough to give a detailed medical presentation at my son's high school, because he knew how difficult it is for a nonprofessional to understand Fabry disease.

My son has now successfully finished two years of high school. Despite the neuropathic pain, he is an A student, which is a new victory for him. He keeps looking, researching and trying to trick the pain, because he still has not found the final solution to it despite professional medical help.

He is patiently waiting and making plans, and he strongly believes there are many wonderful things still to happen in his life, including the embrace of the mountains.

Cabi (45 let)

The View of Life Has Become Clearer and More Relative

My encounter with Fabry disease was cruel. It invited itself (we started hosting it) eight years ago and it struck my beloved 9-year-old son, with whom I looked forward to the variety of activities we would do together in life... Instead of climbing and skiing in the mountains, we had to start fight severe pain which appeared even with minimal exertion. We had to face the acceptance of a life which included all the limitations that disease gradually, but persistently brought.

It started with the supposition of doctor Dragoš. She was recommended to us, to examine closely, unexplained red rashes on my son's thigh. We had been told that the source of all of my son's trouble could be Fabry disease. Because I had never heard of the disease before, I immediately started to browse on the internet for information and with every described symptom the painful truth cut deeper inside. The hardest thing to take was that the disease is progressive and slowly takes over almost all of the body. It also became clear that the only hope that my son could live, more or less a normal life was to get treatment with enzyme's replacement as soon as possible. Official confirmation that my son had the disease was needed for this. Doctor Dragoš immediately established contacts with doctor Vujkovac, head of the Centre for the Treatment of Fabry disease in Slovenj Gradec to perform the genetic blood analysis and enzyme activities through specialised laboratories abroad.

Thanks to great connections with his colleagues from Hamburg, doctor Vujkovac finally revealed – my son and wife have Fabry disease.

Yet the first contact with the team from Slovenj Gradec gave me the impression that they were a very noble and self-sacrificing people, without whom there would not have been a path leading to the medicine. So it only took a month and regular two-week visits to the Paediatric clinic for infusion therapy to become part of our schedule. There we came in touch with doctor Tanšek. She was always ready to listen to our problems and comply with our wishes.

During our acclimatisation to the disease we received a lot of help and comfort from doctors Vujkovac and Tanšek. The feeling that people sincerely wanted to help was priceless. They both selflessly gave us their personal phone numbers and we often had long conversations outside their working hours.

Despite the expectation that the drug would deal with the disease in no time, our problems did not go away, particularly the pain in my son's feet and hands. By the age of puberty it worsened. Only recently has the pain subsided.

When I was recently in contact with sufferers from other countries, I came to the conclusion that we are very lucky in Slovenia, because sufferers can be treated with enzyme replacement therapy. Due to high costs in many other countries, this is sometimes not possible. Therefore, we are grateful and happy that my son can receive the therapy, that his health condition is stable, that he is up to the challenges of school and in particular, that he remains an optimist…

I'm glad that life keeps us on the »main road« despite frequent »crosswinds«. Because of living with Fabry disease my view of life has become clearer and more relative.

Franci (46 years)

Marked

I am grateful to be who I am – Fabry disease, which has marked me profoundly at only 17, is no exception.

No, I'm not crazy, nor have I forgotten the countless hours I've spent in severe pain, or the tears I cried at the start of my life because of the »damn« disease, and I'm writing this despite the fact that I feel neuropathic pain even at this moment.

And how did everything begin?

Even as a young boy, I took karate lessons. At first, I did not pay any special attention to the pain that I felt virtually all the time during practice. But as it got worse through the years, I started asking others in my karate class, if they also felt searing pain in their legs. Some of them said yes (they probably meant muscle pain), and so I went on with my life without being too concerned about the pain. Even a later painful memory did not change my opinion that nothing was wrong with me.

I remember going to the Atlantis water park with my class at the end of a school year. Because our school was relatively close to the water park, we went on foot. While I had no problems walking there, the way back to school was another story. Even in my dreams, I could not imagine how dreadful it would be. The heat was terrible. When we were about halfway to school, I started feeling a tremendous pain, which was getting worse with each step I took. I put some distance between me and my friends, so that I could wage a war in my head. While I was still winning the fight while walking, the pain finally got me on my knees once I entered the school building. I even got cramps because of the severe pain, and I started crying and bawling. The teacher was confused and asking me what was wrong. All I could do was scream: »It huuuuuuuuurts!« She took me to her classroom and called my parents. As I could not even touch the ground, my mom had to carry me home. After my body temperature stabilized, the pain went away after a couple of hours. Unfortunately, we did not take this event seriously either, and I tried to forget it ever happened.

However, a few months later, something happened that I could not just let slide... **I was 9 years old, when I got seriously ill – I will never forget those days. Virtually overnight, I came down with a high fever, without any signs of cold, just tremendous pain that accompanied the fever. I cried from pain, I even yelled out during the heaviest pain attacks.**

No one in my family knew what was wrong or how to help me. First, we went to the general practitioner, who was very concerned and referred us to the Department of Infectious Diseases. They could not find the answer to my terribly painful condition, either. Thankfully, during my exam, the doctor noticed I had angiokeratomas (red spots) on my right thigh. She advised us to see a dermatologist. By diagnosing my condition as Fabry disease, the dermatologist Vlasta Dragoš made my life my much easier. She referred me to the Center for the Treatment of Fabry Disease, where I was examined by internist nephrologist Bojan Vujkovac, who explained my illness to me in detail.

Days went by and I began the enzyme replacement therapy, although the pain would not subside. I felt pain with each physical activity, each cold, practically every day. As the pain grew worse from morning until afternoon, I was hardly able to get home after school, although it was just a few meters away. Every time I took this difficult journey, I had to rest, after which the pain let up and I was able to go on with my day in the late afternoon. This went on almost every day, with few exceptions.

Now that I was aware my pain was not as normal as I was telling myself at first, I became much more focused on this problem. As a result, I was in much more pain every day, on average.

But despite all the problems, I am getting to know my disease more and more, and the painful episodes I described above hardly ever happen anymore. I also got used to the fact that I feel the pain practically throughout the day. The only thing bothering me is that the disease limits my sports activities, which I really love. However, I see some opportunities here as well – lately, in the evening when the pain practically wears off, I try to strengthen my muscles as much as I can to improve my physical fitness. By doing so, I believe that my body will get used to activity and the pain will occur later, when I exert myself.

Things have not been as bad recently, particularly in the last year. As I already mentioned, I have gotten better at coping with pain, I am getting to know the disease and learning how to trick it. I am getting out of puberty and I can already feel the aging of my faithful »unwanted friend«. I have finished the first two years of high school successfully, and I can safely say that the disease has not limited me too much. I have a family that understands and supports me, and encourages me to look for and open new ways for myself. I am happy that my high school accepted my special requirements regarding absence and taking exams. I make individual arrangements with each teacher.

If someone who just found out they have Fabry disease asked me for advice, I would tell them not to look at the disease as the most terrible thing that could have happened to them, but merely as an obstacle in life. Whether they decide to jump over it or even tear it down is entirely up to them. For the strong-willed, the obstacle is certainly surmountable.

