A Case of Cystine Calculus in a two-year-old Infant

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ABSTRACT. A two-year-old boy developed abdominal pain and vomiting at initial onset. After referral to our hospital, diagnostic images disclosed a large urinary calculus in the right pelvis involving hydronephrosis. From the results of urinalysis, a cystine calculus caused by cystinurina was suspected. At first, we considered the possibility of destroying it with extracorporeal shock wave lithotripsy (ESWL) or removing it with endoscopic instruments, but instead selected pyelolithotomy because of the calcular characteristics and the patient's renal dysfunction. The choice of treatment for urinary calculus in a child may depend on the calcular size or component and on the patient's status. The availability of ESWL for the treatment of urinary calculi in pediatric patients has been recognized, but there should be no hesitation in selecting pyelolithotomy in cases presenting with difficulty or complications.

Key words ① **pediatric urolithiasis** ② **Cystinurea** ③ **Cystine calculus** ④ **ESWL (Extracorporeal shock wave lithotripsy)** ⑤ **Pyelolithotomy**

Urinary calculi are often encountered in adults, but are rare in the infants. Such pediatric urolithiasis sometimes results from a metabolic disease conforming to the laws of heredity such as cystinuria. The treatment of pediatric urolithiasis has recently become less invasive with procedures such as extracorporeal shock wave lithotripsy (ESWL), but the adaptation of ESWL and its effect vary with the patient and the stone. We experienced a pediatric case of cystine calculus with abdominal pain and vomiting at initial onset and report it in addition to discussion of its treatment.

CASE REPORT

A two-year-old boy had been well until he developed abdominal pain in the right lower quadrant and vomiting. The family history revealed that two relatives on his father's side had suffered from urinary stones. Because of a high fever, he visited a local hospital. Right hydronephrosis was detected by ultrasonography, and he was referred to our hospital.

In the US scan, a stone with low grade acoustic shadow was discovered in the right dilated pelvis (Fig. 1). Abdominal X-p demonstrated an oval mass shadow in the right upper quadrant (Fig. 2). A CT scan also clearly revealed an oval stone located in the right pelvis, 30 mm in major axis (Fig. 3). This CT scan also revealed a dilated and thickened pelvis and upper ureter. The lower ureter was not dilated.

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Fig. 1. Ultrasonography demonstrating a large calculus in the right pelvis involving hydronephrosis.

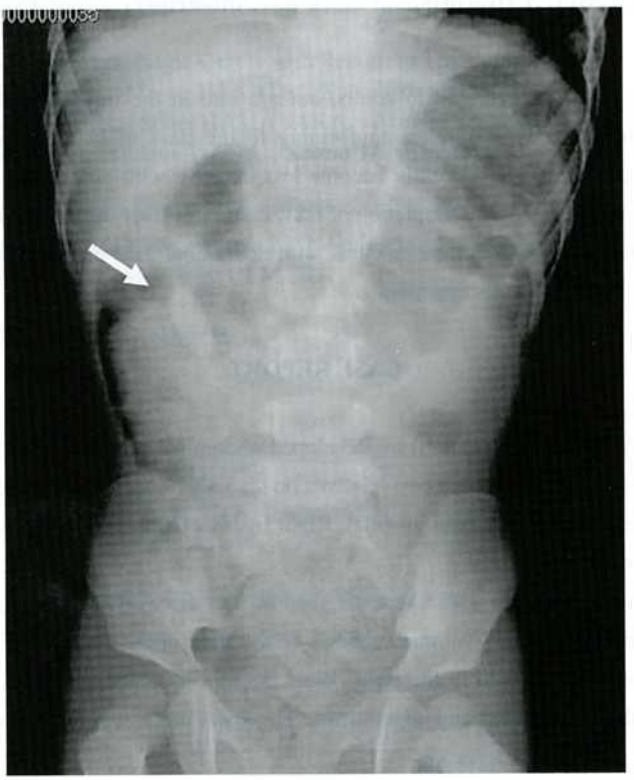

Fig. 2. Oval mass shadow shown in an abdominal X-p.

