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Case Report

A Rare Case of Chronic Kidney Disease with Photophobia

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Abstract

Cystinosis is a rare autosomal recessive disease characterized by cystine accumulation in the lysosome leading to various organ-dysfunction. Kidneys are severely affected, of which nephropathic infantile form is the most common. Juvenile nephropathic cystinosis has a slower progression to end stage renal disease. Rarely, cystine crystals in the cornea may be fewer and diagnosed later in later in life. We report a case of 8 year old female child who was diagnosed to have juvenile nephropathic cystinosis with corneal deposits detected later in life.

Keywords: Chronic kidney disease; Corneal deposits; CTNS gene; Juvenile cystinosis

Introduction

Cystinosis is an autosomal recessivedisorder characterized by the accumulation of cystine in lysosomes resulting in various clinical manifestations, such as Fanconi syndrome, End-Stage Renal Disease (ESRD), hypothyroidism, hypogonadism, insulin-dependent diabetes mellitus, muscle weakness, central nervous system complications, keratopathy, and pigmentary retinopathy [1,2]. Cystinosis is classified into infantile, juvenile and adult cystinosis according to the age at onset and severity. Nephropathic infantile cystinosis is the most frequent form and may progress to ESRD in the first decade of life [3]. We report a rare case of juvenile cystinosis.

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Case Report

An 8 Year old female childborn to consanguineous marriage was brought with complaints of breathlessnessand decreased urine output of 1 week duration, associatedwith abdominal distension and intense photophobia.Shewasdiagnosed to have proximal RTA-fanconi syndrome, rickets and hypothyroidism at 5 years of age for which she was on irregulartreatment. There was no significant antenatal, natal and post natal history. On examination patient was drowsy, pale and had cloudy cornea. Laboratory investigation revealed hemoglobin- 6.4g/dl, WBC count-10200/mm3, platelet -1.60 Lakhs/mm3, blood Urea -364 mg/dl, serum creatinine-13.3mg/dl, serum sodium-135meq/l and serum potassium - 2.7 meq/l.urinalys is revealed proteinuria (spot urinary protein creatinine ratio of 0.9), glycosuria 2+ without any deposits. Her serum calcium level was 9.8mg/dl, Phosphorus-1.1 mg/dl, intact Parathormone level -76.20 pg/ml and 25 hydroxy Vitamin D level -57.18 ng/ml. ABG analysis revealed normal anion gap metabolic acidosis. Ultrasonography showed contracted kidneys with loss of corticomedullary differentiation.

Ophthalmological examination including a slit lamp examination revealed corneal deposits.

In view of Renal Failure, Hypophosphateamic rickets, Hypothyroidism, proximal renal tubular acidosisand corneal deposits, cystinosis was suspected and genetic study was done. A homozygous missense variation in exon 7 of the CTNS gene (chr17:g.3558607C>T) that results in the amino acid substitution of Phenylalanine for Serine at codon 141was detected. Patient was diagnosed to have Juvenile nephropathic and ocular cystinosis. Patient was treated with 52 cycles of acute peritoneal dialysis. As the kidney function did not improve, patient was diagnosed to have end stage renal disease and was initiated on CAPD (Continuous Ambulatory Peritoneal Dialysis)

Discussion

The nephropathic juvenile Cystinosis accounts for only 5% of all patients. They show much slower progression to ESRD than infantile form [3]. Servais et al. reported 14 patients with the late-onset nephropathic form in which Corneal deposits were identified later in life after diagnosis in four patients [4]. In our patient corneal deposits were identified 3 years after diagnosing Fanconi syndrome.

In cystinosis, cystine accumulates inside the lysosomes due to a defect in the gene that encodes cystinosin, the protein that transports cystine across the lysosomal membrane. In kidney, cystine accumulation increases apoptosis of the cystine-laden renal proximal tubular cell, which causes tubular dysfunction [5]. The gene for cystinosis has been mapped to chromosome 17p13. The gene CTNS consists of 12 exons and encodes for a 367 amino acid lysosomal membrane protein, named cystinosin. More than 140 mutations in the first 10 exons and in the promotor of the gene have been described in patients with cystinosis [6]. In our patient geneticstudy revealeda missense mutation ser 141phe. Previous literatures show that ser141phemutations has been reported in only 5 patients in India.

Diagnosis can be confirmed either by, elevated cystine content of peripheral blood leukocyte or fibroblasts, demonstration of cystine corneal crystals by the slit lamp examination orconfirmation of mutation of the CTNS gene. Treatment of cystinosis consists of

supportive therapy, cysteamine administration and renal transplantation for those who progress to end-stage renal disease [7]. Adequate fluids to prevent dehydration, correction of hyponatremia and hypokalemia is important.Plasma bicarbonate concentration andplasma phosphatelevelshould be maintained between 21 and 24 mEq/L andabove 3.7 mg/dL respectively.To prevent rickets, calcium, magnesium, and vitamin D mustbe supplemented. Vitamin D can be given at a starting dose of 0.25 mcg/day of calcitriol andshould be adjusted according to the plasma calcium concentration [8].

Cysteamine therapy should be started as soon as the diagnosis of cystinosis is confirmed as it preserves renal function, prevents hypothyroidism, and improves growth in affected children. Immediate-release preparation of cysteamine bitartrate is the most commonly used formulation. The dose should be progressively increased from 10 to 50 mg/kg per day (maximumof 1.95 gm/m^2 per day), given in four divided doses. Levels of cystine are measured in white blood cells once the maintenance dose is reached, then monthly for three months, quarterly for one year, and then twice a year. The optimal target level is less than 1 nmol half-cystine/mg protein. Blood sampling should be obtained six hours after taking a dose of cysteamine [9]. Only Topical cysteamineis effective in preventing corneal crystal deposition. Renal transplantation is successful in patients with ESRD with excellent long-term renal outcome. Cystine-induced tubular dysfunction does not recur on the graft, although cystine does accumulate in the interstitial cells. After renal transplantation, cysteamine treatment should be given as soon as possible [10].

Conclusion

An early diagnosis of cystinosis is very important. Slit lamp examination of eye inpatient presenting with proximal RTA, hypothyroidism and photophobia should be done to aid early diagnosis but cystine crystals may not be present in some cases of juvenile nephronophthisis. .Early initiation of cysteamine therapy can delay the progression to end stage renal failure .Renal transplant recipients with cystinosis have a good long term outcome but can have non-renal complications. Cysteamine therapy must be continued to prevent or delay non-renal complications.

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