Despite my fears and the limitations it created, the disease has marked me mostly in a positive way. When I am feeling down for reasons other than the disease, this experience helps me get back on my feet. This is why I am grateful.

Franc (17 years)

My Brother And I Had a Childhood Filled With Games…

Fabry disease introduced itself to our family in 2006, when the conglomeration of symptoms exhibited by my younger brother got a single name – Fabry disease. Although the diagnosis greatly upset our family life, I never saw my brother as an ill person. I am still not sure, whether it was a defense mechanism that would not let his illness get to me and change my perception of him or it was something else.

Our childhood was full of games, playfulness, sports and happiness. Although my brother has always been on the quiet side, he would always get involved in his big sister's (that's me) shenanigans and ideas. However, around the time he turned six, he began having problems enduring high temperatures. It was particularly bad each summer holiday at the seaside, when one could hear yelling from our vacation home and feel my parent's desperation, as they were unable to help their child by alleviating his unbearable pain. The pain occurred every time his body overheated. He often had fever as well and would suffer immensely. I often witnessed the desperate look on my father's face, who was otherwise an eternal optimist, the tears of my mother, who never wanted anything but the best for her children, and the sighs and tears of my grandmother, who took care of us lovingly throughout our childhood.

As for me, I understood my brother's sadness and suffering, but I was never able to comprehend that he was ill. When summer came, which was the time for hanging out with friends, I encouraged my brother several times to go out with his friends. Everyone in my family was appalled at my suggestions, because »what will happen, if the pain comes after he is out for several hours« or »how is he supposed to walk from the cinema to the pizza restaurant in this sun and in this heat…«. I could never understand the fact that my brother required much more planning ahead and that most of my suggestions couldn't be realized for completely trivial reasons.

Unlike me, my brother accepted his disease completely, and was aware of all the problems and restrictions that came with it. That is why he would often say that he is not going anywhere so as not to burden his parents and his environment. He also became highly sensible to his pain. He discovered what causes him pain (when his body became too warm, when he was tired…) and tried to avoid those conditions as much as possible. All this time, he has been a wonderful example of strength, will and optimism for me.

I admire him for not giving up on life, for setting goals for himself and looking for hobbies he enjoys. Above all, I value his efforts not to burden our family with his illness.

Jasna (22 years)

Proud of My Older Brother

I always judge my brother by his mood. Every time he is grumpy or shut up in his room, I know that he is not feeling well. But when my brother is happy our family is filled with joy.

I never really thought about his illness. Sometimes I even envied him, because he could stay home due to illness, and not go to school. It's so hard to believe that my brother is sick, because he was always like one of the rest: when he feels good, he plays soccer with his younger brother, he loves jokes, takes the dog for a walk, he is pleasant to talk to, he does some housework, he associates with his friend and he is also successful at school. I think my brother is a hero, because thus far he has endured so much, as I never have, and I hope that never will. Although he faces struggles with his pain, he remains optimistic.

Therefore, I am very proud of my older brother and he is my role model. He has shown me how to solve problems to obstacles in life.

Dora (12 years)

He Prefers Lakes to the Sea

My older brother has been ill since birth. Sometimes he is quite grumpy with me and my sister, he really likes playing football with me, I enjoy my time with him-then. **He rarely goes with us on trips.** He loves sports: football, basketball, tennis and regularly watches matches and reads the sports news and results. He likes swimming. Last year, he swam the width of Lake Bohinj. He prefers lakes to the sea. Sometimes he scares me. Mummy scolds him when he has fizzy drinks and unhealthy food. I do not like it, when he does not want to play with me, but I really like when he is nice with me.

Pavel (8 years)

Grandmother, Mother, Wife and More…

To live as a wife and mother and not knowing that your husband and your daughter are ill is a burden you take on your shoulders and to your heart the moment you find out. The shadows of the genetic disease only took on a clear form after my 9-year-old grandson was diagnosed with it. It was only afterwards that my daughter – a carrier – and my late husband were also diagnosed. I was shocked by the fact that so many members of my immediate family had such a serious disease.

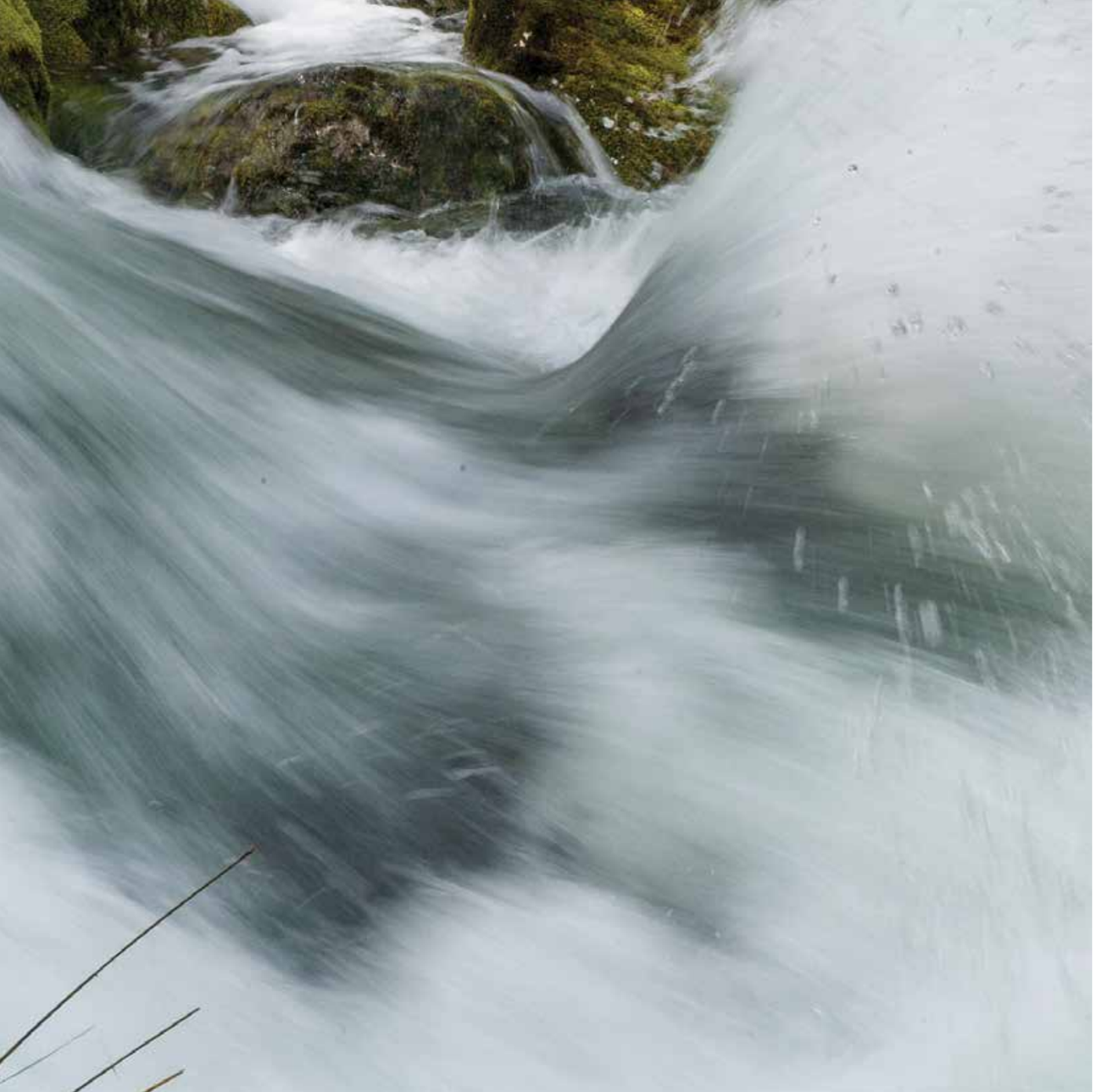
I will never forget the day when the disease was discovered – a painful moment that I wish immensely had never happened. When my grandson was diagnosed, we also found an explanation and a diagnosis for all the symptoms my husband and my daughter had that neither I nor the doctors could figure out: his unbearable pain throughout his body by the end of his life, my husband's depression, my daughter's searing pain in her feet and hands when she was an adolescent, and the headaches. All this was explained by the Fabry disease diagnosis.

