The clue in my eyes

Dr. T.Subhashree 1st year Dch

Prof. Dr.Lakshmi unit

ICH and HC

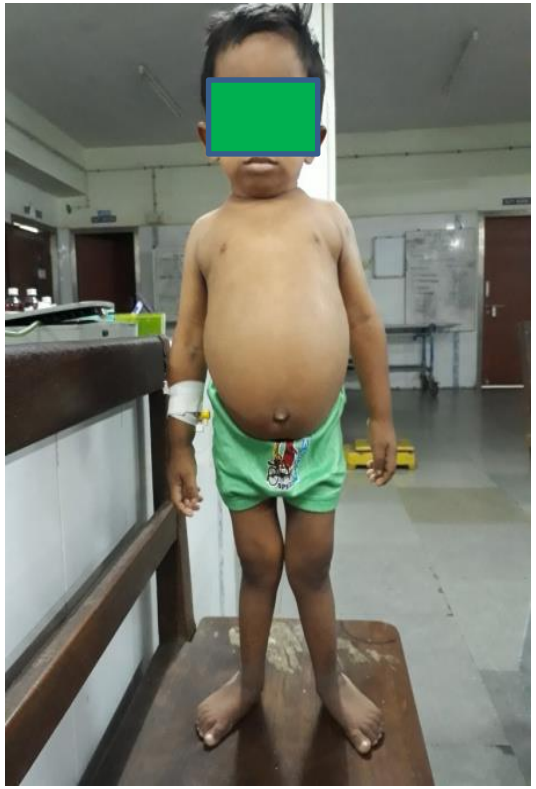
- 7 years old male child 2nd born to 2nd degree consanguineous parents
- C/o not attaining age appropriate height compared to his peers
- Referred as MPS
- H/o polyuria, polydipsia – early childhood
- No h/o chronic diarrhoea, greasy stools, headache, vomiting or visual problems
- Antenatal /postnatal - uneventful
- Nil significant family history
- Mild motor developmental delay +, social and language milestones were normal

- On examination child had short stature

- Height-95cm which is <3rd percentile; ht age – 1 year
- Weight-9.6 kg which is <3rd percentile;
- BMI-10.6 which is <5th percentile
- SMR-stage 1(prepubertal)

General examination :

- pallor+
- Bilateral corneal haziness +
- f/o rickets - frontal bossing, widening of wrists ,b/l genu valgum
- Exaggerated lumbar lordosis
- abdominal protuberance
- vitals -stable , BP – 90/60
- System examination - normal



Problems

- *Short stature*
- *Polyuria, polydipsia*
- *Rickets*

Provisional diagnosis

RICKETS - RENAL

- Anemia -Hb -8gm/dl
- Renal parameters : Urea-69 , Creatinine-2.3
- **Hyponatremia** - sodium-117 ; **Hypokalemia** - Potassium-1.9
- **Serum bicarbonate – 14 meq/L**

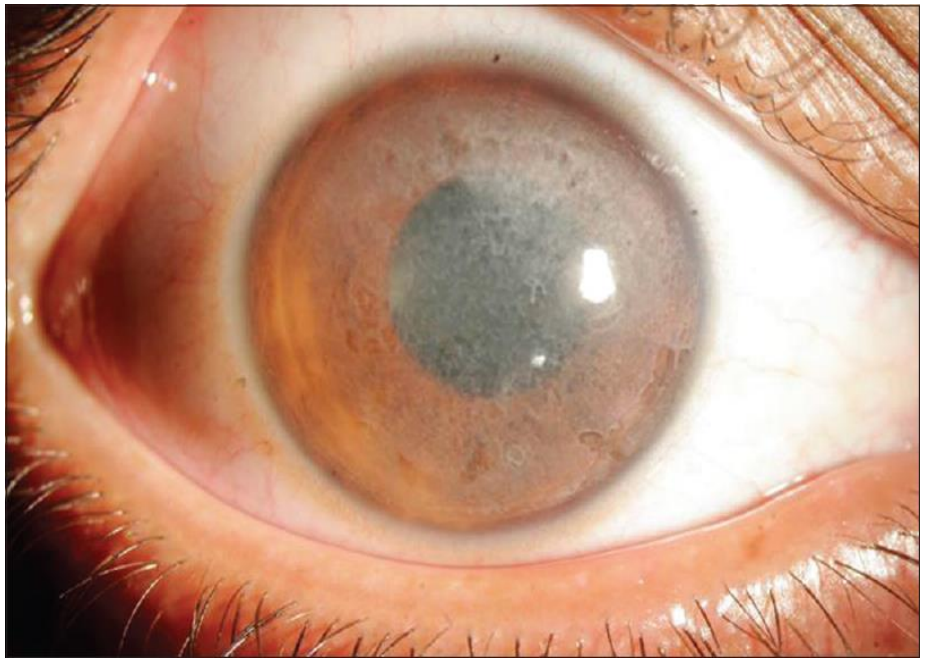
- Urine analysis – **urine glucose – trace** ; blood sugar - normal
  - **Proteinuria** - albumin- 1+ , spot PCR - 1
  - **Phosphaturia** + + ;

    tubular reabsorption of phosphate – 70% (decreased)
  - **Urinary spot potassium** -20, (increased)
  - Calcium creatinine ratio (0.2) - normal
  - **Urine Ph : 5.2 (acidification intact)**
- **ABG -**

