Patient report of family Pfeifer with Eva (20 months old), PH type II

Eva was a very restless baby from birth. She slept little and cried a lot, often stretching herself backwards. There were also situations where she screamed until she vomited. Only later did we realise that this might have been related to her illness. But perhaps this happened completely independently.

The first symptoms visible to us appeared at about 5 months of age, when we found a stone-like structure about 4 mm in size in her diaper while she was changing. At first we thought it was dirt and disposed of it with the diaper. A few weeks later we found another almost identical stone in the diaper. We took it to the pediatrician for further clarification. The paediatrician told us that it could be a kidney stone, but he thought it was impossible that our child, who was only a few months old, could have got rid of such a large stone without any problems. So we were sent home without further diagnosis.

In fact, Eva's behaviour was not conspicuous in the run-up to the stone findings in her diaper. When after hardly a fortnight the next stone was found in her diaper, we took both stones to the pediatrician again. There we explained that we wanted to find out whether they were kidney stones, since father and grandfather had also developed kidney stones in the past. The stones and a urine sample were then sent to the laboratory for diagnosis. As it turned out, they were calcium oxalate stones. Eva and I were then referred to the kidney outpatient clinic at the Children's Hospital in Heidelberg for further clarification.

Again, several weeks passed before the appointment in Heidelberg. There a blood and urine examination was arranged. The results directly indicated primary hyperoxaluria. A further urine sample sent to Bonn for examination led the attending physician to suspect PH type II due to the biochemical constellation. A subsequent genetic examination confirmed the assumption. So about three months after the first diagnosis by the pediatrician we had a definite diagnosis.

The medication was initially administered with a combination of uralyte and magnesium to inhibit stone formation. In addition, we tried to increase Eva's drinking volume as far as possible in a nine-month-old child. At the same time, we presented her at the age of twelve months to Prof. Hoppe at the University Hospital in Bonn. There, for the first time after the initial examination by the paediatrician, an ultrasound of the kidneys was carried out, showing another stone about 4 mm in size in the left kidney. In the past months, Eva had occasionally deposited larger and smaller stones, which we found in her diaper. The stone detected by the ultrasound in Bonn was removed the very next day without complications, as far as we can judge.

After consultation with Prof. Hoppe, the medication was changed from Uralyt to modified Shol's solution. In addition, Eva continues to receive magnesium. The amount to be drunk has been steadily increased through constant offering, so that Eva now drinks 1 - 1.5 litres of water daily (with a current body weight of 10 kg and a height of 80 cm). She wakes up several times during the night and then drinks around 300 - 400 ml spread over the night.

After switching to the modified Shol's solution, we could not detect any more large stone leaks. Also in the following months, the ultrasound of the kidneys showed no more stones. The recently performed heart and eye examination was inconspicuous.

The daily repeated administration of Shol's solution and magnesium has since become routine for us. Eva's development has so far been completely normal and - as far as we can judge - she has no further limitations due to her illness.

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