And yet, my husband an my daughter were great at overcoming their problems. They exercised regularly, ran the marathon and the half marathon, and went hiking, so I never saw them as patients. We lived like any normal healthy family. Maybe even better.

It has been 8 years since my grandson started the enzyme replacement therapy. I help him as much as he wishes: by driving him places, preparing him lunch or snacks…

However, I mostly help him by treating and seeing him as a person who is capable of handling his own problems.

Mila (69 years)



The Wind »Ruffling« Our Lives…

At first glance, we look like a completely normal family with normal problems, and yet, we are not. My family, which looks average when you first look at it, has to face the truth each day at the time, and each day is a new struggle. However, each struggle also means hope.

Not so long ago – only six years ago – my husband and I, and our two girls lived like all regular families. We had wishes and needs like most others, until… until disease came into our lives. We found out that my husband has a rare genetic disease that has remained incurable so far. This news was followed by others that were far from pleasant and gave us no hope. Our entire world shattered into a million pieces. I tried to pick up the pieces and put together a new, smaller and different world, but every time I laid down new foundations, they were torn down by more news about the disease and my husband's health.

Our lives turned upside down. Fear and pain came, growing stronger every day, seeming unbearable. We felt like we were in the middle of a dark tunnel, not knowing which way to face and how to continue on our journey. We were running around in circles in the middle of this mysterious tunnel, lost and frightened. You cannot imagine all the things that change when disease comes into a family. In addition to the fact that a family member is ill and faces a disease, your social status changes, you lose or change work, or you come up against financial problems… All this stress and tension changes a person, weakens a family, no matter how strong, and compromises relationships. Each person tries to face the situation in their own way – some through sadness, others through anger, still others through fear… and the pain is always there.

What about our family today? We still have not given up. We fight, each in our own way. We try to live as any other average family, except that while others are choosing the destinations they want to visit on their holidays, we first look for dialysis centers and hospitals nearby before deciding on where to spend our holidays. Last minute deals are out of the questions for us, because we usually have to prepare and send all the necessary documents months in advance. Every time I go on vacation, fear follows me, because I feel safest at home, in Slovenia. But I have to hide my fear for the sake of my husband and children, if for no other reason, pretending that everything is okay, and go on a holiday outside Slovenia. I know that you must not let fear control you.

And our girls? They have known since day one what is going on with our family. I felt that this would make it easier for them to accept their father as he is. My husband and I believed that it would be easier for us to live a normal life, if we talk to them and do not hide the illness, which is a part of our lives now anyway. It was difficult at first, because my husband and I spent a lot of time in hospitals and clinics and the girls were in the care of different people at the time – friends, relatives, and neighbors. I would like to take this opportunity to thank all of them for their care, help and support. Each of them left their mark, helped us in their own way to get back

on our feet, and helped the girls perceive this difference as a normal way of life. Today, all this is familiar to them, and they sometimes go visit their father during his dialysis treatment, where they lie down next to him without any fear. In school, they have written essays about their father, who is ill but still loves them endlessly. He likes to play with them, and his disease does not make him any different from other dads. Of course, there were times when the girls were scared or hurt, but at those times, we turn to our child psychologist, doctor Ahčin. I remember one time when I was working the night shift and my husband called me to say he had a stroke. It was difficult for him to talk and he could not move. That time, my older daughter, despite being scared and in tears, played an important role and replaced me in my absence. She helped her dad, unlocked the door and waited for the ambulance, all the time making sure that her younger sister slept through the entire event, thus protecting her from the pain and fear, even if she was frightened and sad herself. Nevertheless, we lived through that as well.

What about me? What can I say? I took on a great burden, one that is usually shared by two people. I've been depressed, I've been desperate, I've been sad, angry and afraid, but I couldn't show it, because everyone around me had broken, and someone had to ensure the family gets back on track. For five years, it was as if I were sleeping. It was as if I weren't living my own life, with locked away emotions, hidden pain and suppressed fear. I was terrified of the future and the things to come. At night, I woke up many times, looked at my sleeping husband and cried. Sometimes I was afraid of the morning – waking up – I am still afraid of that - that I'll wake up and he'll no longer be with us. I am afraid that our little girls will be left without a father when they need him the most. I am afraid to think of the time that he will not be returning home with three flowers or chocolate hearts – one for each of his girls.

However, a life lived in fear is a life half lived. This is why I woke up from this world of pain and fear and decided to do something for myself and for others. I have adjusted and learned to live with what I have. I got my life back. Once more, I have secret desires, my happiness back... I have not stopped, I am trying to realize my dreams and my family is supporting me. I still get sad, angry and afraid sometimes, but I am also happy and grateful, remaining strong – always. Finally, we decided and changed our course. We are heading out of the tunnel. We have begun a new life, even if we do not know how long it will last. My husband is taking a medication that could help slow down the spread of the disease. We are living one day at a time, and while others are living their lives to the fullest, we are picking up life's breadcrumbs and moments, using them to build our own world, one day at a time. The time we spend together is important to us, all the tiny moments that signify life and without which there is no happiness. Every minute we spend together means happiness and joy to us, and each morning gives us new hope. We cannot sail against the wind ruffling our lives, but we can change its direction, so that the ruffles are smaller. We are prepared for the worst, we bravely march towards destiny and we are proving that things are not as gloomy as they seem.

Where there is a will, there is a way!

Zvezdana (36 years)

My Dad

When I was little, my dad got sick. **The disease caused him renal failure, therefore every Monday, Wednesday and Friday he has to go to haemodialysis.**

Now, he is not allowed to eat every type of food. Sometimes, when my mother is not around, he secretly drinks a drink, although he should not. When mother finds out, she gets very angry. Sometimes, even dad is a little angry and sad, because some food is bad for him. Although he has this disease, to me, everything seems quite the same as before. He plays with me.

Lara (8 years)

My Farm Keeps Me Going…

My mother was a Fabry patient. Up to the age of 60 she was healthy, then she had a stroke and died. My brother Vinko is also a Fabry patient. We have a farm and there is always something to do. I take care of 14 heads of cattle; we also have pigs, chickens, three dogs and four cats. Now

I am retired now, because I have a category 3 disability.

