

D313Y - patients, symptoms and therapy experiences



1. Female, first symptoms at the age of 4, severe leg pain so that she did not want to walk, frequent abdominal pain in elementary school, circulatory problems with fainting. She tried to overcome burning pain in her feet and hands with sports.
Trained as a nurse. Severe cardiac arrhythmias, pacemaker at 19 years. Countless visits to the doctor and hospital stays. Incapacitated and wheelchair at the age of 29, diagnosed at 30. After 4 years of chaperone therapy without a wheelchair. Volunteer patient supporter as a slow entry into the profession.
2. She is the mother of a son. At the age of 3, he often complained of abdominal pain, burning pain in his feet from the age of 4, therapy should start soon.
3. Boy, 10 years old, symptoms since birth. Constantly diarrhea. In the first 2 years of life, constant visits to the doctor and clinic. From the age of 3, stabbing and burning pain in the feet. Parents describe countless difficult nights. The boy often vomited in the morning. The father took a year's leave of absence due to the heavy burden. All therapeutic measures showed no or only very short-term success. Diagnosis at 4 years of age, immediate onset of ERT. Very rapid improvement of all symptoms, symptom-free after a few months of ERT.
After 4 years of therapy, sudden recurrence of all symptoms. Doctors were not aware of any habituation effect and no one had an explanation for it. The boy had been treated at home for a long time. With the change of home therapy nurse, all the symptoms subsided. It was only after a lengthy investigation by the parents that it turned out that the nurse who administered the ERT had a hitherto unknown alcohol problem. He had probably lost his apartment and slept in his car. Since it was summer, the drug was not refrigerated, became very hot during the day and probably lost its effect. So the home therapy company determined this, he was fired. With the new nurse and the medication now cooled again, the symptoms also subsided again.
For parents and doctors, this was the best proof that their son was getting the right therapy. There can be no talk of a placebo effect.
4. Female, 42 years. First symptoms in early childhood. Always sick, weak constitution, constantly wandering pains throughout the body. Digestive problems, severe heat intolerance and headaches were part of everyday life. Despite the conspicuously sick appearance, the symptoms were always dismissed as psychosomatic. She became very limited in her movements and felt that her body was hardening.
Speech and word-finding disorders were added. The patient lost many of her cognitive abilities and became almost completely bedridden. She was fully cared for by her husband and a caregiver. Self-sufficiency was no longer possible. To outsiders, the patient was demented.
A pain therapist had her tested for Fabry disease. She immediately started chaperone therapy. Many symptoms improved over time. After almost 5 years of therapy, she is more and more often able to actively participate in life. However, it needs concomitant pain therapy.

The patient also regained a lot cognitively and is once again able to read in a meaningful way. I visited the patient in 2019 and was able to see her dramatic state of health. For outsiders, the improvement is clearly visible.

- There is another patient in this dramatic condition in Switzerland, who unfortunately is not being treated. She is also cared for by her husband. It should be treated and observed over the next few years. Here, doctors and scientists would have a great opportunity to observe developments themselves and to find out how such dramatic conditions can occur. We would be happy to put you in touch.
5. Male, mid-40s, diagnosed with hypertrophic obstructive cardiomyopathy in 2010, and in 2013 he had an unsuccessful Transcatheter Ablation of Septal Hypertrophy at the Heart Center. 2017 Climbing stairs only possible with breaks, daily walks in the forest. Circulatory problems, cardiac arrest. Two more unsuccessful attempts at Transcatheter Ablation of Septal Hypertrophy.
Septum thickness in the heart 20mm. Heart biopsy, result: 3 monogenic cardiac variants that supposedly do not make you sick and Fabry disease D313Y. The only treatment option for Fabry disease. Start with Chaperone. Over time, the general condition improved significantly. Climbing stairs and even cycling was much easier. 2018 Atrial fibrillation, an ICD was implanted. The thickness of the septum has decreased to 15 mm.
 6. The daughter of this patient complained of migrating joint pain from the age of 10. Once in the knee, then in the hip and the next day in the shoulder. Initially, the pain was in the large joints, later also in the fingers. At the age of 16, she had more and more problems with walking because the pain was too great. Climbing stairs only in extreme pain. At the age of 17, her condition became threatening, she fainted more frequently and convulsed, her fingers turned deep blue or became translucent white. She couldn't write her exam with a pen. A two-week hospital stay to search for a diagnosis resulted in fibromyalgia.
Shortly thereafter, therapy started because of the diagnosis with the father. After several years of therapy, the patient is doing very well again. She studies and lives on the 2nd floor and is happy to be able to jump up the stairs every day.