

My name is Stephanie Schulz and I live with my two children in the south of Bonn. I met my husband Jörg in 1997 when we were both in our late twenties. He casually told me at some point that he was a "stone carrier", which would mean that he has been forming kidney stones since birth. He also told me that his parents' first son died of it as a baby and that he himself had been in hospital as an emergency since childhood, which always drove his parents into despair, as they were afraid of losing him too. Nobody in the family knew a reason for the illness. My mother-in-law later always told me only that Jürgen, the first child, had died "of kidneys". After his death the parents had received a medical report which they never understood. The whole family was completely helpless in the face of the children's illness. Again and again his parents had to take the unconscious toddler to the hospital and each time it was a matter of life and death. At least that is what everyone in the family told me.

When my husband grew older and it was clear that he could withstand the colic better than his deceased brother, his frightened parents did everything to keep possible dangers away from him. They rarely went on holiday with him and he was not allowed to learn how to swim or ride a bike. Later he made up for all this. He never missed an adventure, was literally looking for dangers and was obsessed with travelling the whole world.

When I met my husband, he had deep scars on his back, where they had operated out the stones in emergency surgery, because breaking them up was not possible at that time. Once this happened when he was a student in Australia. He barely survived because he got colic while he was in the outback and had to be flown to the clinic. He told this story often, I think it was a sign for him that he was stronger than his illness.

In 1998 I experienced him with a colic for the first time. The attending doctor in the emergency room said to me at that time that these were the only pains that could be compared with pain in labour, I remember that exactly. My husband was being treated by a urologist in Cologne, where we lived. After each colic and the subsequent breaking of the stone that had caused the pain, my husband took the stones with him and added them to his extensive collection. None of the doctors, neither in the hospital nor the family doctor or urologist ever talked about primary hyperoxaluria or analyzed the stones. They told my husband to drink a lot. He also took Allopurinol (which in retrospect was completely ineffective). He was obsessed with drinking, got up several times during the night to drink water and encouraged those around him to drink a lot. He had a panicky fear of not drinking enough. When I was pregnant and the gynecologist asked us about hereditary diseases, my husband said nothing. I then told the doctor that there was a kidney disease in my husband's family from which a baby had also died. My husband resented that. He always felt this disease as a flaw, something that could not be controlled and which he preferred not to mention. He never woke me up when he had colic at night, but sat in the living room until the ambulance came. He had no emotional access to his illness.

When we moved to Bonn after the birth of our second child, my husband changed the urologist who referred him to Professor Hoppe. He immediately suspected primary hyperoxaluria and let my husband participate in a study for two weeks in the University hospital. Afterwards it was clear that he was a PH1 patient. At that time my husband was 39 years old. He hardly talked about the diagnosis, neither with me nor with his friends.

In 2009 we flew as members of the support group for primary hyperoxaluria, PH1 Bonn, to a symposium in New York. There I met American families who had sick children. They were incredibly happy to see that it was possible to live with the disease as an adult; there were many emotional reactions that shocked me at that time because I realized that my husband had never really talked about the extent of his disease. In the following years Jörg participated in various studies at the University Hospital under the direction of Professor Hoppe; he had great hope that a drug would be found that would spare him the transplantation of which he was very afraid. However, as the development process seemed to be too long, he increasingly lost hope. At the same time, his creatinine value deteriorated and Professor Hoppe often spoke about dialysis. My husband, who did not want to admit it, concentrated even more on his career, we made long journeys more often and he did not talk about his fears nor about his deteriorating values

In the winter of 2015 I noticed a change in his behaviour for the first time, which intensified in the following months. He often fell while skiing, was unfocused and slowed down in his movements. I advised him to go to the doctor and have a checkup on what he was putting off.

On a trip to South Africa in March 2016 he got a sinusitis, from which he did not really recover, and he collapsed from the antibiotics he was prescribed. The ENT doctor diagnosed an allergy to the drug and prescribed another one. Since he just changed his job and had a promising job offer, my husband stubbornly refused to have his creatinine levels checked. Later, I learned that it was this infection that had given the kidneys "the rest", so to speak.