Because of my success, I have nice memories of my school years. I did not have serious health problems. **In elementary school I had a burning pain in my hands and feet. Since I was born I have had red spots on my skin, which do not bother me.** I remember that when I walked for a long time and was exposed to the sun I had to stop several times, because of the pain. After this I sat down and then began to sweat only from my forehead.

In my early years I loved sport, so I trained full contact-karate and football, which I played up until the age of 42. I trained to be a carpenter. As a 19-year old I started working in the Trbovlje mine. It was hard physical work over the conveyor belt. I had to dig under a bunker several times. After fifteen years of working in the mines, I became redundant and then worked for a construction company. Again, I was doing heavy physical work. After I reached the age of 24, pain in the arms and legs disappeared.

Ten years ago, the problems worsened. I was driving a truck down the road, when I lost consciousness. For some time my heart stopped beating. After this I got a pacemaker. In May this year I started getting dizzy which is getting worse. In 2009, I had a slight stroke and since then the mobility of my right sight has deteriorated. Because of this I stopped playing football. When I walk, I am sometimes unstable. This is very annoying and hinders me. I have muscle pain only when I walk down a hill. I like to rime my four-wheeler through the hills of Zagorje. I live with my partner with whom I have a 12-year-old son who is healthy.

Marjan (48 years)

Red Spots

When I was seven, I began to feel terrible pains in my arms and legs. It was strong and searing. Sometimes the pain was so bad that the only way I could move was with a wheelchair. It lasted for about 10 years. Once, I was admitted to the Peter Držaj Hospital in Ljubljana, where I was diagnosed with rheumatism. Ever since my childhood, red spots have been appearing on my skin. As a child, I could not tolerate heat or cold. When it was cold, my fingers turned very pale, and when they warmed up, it really hurt.

I was a good student. I could not play sports. One of my brothers died when he was 35 – they say the reason was multiple sclerosis. My mother had Fabry disease as well, and so does my brother Marjan. My 22-year-old son is healthy, while my daughter died at birth. My brother and I are receiving treatment at the Center for the Treatment of Fabry Disease in Slovenj Gradec.

After finishing high school, I began working as a mail carrier. I rode the motorcycle in all weather conditions. Many times, I felt pain in my arms and legs, so I rested. I also got frostbite. I was hurt in a car accident, so I have screws in my leg. Later, I spent 15 years doing manual labor, working in separation in the Trbovlje mine. Since then, I have been having problems with my hearing. I have always sweated less than other people.

I have problems with my vascular system. I have dialysis treatment regularly, because Fabry disease caused my liver to fail. I got a pacemaker because of my heart condition. I received electrical shocks several times because I had arrhythmia. I also survived a heart attack.

I am happily married and I live on a farm. I never run out of work. We have 60 acres of arable land, 4 heads of cattle, chickens and two dogs. I really enjoy the beautiful nature of the Zagorje hills.

Vinko (1957–2013)

When One Is Constantly Sick…

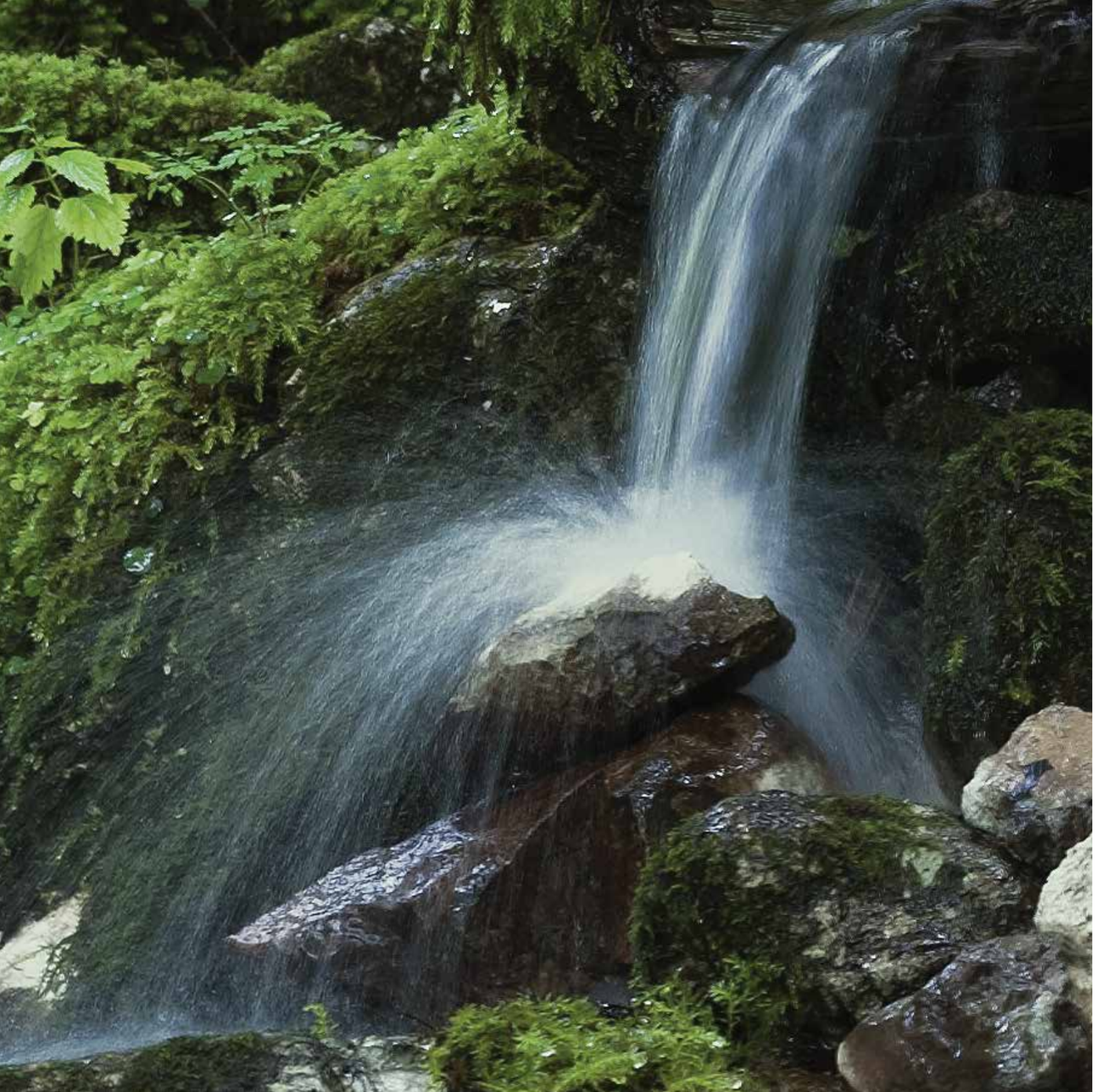
Very early in my childhood – when I was five or six years old – I felt terrible pain all over my body. Mum and I visited several doctors. My mother was very exhausted because of my health condition, as she was tense and anxious. The pain could be compared with putting your hands on a hot stove. Later insomnia set in. Check-ups with the doctor unfortunately, did not point to any disease, so they thought that I was faking'.

