No longer living in fear

This moving personal story about the challenges of living with a rare disease is written by Rocco Falchetto, a biochemist and director of analytical sciences and imaging at the Novartis Institutes for BioMedical Research (NIBR).

By Rocco Falchetto | Sep 18, 2014

Rocco suffers from erythropoietic protoporphyria (EPP), a genetic disorder that makes skin extremely sensitive to sunlight. The often almost invisible burns and the resulting severe pain are limited to the areas of skin exposed directly to light. Depending on their intensity, the symptoms can last for several days and may even require hospital treatment.
Diseases like EPP may be rare but their impact is great. There are approximately 7,000 rare diseases affecting more than 300 million people worldwide. Novartis is committed to research to deepen the understanding of rare diseases and to help patients underserved by today's medicines. In addition, research into rare diseases teaches us about the fundamental mechanisms of human biology and disease, often applicable to more prevalent disorders.

The views expressed in this story are Falchetto's and not the views of Novartis.

Sometimes the pain takes hold of the psyche and then takes over your life completely. At such moments, I have often wished that my illness could somehow be more apparent to others. Sometimes when I am treated with condescension because the pain – which feels like boiling oil on my skin – is not visible, I think about people who are blind or paralyzed. Nobody doubts a person's disability when they see them with a white stick or a wheelchair.

Rocco Falchetto can now enjoy being outside in the sunshine without worrying about his skin burning after a few moments' exposure. (Photo provided by the author)

Thoughts like this are irrational – of course I am thankful that I don't suffer from the additional problem of a visible disability. Despite this, the disease has a significant effect on my quality of life and it is hard to get away from the negative mindset – especially when doctors downplay the symptoms because they don't know what they are talking about or as a result of their lack of specialist knowledge about the illness. I always get angry when I hear similar stories from other sufferers.

While I would like to experience more understanding and sympathy for my condition, I can't stand pity at all. I have the disease, but Rocco Falchetto is not the disease – he is much more than that. While my illness has

partly shaped who I am today, I still lead a fulfilling life and have achieved things that I can be proud of. This is another reason why it is so difficult for me to talk about it. People should get to know me first without knowing anything about the illness.

However, I now know just how important it is to tell my story.

It is high time that society takes the needs of sufferers of rare illnesses and diseases into account and the difficulties we face in finding treatments.

A difficult childhood

EPP meant that my childhood and teenage years were difficult. The many associated problems and limitations were difficult to accept. The desire to be “normal” was often much greater than common sense dictated, and put me in situations that led to a lot of pain and suffering.

The situation was also difficult for my parents, especially as one of my two older brothers also suffered from EPP. They tried their hardest to support us in leading as normal a life as we could. Of course, this was not always possible. I still remember their helplessness when my brother died of liver complications at the age of 16 in 1974, something which can occur to EPP patients in rare cases. Back then, the doctors could do nothing for him. Nowadays, his life could have been saved by a liver transplant and immunosuppressants.

The thought that this might happen to me as well – together with the need for such a complicated operation – is always at the back of my mind and becomes apparent in certain situations. For example, I am always nervous as to whether all my liver values are normal after every check-up.

Sometimes they deviate from the norm, meaning additional tests are then necessary to make sure that they don’t deteriorate further. In such moments, I often think about my brother’s death and what my parents had to go through at the time – the hospital visits, the trip by ambulance to the University Children’s Hospital in Zurich and the moment that the doctors announced they could no longer help him. I was too young to really understand what was happening back then. However, I clearly remember my mother’s devastation at losing her child and the feeling of helplessness and injustice.

The long path to acceptance

After living with EPP for many years and unsuccessfully trying out various remedies and products in an attempt to alleviate the debilitating symptoms, I finally began to accept the illness.

Whenever possible, I didn’t leave the house at all on sunny days. When going to work, for example, I was always afraid of the painful symptoms of EPP. I walked in the shade as far as possible, wore long-sleeved shirts and gloves, and even covered my face in extreme cases.

However, keeping up these precautionary measures for long periods was not realistic as it would have led to almost complete social isolation. I then had to expose myself to dangerous situations from time to time, even though I knew about the possible adverse effects that may have occurred as a result.

Hopes for a normal life

The turning point for me came when I first learned about research by another company into a substance produced naturally in the body that stimulates tanning of the skin. I discovered it online completely by accident at a time when it was still being tested in other applications. I saw its potential in treating EPP and talked to my

doctor. Within a matter of months, she and the researchers concerned had organized the first human clinical trial for it.

I was one of five patients in Switzerland who took part in the clinical study.

I will never forget the day when I first received the drug– I summoned all my courage and went to sit in the sun. I then waited, afraid of what was going to happen. Ten minutes passed, then 20, then 30 (where I would normally already be in pain), then 40 minutes and more in the sun – and all without the typical painful symptoms! After over 40 years with the illness, I was convinced that I had finally found a drug that I believed tackled the EPP symptoms.

I took part in a further clinical trial. I had to travel to Zurich every two months for a new injection of the drug implant, which was a small price to pay given what I felt was its positive effect on my quality of life.

I am currently benefitting from a compassionate use program which allows me to continue using the drug and, thanks to special legislation, most Swiss health insurance companies now reimburse the cost of such medication.

Difficult regulatory environment

However, the current situation is only temporary – if the product is not formally approved, then the reimbursement and the treatment can be withdrawn. This would be a heavy blow.

I don’t like to think about what would happen in this case, but the increasingly challenging context surrounding drug approvals does make me nervous as a patient.

Some countries have even started to put a price on the quality of life. For example, at the end of 2010 the Swiss Federal Court passed a judgment ruling that health insurance companies were not obliged to reimburse the costs for treating Pompe disease, a rare, serious metabolic disorder. Treatment of Pompe disease can cost several hundred thousand Swiss francs a year. While this is undoubtedly expensive, the therapy can significantly ease the symptoms and allow patients to lead a relatively normal life.

This shows how difficult it still is for people with rare diseases to make their voices heard, not to mention how inadequately their needs continue to be met. I am also worried that our modern society is not more committed to discovering creative and innovative solutions for the benefit of these patients.

This medication has radically changed my day-to-day life I can now do things that I would never have been able to do before without pain. Not only my friends and family benefit from this – my working life has also become much easier.

I can now go to the office without having to always move about in the shade and go on business trips without thinking so much about the unfamiliar surroundings. Lastly, I can now move freely around at work, meet with colleagues and simply enjoy our wonderful working environment.

The EPP sufferer community is a small one in Switzerland, gathered in a patient organization that I chair. We all know one another. It is very moving to hear how patients who have undergone treatment with the drug felt the sun warm their faces for the very first time without causing pain and suffering.

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