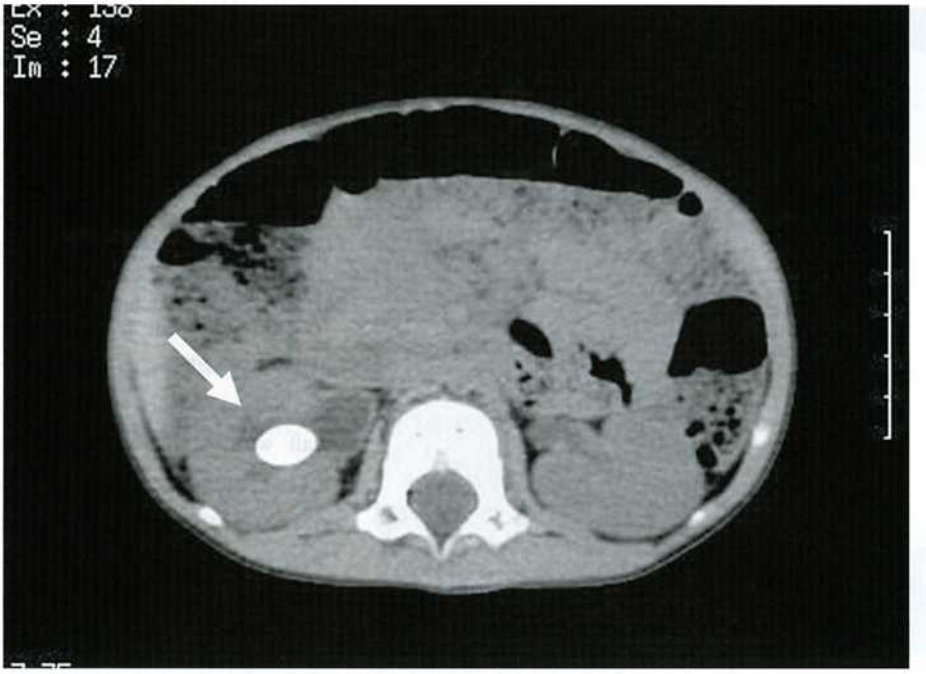

Fig. 3. CT scan demonstrating an oval calculus clearly in the thickened pelvis.

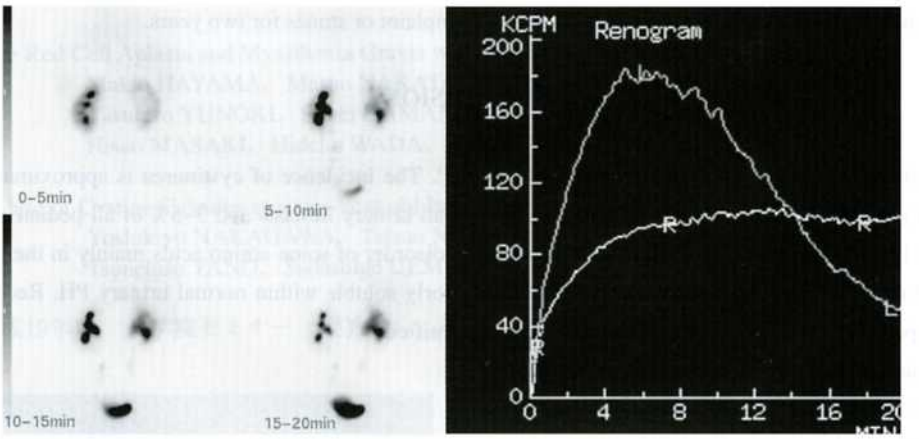

Fig. 4. A renogram showing a failed accumulation to the right kidney and an incompletely obstructive pattern.

A radioisotope renogram was made to evaluate his renal function. It revealed poor accumulation and a discharge delay of the isotope in the right kidney, indicating poor renal function and incomplete obstruction (Fig. 4). In retrograde ureterography using a cystoscope, a stone in the dilated pelvis was detected. A stricture in the lower part of the ureter was suspected, although a good passage of the contrast medium was observed (Fig. 5).