I remember how often I looked for shade, and hiding to splash cold water on myself. I had a bad tolerance of physical exertion and heat. My skin, feet, and hands were smarting me. My face got swollen. Even a little effort was exhausting for me. Many times I was forced to give excuses and go back to the toilet.

It was very difficult, for me to participate in sports days, school trips, hiking or even PE. The worst part was when I should have served my military service, which of course I could not do. Many times I came home crying and I thought I would commit suicide. It was like living with the feeling that I would jump to the sky, if I only could). Of course, no one understood my pain and feelings, because I looked like a healthy man.

In situations of severe pain I had to put vinegar compresses on my hands and feet and shower with cold water to ease my body heat. Over the years I have noticed, that I am different from my brother and peers…

Igor (45 years)



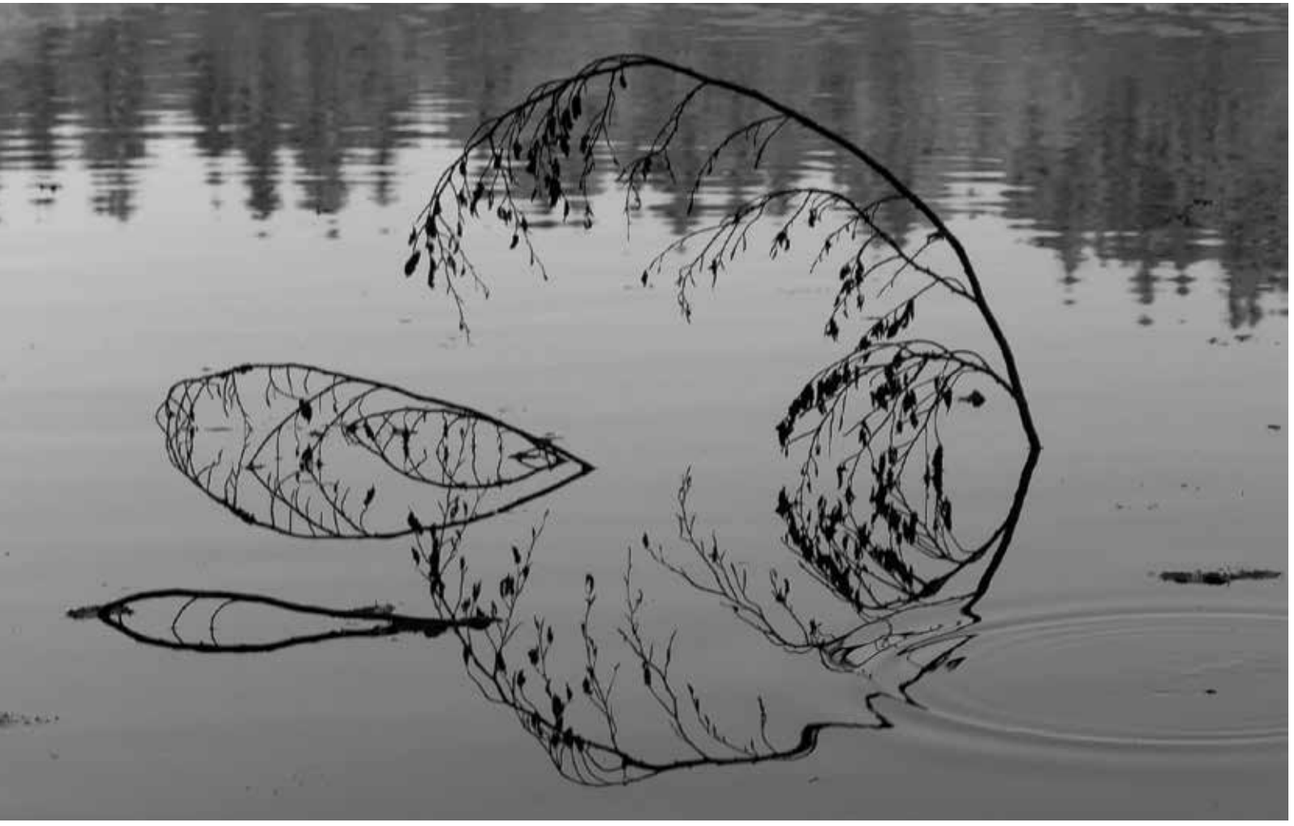
And the Disease Gnaws Away…

It was hard for me to force myself to start combing through my memories, which are fraught with pain and sadness. Eventually, the sadness retreats into the depths and eventually nice memories float to the surface. It seems to me that this writing will not be a realistic representation of events. There was too much pain and sadness in the time I have spent together with my husband.

I met my husband when we were still young and we decided to continue our path through life together. We have created a family. My beloved husband was mentally strong with solid personality and an unending source of energy. He never spoke much about any health issues, except that he had a hard time tolerating heat and that he was suffering from indigestion. It seemed that these problems were not a result of his lifestyle. He valued healthy food and ate in moderation. I believe that he was strong and stubborn in facing with his health issues, which were becoming more severe through the years and were joined by new problems. Of course, he only managed to keep on going until the disease had progressed too far. **After he turned forty, his face bore the marks of unending problems and fatigue. He came from work increasingly tired and irritable. His ankles became swollen. Eventually, the swelling never went down but began to spread upwards towards the knee. The more he neared his fiftieth birthday, the more difficult it became for him to deal with his illness. His problems were becoming more pronounced and there were more of them. Increasingly, he complained of burning chest pain. He began having trouble breathing, even at night. He got increasingly severe cramps in his legs.** It became increasingly difficult for him to bear any type of exertion, so he asked for reduced working hours. Despite taking medication, walking up the stairs or even just a stroll around the house was increasingly difficult. During this period, he began with his replacement enzyme therapy. He hoped that his problems would become less severe. For a while, some health issues were less severe. He mentioned several times, that it was time to go for the second dose. But despite the therapy and several types of medication, his heart-related problems became more pronounced. Surgery was necessary. He underwent quadruple bypass surgery. His recovery was very slow. Between the recovery after surgery and another turn for the worse, his health condition was relatively stable for three years. Then his heart began to weaken again and he had a pacemaker implanted. However, it did not help much. During his last year he often complained that he felt worse after therapy. It is hard to make any sense of the events that happened in the last couple of years before his death. A person cannot comprehend how an illness of a loved one can impact the family life. Five years after surgery, his heart stopped forever.

This is how I saw and lived through his fight with the illness that he never liked to talk about. He always turned the conversation to other topics. The illness slowly began to gnaw away inside him when he was at the height of his strength and most creative. He had so many goals and wishes that he wanted to fulfill. However, when he should have been reaping what he had sown, the disease threw its most powerful punch. Despite the treatment, supported with medication and surgery, it seems that the illness developed unimpeded and ended his journey. I was left with a painful experience and untold sadness.

Valentina (48 years)



Fear Within Me…

I have thought long and hard whether I should write a couple of lines about my life story and my disease. There were six of us in my family. Dad, mom and four children. Our childhood was tough, as our dad was an alcoholic. I remember I was a very sickly child, just like my brother. I could not learn a profession as I had difficulties learning and we did not have the money. That was why I got a job after high school. I found employment in a Gorenje factory in Velenje and later found a position in another Gorenje factory in Nazarje, which was closer to home. I have worked there to this day.