In July 2016 we went to France for three weeks. On the second day of the holiday, my husband could not eat any more, was fidgety and had pain in his legs. He refused to see a doctor in France and suspected he might have diabetes. In the evening he had muscle cramps and seemed increasingly disoriented. He asked to see his family doctor. I packed everything and drove us home. The family doctor immediately diagnosed kidney failure; my husband was admitted to the University hospital as an emergency, where he was given emergency dialysis in the admissions department. He had a creatinine value of 21 mg/dl at that time, blood urea was about 500 mg/dl and potassium 8 mmol/l. All life-threatening values. At the University hospital, doctors from various departments gathered at his bedside to see who had survived these values. After weeks of slow dialysis the values returned to stable elevated and he was discharged home with regular hemodialysis.

For us as a family, this summer has changed our whole life from the ground up. My husband could no longer work, could no longer pursue his career plans, could no longer travel. From then on, he struggled to get a transplant as soon as possible. He was against KALT (Kidney After Liver Transplantation) surgery from the beginning, preferred to have both transplants at the same time, but the doctors did not agree, because the remaining high concentration of oxalate in the body would have destroyed the new kidney immediately. The wait for the surgery was exhausting and difficult, my husband fought for every point on the list. At the same time, Professor Hoppe advised him to rather wait, as there might soon be a drug that would make the risky surgery unnecessary. But my husband no longer believed in a drug and pushed ahead with the operation. His condition worsened as more and more oxalate was deposited in his body, which the dialysis could not filter. It was a race against time. After 17 months, on Christmas 2017, my husband was given a new liver. Everyone was very euphoric, the team was relaxed and in a good mood, the surgery went very well and the liver started working. Jörg stayed in the clinic for 6 weeks and was then discharged into the rehabilitation. From the very beginning he struggled with fluid accumulations in his body due to the lack of kidney function, which could never be completely drained. Among other things, this led to effusions in the lungs and often threw him back. In the rehabilitation he got worse and worse, and back home he got repeated infections which were difficult to treat due to the immunosuppression. During the whole time, however, the attending doctors at the University hospital were confident that he would make it. In May 2018, my husband was so bad that he had to go to the clinic repeatedly. Since there were no kidneys to manage the fluid balance in the body, he suffered from severe ascites (fluid in the abdomen) and infections. However, the nephrologists informed him that a kidney transplant was out of the question due to his poor condition. My husband was a great fighter, but I believe that in that moment he gave up.

At the end of June, I took him to the emergency room, the bilirubin had risen sharply, the bile was not working properly, which also indicated that the liver was deteriorating, but the doctors at the clinic were still saying that the liver was doing very well. My husband fell into a coma and was taken to the intensive care unit, he woke up again shortly after Germany had to leave the Soccer World Cup. I remember that he asked me about it and said that he hadn't missed anything then. He had a very good sense of humour.

At the beginning of July he fell into a coma again with sepsis, shortly before he called me. That was the last time I heard his voice. For five weeks the doctors fought for his life and since that time in the intensive care unit I know that this is not just a phrase. In the end, even the most optimistic doctors had to admit that the liver was no longer that the liver was no longer working.

All that time there was no doctor I knew except Professor Hoppe. I did not see any of the doctors who had declared at the beginning that my husband would survive this operation without any problems and be back home in top shape after two months, in

the intensive care unit. When my husband was dying, a receptionist gave me 60 seconds to talk to the head doctor.

The changing doctors in the intensive care unit and especially the nurses, to whom I am still grateful today, really fought hard. On July 30th, exactly two years after the kidney failure, my husband opened his eyes again after five weeks of coma. One day later the values collapsed. On the morning of the second of August, the doctors stopped the life-support measures. At noon my husband died. He turned 49 years old, our children were 13 and 14 years old at that time.

Looking back, I think that the disease was diagnosed too late. If this had happened earlier, the path would have been different and Jörg might still be alive. I also think that we should have been better informed, so I know today that kidney failure can be recognized by clear signs, such as the typical smell of ammonia or confusion because of the high urea level.

Furthermore, I think that once diagnosed, it is essential that the patient has regular and close monitoring, especially if creatinine levels deteriorate. I find it cynical and calculating that patients are only put on the transplant list when they are already sick and on dialysis.

I am convinced that the clinics need psychologically trained personnel who act as a link between the doctors, who, in order to protect themselves, can hardly show empathy in such difficult situations, and the patients and relatives, who do not receive the support and comfort that is so incredibly necessary in the technical operation of high-performance medicine. In my opinion, the psychological damage that is caused here is hardly reparable and also detrimental to the possible recovery of the patients.

I hope that the medicine that my husband would have needed so urgently will soon be available to every patient, and that no one suffering from this disease will need to be dialyzed or transplanted any more.