Urinary pH was 6.5 and occult blood was 1+ and no bacteria were detected in the urine. The calcium concentration in the urine was proven to be within normal range. A diagnosis of calcium-based stones, most

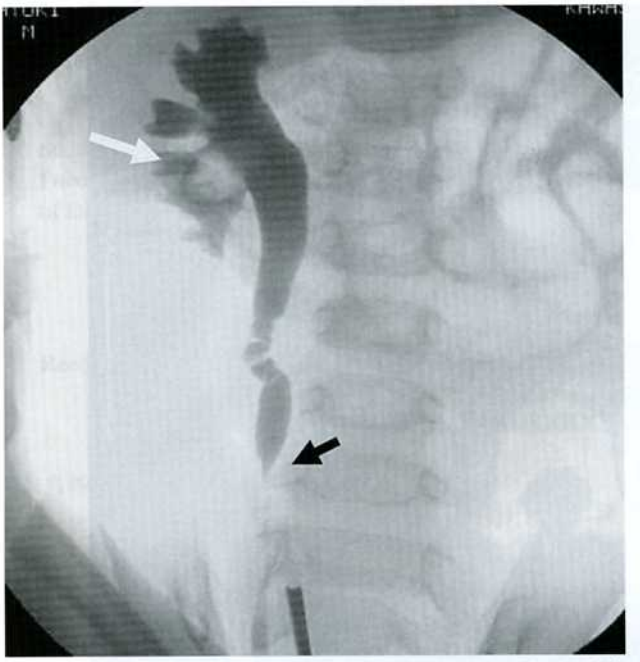

Fig. 5. Retrograde ureterography showing the stone in the dilated pelvis (white arrow) and suspicious stricture in the lower part of the ureter (black arrow).

commonly seen in childhood, was ruled out. Repeated urinalysis revealed a cystine crystal. Based on this result, we measured the total amount of cystine per a day, which was 725.2 μmol (normal range 20~200 μmol). He was diagnosed to be suffering from cystinurina which caused the cystine calculus.

He underwent pyelolithotomy under general anesthesia. The right pelvis and the ureter presented a hard and thickened wall with fibrous adhesion around the tissue. The stone, 24 × 11 × 8 mm in size, was very hard, yellowish and waxy (Fig. 6). In analysis of the component, the stone was found to be made of almost pure cystine. The postoperative course was uneventful. It was suggested that he drink enough water and sodium bicarbonate was prescribed for urinary alkalization to prevent recurrent stones. At present, he has been free of complaint or stones for two years.

DISCUSSION

Cystinurea causing cystine calculi is a rare disease[1]. The incidence of cystinurea is approximately 1 in 16,000 in Japan[2]. Cystine calculus comprises 1~2% of all urinary lithiasis and 3~5% of all pediatric urinary stones. This clinical condition results from a resorption disorder of some amino acids mainly in the proximal urinary tubule. Among these amino acids, cystine is poorly soluble within normal urinary PH. Recently the gene responsible for this hereditary disease has been identified[3), 4)].

Management of pediatric urolithiasis has changed[5), 6)] and the possibility of destroying the stone with ESWL (extracorporeal shock wave lithotripsy) is initially considered. ESWL has been reported to be useful and has become prevalent. Application of ESWL for children, first reported by Newman et al in 1986[7)], also has been proven to be suitable and highly effective in about 60~90 % of cases[8), 9)]. However, the indication for ESWL differs with each patient. It is generally contraindicated in patients with congenital urinary tract deformity, obstruction in the lower urinary tract and renal

Fig. 6. The hard stone was removed.

failure. Calcium-based stones, which occur frequently in children[10], are comparatively easy to break up with ESWL, whereas cystine calculi have been reported to be hard to destroy with ESWL[8), 11].

In our case, the application of ESWL was rejected. Repeated use of ESWL would be needed for such a large sized stone, and renal dysfunction due to the stone which was identified with a renogram, already existed and could deteriorate with the use of ESWL [12]. The contrast study also led us to suspect stricture in the lower part of the ureter. We also considered percutaneous nephrolithotomy (PNL) [13], but PNL for this patient with a large cystine calculus was assumed to be a complex and invasive procedure, and his parents did not agree to it being undertaken. Therefore we performed an open pyelolithotomy. Though the operation is more invasive than the other methods, the advantage of this procedure is that the stone can be removed quickly and preventive therapy can be started immediately.

The choice of treatment for a urinary calculus depends upon its size or component and/or the patient status. Although ESWL or PNL are available for pediatric urolithiasis, one should not hesitate to perform an open pyelolithotomy if these procedures were predicted to be difficult or complications could possibly be involved.

Due to the high incidence of recurrent stones, long term follow-up is mandatory. As this disease conforms to the law of complete or incomplete recessive heredity, inspection of the patient's family history; i. e., his parents or siblings, should be carried out.

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