Because of the pain, I often had to take sick leave. I saw many doctors and all of them told me that I was suffering from rheumatism, that it is no big deal and that I will have to get used to the pain. They prescribed analgesics and I ate them like candy, because I was suffering from severe pain. This way, I could at least work more as I was having difficulties getting sick leave. I remember the surprise on the faces of the members of the Invalidity Committee. They believed that as a 36-year-old I had no business asking for disability pension. However, even at that young age I had great difficulties performing my work. I had to take large numbers of pain pills to do any work. Each day, I was so exhausted that I collapsed onto the bed after arriving home from work. I rested for a bit, but I had to take care of my entire family. I had a husband and a son. My problems were compounded by the fact that my husband was an alcoholic and that I had to do everything on my own. Since I was a child, my greatest fear was that I would marry an alcoholic. I suffered greatly because of my addicted father. That was exactly what happened.

Without a doubt, the greatest event in my life was the birth of my son. It gave me more strength to keep going. But when you have a child, you have to stay awake all day and all night long. I do not know where I found the strength to raise him. Day after day I fought the pain and worked hard to ensure that he learned a profession.

After my brother became seriously ill and experienced renal failure, he was diagnosed with Fabry disease. That is why me and my sister got tested. I quietly hoped that I did not have it, but in light of all my problems, it was confirmed that I have the disease. At first, after hearing the diagnosis, I was in shock but then I slowly accepted it and fought on. Thank God there is a cure. I wish to thank the General hospital in Slovenj Gradec, which also hosts the center for this disease. I particularly want to thank doctors Bojan Vujkovac and Andreja Cokan Vujkovac and nurses. I thank them for their kindness and patience, which is important to us patients.

I now work 4 hours per day, which greatly relieved my burden. Every two weeks, I receive treatment (replacement enzyme) in the Center for the Treatment of Fabry Disease. At work, they understand that I am ill and that I work as much as I can. But I am sorry to say that at home they still do not understand that I am ill. I still have to perform all household chores on my own, even though it is sometimes very hard to do. I am very stubborn and keep fighting, even though I sometimes just want to collapse onto the floor. However, if I were not so stubborn and patient, I would surely have never made it. The same goes for my brother.

I also want to write this – things would have never been as good as they are, if it were not for the doctors in Slovenj Gradec, who have helped us with their treatments, advice and talks. Of course, my stubbornness also played a part. I again wish to think the healthcare personnel in Genral Hospital Slovenj Gradec. I wish patience and success in their treatments to other patients, my brothers in suffering.

Terezija (54 years)

We Are Not Second-class Citizens…

Even as a child, I had numerous health problems: frequent fevers, flu, pains all over my body, heat episodes that felt as if somebody put hot coals in my hands and feet. **Whenever I visited a doctor, he told me I was a malingerer, who does not want to go to school and, later, work. I felt greatly distressed because not even my family (mother) did not believe me about the pain I was feeling.**

Tests (blood, urine) never showed anything out of the ordinary, so not even doctors believed my problems. At the dermatological department at the General Hospital Celje, they used liquid nitrogen to burn away the angiokeratoma across my entire body. The procedure was very painful and unpleasant so I was always hiding from my mother before new procedures and hid my condition, so I would not have to undergo any more treatments.

These pains influenced me greatly and I stopped seeing doctors. I became withdrawn and secretly took pain medication from my mother.

In elementary school, physical education was an enormous challenge. The teacher refused to believe how hard it was for me to be physically active. Even getting to school, which was 500 meters from our house, was a tremendous effort for me. On my way to school, I had to rest up to 15 times.

Then the time came for me to attend high school. The school was 5 kilometers away and I again had to attend physical education and practical training. The story from elementary school repeated itself. Nobody believed that I was in terrible pain, which was compounded by poor mental and physical state.

Despite that, I managed to complete the Technical High School as a precision mechanic. My father's friend, who owned a precision mechanic workshop, solved my troubles with internship. After completing my internship, the time came for me to perform my military service. Because of my intense pain, I was avoiding the service, which landed me in trouble with the Misdemeanors court and resulted in me having to pay a fine.

This was followed by years I spent searching for myself – I had no real will to live. My mother

died before she was diagnosed. She died without knowing about her - and my – disease. She was suffering from severe health issues: he had a heart attack and was immobile for 6 months, after which she received a pacemaker. After my mother died in 2004, I was left alone.

This was a very difficult period of my life as I was used to living with my mother. My father died when I was two.

In 2005, I met my current partner, who is very supportive and forced me to see a doctor again. Luckily, I encountered a very good dermatologist at the General Hospital Celje, who took an interest in me. Before that, specialists from different fields, who never found anything, examined me. The dermatologist from Celje then referred me to doctor Bojan Vujkovac for an examination in the Center for the Treatment of Fabry Disease at General Hospital Slovenj Gradec. After examining me, he told me that he is almost 100-percent sure that I have Fabry disease and wanted me to have additional tests done, which confirmed his suspicions. Before I was diagnosed, I visited at least 25 doctors in 25 years.

After I got the diagnosis, the first thing I did was to go online to try to find out what this disease actually is. Reading about it, I was frightened, horrified and filled with negative emotion.

People in the General Hospital Slovenj Gradec gave me the emotional support to overcome my problems and I know that I was helped in many ways by honest talks with doctors Bojan Vujkovac and Andreja Cokan Vujkovac and other doctors and healthcare professionals. Of course, my partner gave me the greatest support with all the difficulties I encountered.

I feel the greatest problem is that patients with Fabry disease are regarded as second-class citizens and that even addicts get priority before us. All Fabry disease patients need to have their status fixed and should receive full social relief.

I could write more, as there are many challenges I have not even touched upon in my writing. I hope that I contributed at least a little to raising the awareness about this serious and rare chronic disease. I hope that people will listen to us briefly and believe the challenges, terrible pain and unpredictable complications people suffering from Fabry disease have to face.

Matjaž (41 years)

How Will I Endure This?

We made the connection only when my son was tested and diagnosed with renal failure.

I am 60 years old and five years ago, I was diagnosed with Fabry disease. We never knew about it until my son was diagnosed with it.

When I was very young, I started getting headaches and not a day has passed without me getting one. Doctors always diagnosed me with migraine. I often felt apathetic and weak and all too often my only wish was to stay in bed for the entire day. My despondency was misunderstood – first at school and later at work; even my own family misunderstood it. They told me that I was simply lazy.

When the time came for my husband and me to take our children vacationing to the seaside, I was always worrying about only one thing: »How will I endure this? How will I endure the heat, the sun and sunbathing on the beach?« However, it was clear to me that I needed to do it; otherwise, I would have to face reproaches from my family that I refused to take them on holiday. **This intense intolerance of the sun and heat was never linked to an illness; not even my doctor ever suspected anything.**

We made the connection only when my son was tested and diagnosed with renal failure. After running various tests, he was diagnosed with Fabry disease. After my son's diagnosis was confirmed, I was tested along with my daughter, my three sisters and their sons. The doctors found that only me and my son were suffering from the disease.

