

## Case report Dr. Ulrike Hoffmanns, Germany

„My case“ first manifested in 1973 at age 19 years with recurrent kidney stones, which had to be surgically removed from my kidneys. Analysis of the stones revealed calcium-oxalate as the pure component. At this time my serum creatinine level was approximately 2.0 mg/dl, which translates to a kidney function below 50%. No physician made any specific comment neither regarding the necessity of further diagnostic evaluations nor with respect to my reduced kidney function.

Some 7 years later I experienced end stage renal failure and since I was treated by a big nephrology outpatient clinic of a University hospital in Germany. At this time the working hypothesis of my underlying renal disease was “chronic pyelonephritis”, means loss of kidney function due to recurrent urinary tract infections. Further symptoms appeared (see below), but this did not lead to additional evaluations. The diagnostic examinations making the diagnosis of “Oxalosis” at least an option were either neglected or misinterpreted.

Up until my first isolated kidney transplantation in 1981 I received chronic hemodialysis treatment. Luckily enough I received the kidney graft early after initiation of end stage renal failure, but nevertheless, the kidney graft function deteriorated soon and in 1989 I needed dialysis treatment again. Fortunately, this time on dialysis was even shorter and I again received an isolated kidney graft. Due to hampered early kidney function three biopsies were performed in which typical birefringent calcium-oxalate crystals were found. A severely hyper-echoic kidney graft was seen ultrasonographically, and, in addition, small calcium depositions were visible all over the kidney parenchyma. In 1997 I experienced bone and joint pain and in September 1998 kidney graft function rapidly deteriorated, which made chronic peritoneal dialysis necessary. The working diagnosis was still not revised.

Early 1999 I experienced a loss of vision and the eye doctor described retinal calcium-oxalate depositions. Next to the pathologist, who had examined the kidney graft biopsies, he was the first physician to suspect “primary hyperoxaluria” to be the underlying disease instead of chronic pyelonephritis. At the same time the rheumatologist diagnosed a crystal arthropathy and next to that calcium-oxalate depositions were also found in skin and other tissue. In the year 2000 a retinal ablation occurred. Due to the increased bone and joint pain a bone biopsy was performed, which revealed oxalate osteopathy. The pathologist openly discussed primary or secondary hyperoxaluria as the underlying disease and the current attending physician commented that “we have to re-evaluate the findings”.

Now, another University Hospital performed a liver biopsy for diagnostic evaluation. Result: decreased activity of the liver specific enzyme AGT, which led to the diagnosis of primary hyperoxaluria type I. Finally, I successfully received a combined liver/kidney transplant in November 2002.

Currently I am mostly blind, I can only read with heavy glasses, I cannot drive a car and I cannot work any longer (I am a physician myself) as I have ongoing bone and joint pain. Due to the calcium-oxalate crystals in the skin I also have increasing neurotropic pain. Both liver and kidney function are stable, but ongoing hyperoxaluria in the range of  $> 1.5 \text{ mmol/24 h}$  ( $> 135 \text{ mg/24 h}$ ) already led to a decline in kidney function.

Nevertheless, I did not yet bury my hope that I will gain a better quality of life, possibly with newer treatment options to come.