  metabolic acidosis with compensatory respiratory alkalosis

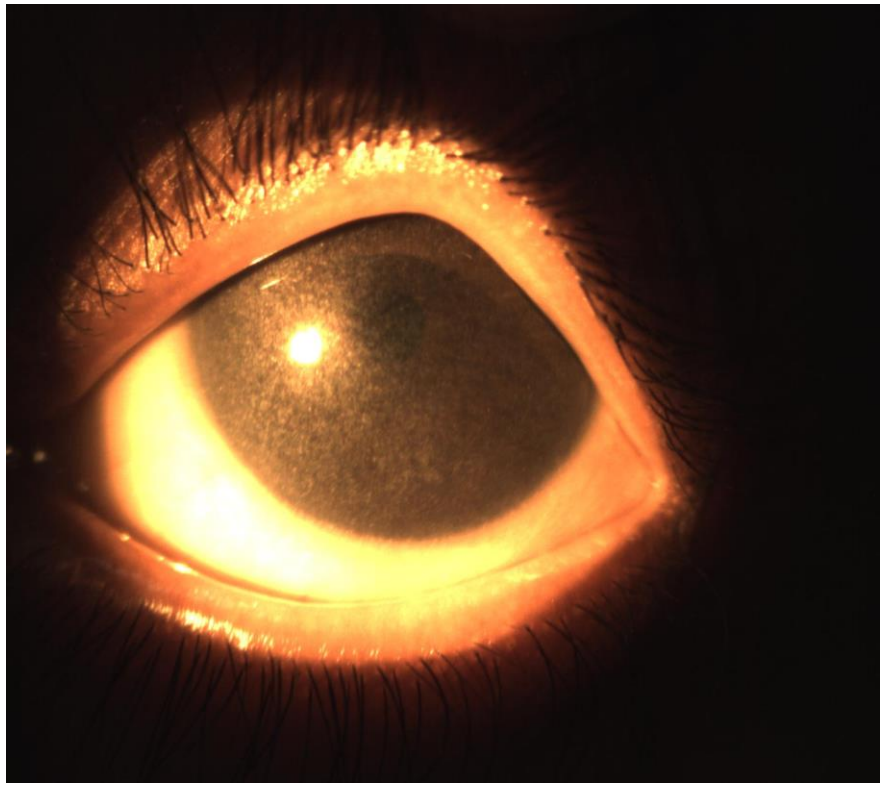
PROXIMAL RTA

- Slit lamp examination of cornea - cystine crystals
- Blood leucocyte cystine levels - awaited

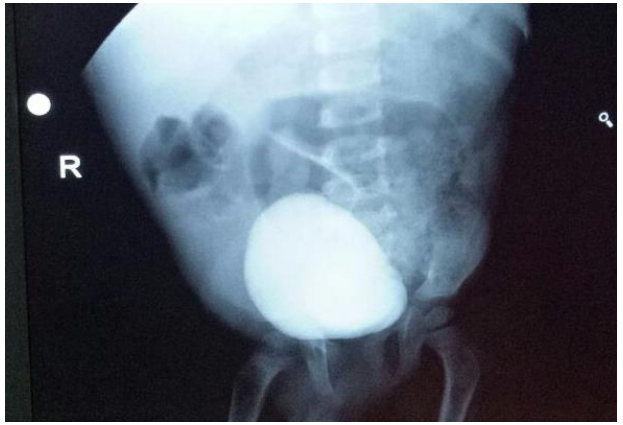


FINAL DIAGNOSIS

Nephropathic cystinosis



- Bone age: delayed bone age of 1and half years
- Thyroid function test - hypothyroidism
- USG - B/L hydro uretronephrosis
- MCU - Grossly distended bladder - megacystis. No obstruction/reflux.
- secondary to many years of massive water drinking and renal handling of excessive fluids., compensatory increase in size of entire renal system



CASE REPORT

Year : 2016 | Volume : 27 | Issue : 3 | Page : 598-601

Infantile nephropathic cystinosis with incomplete fanconi syndrome, hypothyroidism, hydro-uretero-nephrosis, and megacystis

Vaishali More, Preeti Shanbag

DISCUSSION

- Cystinosis is a autosomal recessive disorder
- Incidence:1 in 1 lakh to 2 lakh
- Pathophysiology: mutation in CTNS gene encoding cystinosin
- Cystine accumulation is associated with development of Fanconi syndrome with progressive renal damage.
- Affects – eyes, kidneys and thyroid
- Corneal crystals - seen by one year of age, which are patchy in distribution and become hazy by 10-20 years
- Rickets - 2 years
- Serious, non-renal, manifestations - second decade of life

Diagnosis

- Cystine crystals in cornea
- Increased cystine in leucocytes
- Antenatal diagnosis: Increased cystine in cultured fibroblast from placenta
- CTNS gene mutation study

Treatment:

- Fluid management – RFT normalised
- Acidosis – oral bicarbonate supplementation
- phosphate supplementation
- Hypothyroidism – Treated

PROGNOSIS

- Cystinosis progress to ESRD within 5 to 10 years but PROGNOSIS has greatly improved with specific treatment

Specific treatment

- Rare renal disorders which has specific treatment
- CYSTEAMINE - breaks down cystine into cysteine and cysteine–cysteamine disulfide, which are removed by the cysteine transporter and the lysine/arginine (PQLC2) transporter, respectively. This facilitates lysosomal transport and decreases tissue cystine
- Not available in India . Imported from Europe . Cost 2 lakhs for 3 month treatment
- Ocular - cysteamine eye drops

Launch of Cystinosis Foundation, India

TAMILNAD KIDNEY
RESEARCH FOUNDATION

ALMOST THERE - IIT-M develops drug for rare genetic disorder

U Tejonmayam| TNN | Aug 27, 2015, 07:40 IST

A- A+

Only 11 Of 19 Children In India Diagnosed With Cystinosis Are Alive Researchers in

THANKYOU