Since I have begun treatments at the General Hospital in Slovenj Gradec, I have been feeling much better; however, I am not well enough to say that I am completely healthy.

Lenka (60 years)

I Trust My Family

In 2012, after a myocardial biopsy and additional tests at the Center for the Treatment of Fabry Disease at the General Hospital Slovenj Gradec, I found out that I am suffering from Fabry disease.

I was not too surprised when I found out. I began looking online to find more information about the disease and to see, if it can be treated and how. I put my trust in my family, friends and coworkers.

I had no troubles with accepting the disease, as I had had medical issues for years but could not explain them. The disease was most challenging during physical exertion and I got tired quickly. In my youth I played handball. After completing my military service, I got married. This meant increased family obligations and I gave up sports.

I am glad that my family and the environment in which I live have a completely normal reaction to my condition. I am working hard to ensure that people do not pay too much attention to me. I continue performing my duties at home and at work. Everybody is prepared to help me as much as needed.

I do not think about the dark moments of my disease and prefer to focus on the positives. I am in the 37th year of my employment term and am nearing the retirement age. My wife and I have been together for thirty-three years. We have a son and a daughter. They are both independent and living on their own. My greatest happiness are my two grandsons. If I have the time, I like to go out for coffee with my friends.

The only advice I want to share with other people suffering from Fabry disease is to think positive.

Novo (56 years)

Experience With Home Treatment for Fabry Disease

We had heard about rare hereditary diseases, but we had never heard of Fabry disease. We had also heard about home treatment. Both – Fabry disease and home-based therapy - became our reality in 2009.

Ever since childhood, my husband Tomi has had different troubles that can now be explained as symptoms of Fabry disease. Since 2004 he suffered from serious heart problems. Despite treatment, every couple of months his heart went into atrial fibrillation with a very high heart rate. Each time, a defibrillation with electrical cardioversion was required. After one of these episodes, doctor David Žižek, an internal medicine resident at the time, consulted with his father doctor Bogomir Žižek about his suspicions that my husband was suffering from Fabry disease. The diagnosis was made in March 2009 and in October he began enzyme replacement therapy.

During initial examinations and before beginning with the enzyme therapy, we met with the selfless and motivated team, led by doctor Bojan Vujkovac, in the hospital in Slovenj Gradec, which hosts the Center for the Treatment of Fabry Disease. Very soon, they told us that the enzyme therapy could also be performed at home. Tomi underwent the enzyme treatment without any issues, so we thought it was an excellent option. Every fourteen days, I accompanied Tomi to Slovenj Gradec, where nurse Vesna Korat, who coordinates work of the Center for the Treatment of Fabry Disease, walked us through the procedure in detail. I had no problems preparing the enzyme solution for the infusion and keeping it sterile, as I work in a microbiology laboratory. It was somewhat more challenging to insert the intravenous cannula. It was not so much that the technique was difficult; instead, I was afraid that I would cause pain or hurt a person, particularly someone so close to me. I turned to the Medical Faculty in Ljubljana for help, where they were kind enough to allow me over an hour of practice on a hand model with veins. The first »live model« that I inserted the IV cannula into was nurse Vesna, who calmly and bravely watched my shaking hands. Everything went well and on 24th December – under Vesna's supervision – I inserted the intravenous catheter into Tomi's arm for the first time. That was when we received three-month supplies to administer the infusion as well as phone numbers that we could call for advice, if something went wrong.

At home, we realized that kitchen is the best place to administer the infusion and insert the intravenous cannula. The kitchen table is the easiest to clean and sterilize and offers a lot of

space. Afterwards, we prepared the hooks for suspending the IV bag (we used similar hooks that we use for baskets, when we pick cherries, although ours were a bit smaller). We placed one of the hooks in the kitchen for when the treatment was being prepared, while we installed the other on a bookshelf above the couch for during the therapy. We decided to perform the first therapy in the morning, during work hours, when all medical services would have been available, if something had gone wrong. Everything went smoothly but we kept up the morning ritual for several months and once, it certainly turned out to be the right decision.

In March 2010, I just could not find and hit the right vein to set up the cannula. After several attempts, Tomi and I were becoming slightly nervous. My hands were shaking and the veins »hid« even more, so we decided to stop trying and seek help at the nearby healthcare center. I filled up a bag with the required material, documentation and the prepared infusion solution. At the healthcare center, I knocked on the door of the phlebotomy lab and explained my problem. Even though the nurses had no experience in setting up an IV cannula, they were glad to help me, found the right vein and helped me set up the infusion. Just in case, we walked home with the cannula in place.

When thinking of how to best solve our problems, if another similar situation arose, we realized that we count a nurse and a male healthcare professional among our friends. Both have experience with inserting intravenous cannulas. They both agreed to come to our aid in such events, if they are home, of course. During all this time, we needed their help twice.

In time, we gained experience and learned different tricks. We found that moderate whole body exercise (around 30 minutes) fills up the veins better and makes them easier to see. A couple of minutes of squeezing a ball also helps with that. Any physical activities helps, including biking, gardening, shoveling snow… another helpful activity is vacuuming the apartment before the infusion, which has become routine in our house. In addition to exercise, a warm environment is also important to ensure warmth and good blood flow. Drinking slightly larger amounts of liquid before the infusion also contributes to making veins fuller and more visible. When thinking about the hospital, where they usually insert the IV cannula on their first try and without any problems, these »preparations« might seem extreme, but you have to take into account that things are different at home. Healthcare professionals in the hospital are trained and have to perform the procedure several times a day. If things go wrong, there is help on hand. I only do it once every two weeks and have much less practical experience, which makes every little thing that makes it easier to insert the IV cannula very welcome. Tomi and I are also careful to change the location of the insertion point and to insert the IV on different arms. We avoid the elbow joint, which is usually used during examinations to draw blood, and use veins lower on the forearm.

It will soon be 4 years since we have begun performing the treatment at home and all the advantages this brings definitely outweigh any occasional headaches. The treatment can be delivered within a four-day window and can be adjusted to our needs and other activities. We can choose the time of day and these days we usually begin at 5 p.m. We don't have to go to a healthcare center and while the infusion is administered, Tomi can use the computer, watch television, eat dinner, talk to visitors or sleep for a while. We administered the infusion twice on skiing vacation in Italy and once, while we were on holiday in Croatia. Of course, we put a lot of effort into transporting the enzyme and ensured that it was stored at the correct temperature at all times. I also carefully prepared the package of things we need to administer the infusion.

Of course, people are different and we all have our own different fears, reservations and skills. However, I would like to encourage people, who are thinking of home treatment to not get scared. It is useful, if the person administering the therapy and the patient actively participate, help each other, talk about problems and cooperate. Most of us are capable of learning how to perform the enzyme therapy at home, getting through initial challenges and find a way, even if not everything is going smoothly. The great advantages of home-based therapy are less reliance on healthcare institutions, comfort of staying at home during therapy, time flexibility and others.

Katarina

Enzyme infusion during skiing holidays in Italy March 2011, under careful supervision of an expert.

6. Don’t Let Everything Revolve Around the Disease!

Never, never, never give up!

W. Churchill

From the Miracle of Birth to the Shadows of Disease

As any childbirth astonishes and shocks the mother so does the news that her child may be seriously ill. It is amazing how thin is the borderline between the joy of a healthy and careless life and the »curse« of the disease which hovers over the family like a threatening cloud.

When expecting four miracles of life I hardly imagined that there is another companion coming to our family - a rare chronic disease. When this fact hit in our minds then a period of sleepless nights filled with painful questioning and even crying began. Sometimes waves of despair brought us completely down, but we rose again with the hope to be able to mitigate son's disease by the help of the enzyme replacement therapy and different choices of pain relief drugs. We strongly endeavoured to adapt our life in such a way as to reduce the load of disease with different faces: face with burning pain in feet and the hands, intestinal disorders, severe headaches... And all the time we tried to fool the disease and ourselves, as we can easily go forward, upbeat and active.

The Loving Heart Is Difficult to Outsmart

But the loving heart is difficult to outsmart. Feelings of fear and insecurity, sometimes anxiety were becoming stronger and louder. As a family we were more and more rarely leaving for a trip, we abandoned arduous activities, and we were getting tired of the repeating questions of our relatives and friends: »Where is your son, is he unwell again?«

Open Discussion About the Disease

Although we were very open with our friends and relatives, once we faced the son's disease, too much discussion about it soon became quite annoying. People can be sometimes quite clumsy when talking about other people's disease, death, taboos... Indeed, a great deal of sensitivity and subtlety is needed in order to properly perceive somebody in trouble. Only after a few years, we learned that we did not need to be very comprehensive in explaining each detail of son's health problems. We felt some relief.

With Every Disease, Distress, Fears, Worries and Black Thoughts Are Accumulating

All the distress, fears, worries and black thoughts that I fought with steadily and slowly accumulate until they threatened my health. I needed an urgent »sobering«. During my sick leave I started thinking about myself, about my life, about the needs of the whole family, about the fact that we will probably never completely free from the bondage to son's disease. I decided to take such a way as to avoid loading my son too much with my fears.

When One Family Member Gets Ill, Every Other Member of the Family Should Necessarily Also Take Care for Himself. The Disease Should Not Be in the Focus of the Family!

I realized that I need to create a distance to the son's disease and to define a solid source of energy which would regain my confidence in positive aspect of life. I realized also that I deliberately need to take time for myself, my husband and other family members. Only the solid inner balance can be the basis for the healthy community, spontaneity and sensitivity. To maintain a balance between the disease on one, and pleasant moments of life on the other side, is extremely important, because the disease constantly challenges the mood of the family, relations, and changes directions the family wants to take.

Inner, Personal Responsibility of the Ill Family Member

In this struggle to cope with my own fears it was also my son who took a step towards me. With his exceptional sensitivity he recognized my fears and clearly delimited the teritory saying »From now on I will take control of my disease and my life. I'll let you know, when I need your help.« This has happened after countless night-hours of open conversations about our thoughts, feelings and fears. Experiencing such moments with the children and the husband is mercy and blessing. They are rare, but extremely valuable. We did not know where all this is leading to, but we trusted our son completely. Later it turned out that this was a watershed moment in his life, as he proved to act responsibly through increased self-discipline, patience, regular exercise and learning...

Social Inclusion or Exclusion

One of my biggest concerns were related to the potential lack of the social contacts that my son inevitably had to face with due to disease. I knew that social interactions with peers are critical for growing up. The process of understanding by peers often went in the following order: first astonishment and surprise that my son is sick, then partial understanding and surface tolerance, and lastly misunderstanding and selfishness. Peers were up to some extent tolerant to his illness, but sometimes they even thought that he pretends and exploits the situation. When he entered the high school he decided to present his disease in front of the class openly and thoroughly. This was a moment of reconciliation between him and his social environment. In order to prevent the harmful effects of the societal misunderstanding it is important to constantly raise awareness of the social environment. I will never forget the initiative of doctor Vujkovac, who was willing to come to the son's high school and explain the consequences of Fabry disease. And still, despite this excellent initiative, as well as an exhaustive two-hour interview some professors had difficulties to understand my son's physical conditions. Thus, it is not easy to find the proper degree of understanding and assistance in the local environment.

Social Contacts Are Necessary

My son has compensated the lack of social contacts successfully with regular telephone calls mainly with his friend from the early age, and older sister, who is his »information retriever« from the »outside world«. She is a wonderful friend and perfect interlocutor in the night hours. My daughter gave him the feeling that he is fully equivalent partner who should not be treated as a patient. In our family we build on the strong and frank relationships between family members which have often proven to become an important source of energy in situations when we feel all the burden of disease.

Ill Child Is Primarily a Personality Not a Sufferer

It is important to recall day by day that disease is only a small part of someone's personality and existence. The child-sufferer is primarily a personality, which is unfortunately by accident also sick.

Disease should never take the position in the focus of the family because the scope of each family member is much broader.

Karmen

7. Epilogue

If courage is stopping you – exceed your Courage.
Emily Dickinson

It took courage for all the written stories, published in this jubilee collection.

The courage to stop in this crazy fast world and to stare in the face of pain and suffer. The courage to talk frankly with the disease and to pour it on paper. The courage to move forward with our history and to share it with others – to those who may have just started facing the first signs of the disease.

Certainly there are many words that are not written on paper and in our heart which can't be expressed, but we believe, that when the moment is right, we will break off that wise silence and express the difficulty and mystery.

For this shared courage, a record of personal stories, I am grateful. I cried with them, sometimes I had to hang up the editorial pen, so I was able to bear all the pain on the page. While I finish this final thought, I feel an immeasurable wealth of the shared comfort, which we were able to give each other, so we are can make new steps into spiritual power, wisdom, and the art of living.

As in the last chapter of the Essays, Nietzsche's admired Montaigne explains, the art of life is that we can turn problems in our favour:

»We must learn how to endure what we cannot avoid. Our lives are like all the harmony of the world composite out of tune tone, gentle and sharp, the major and the minor, quiet and loud. If a musician would like only a few of them, how could he sing. He must learn, how to use all of them and knit them together. Likewise we have to with good and bad, which are made of the same material as our lives.«

Alain de Botton, The consolations of Philosophy, p. 231-232

Karmen Bizjak Merzel

Fabry Heart

Believe Us!

Anniversary Booklet Celebrating the 10th Anniversary of the Slovenian Association of Patients with Fabry Disease and the Centre for the Treatment of Fabry Disease

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3. Društvo bolnikov s Fabryjevo boleznijo Slovenije
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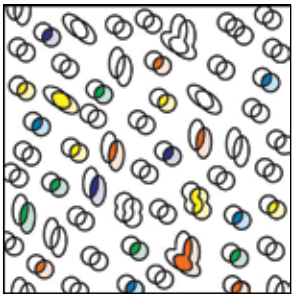

CENTRE FOR THE TREATMENT OF FABRY DISEASE

DRUŠTVO BOLNIKOV S FABRYEVO BOLEZNIJO

Dobja vas 153, 2390 Ravne na Koroškem

SLOVENIAN ASSOCIATION OF PATIENTS WITH FABRY DISEASE