

A girl with cystinosis with bone and eye complications: case presentation

Nicholas J A Webb
Royal Manchester Children's Hospital
Manchester, UK

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Case history

- Born 29 10 2005 BW 4.3kg
- Parents first cousins. Pakistani migrants to UK.
- Presentation Nov 2006 at 13 months of age

- Polyuria
month

- Previous

- Acute
function

- Renal

- Diagnosis

- WBC c

- Eye ex

- Homozygous for c.18_21delGACT mutation in exon 3 of CTNS

- Also found to have right duplex kidney with non-functioning upper moiety. Radiological evidence of bilateral renal dysplasia



thrive from 9

d

d impaired renal

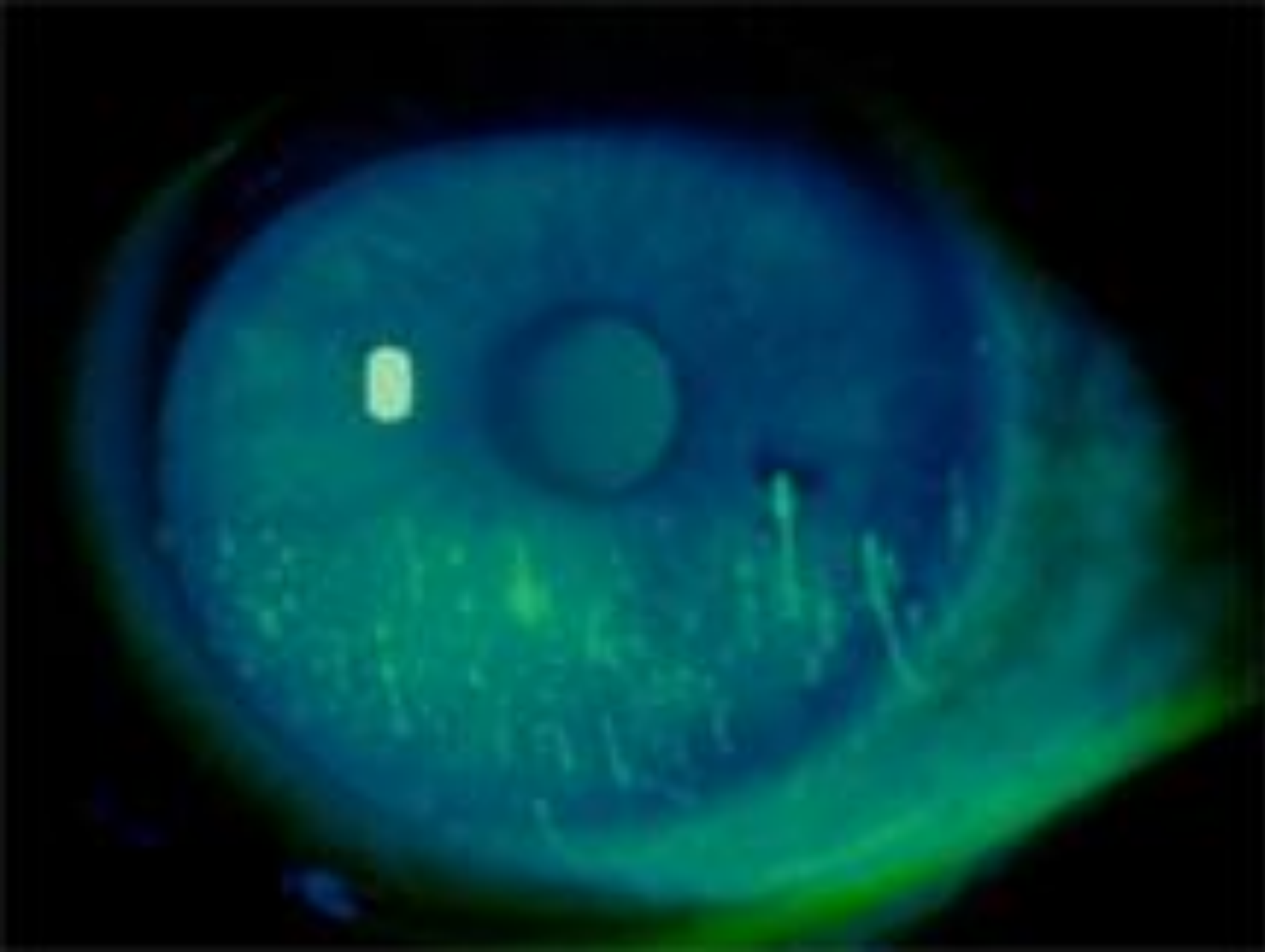
3 of CTNS

Case history

- Commenced treatment
 - Mercaptamine bitartrate
 - Cysteamine eye drops
 - Phosphate Sandoz
 - Potassium citrate
 - One alphahydroxycholecalciferol
 - Indometacin
 - Omperazole
 - Sodium bicarbonate
 - Erythropoietin
- Discharged home with stable biochemistry
 - Plasma creatinine $43\mu\text{mol/l}$

Case history

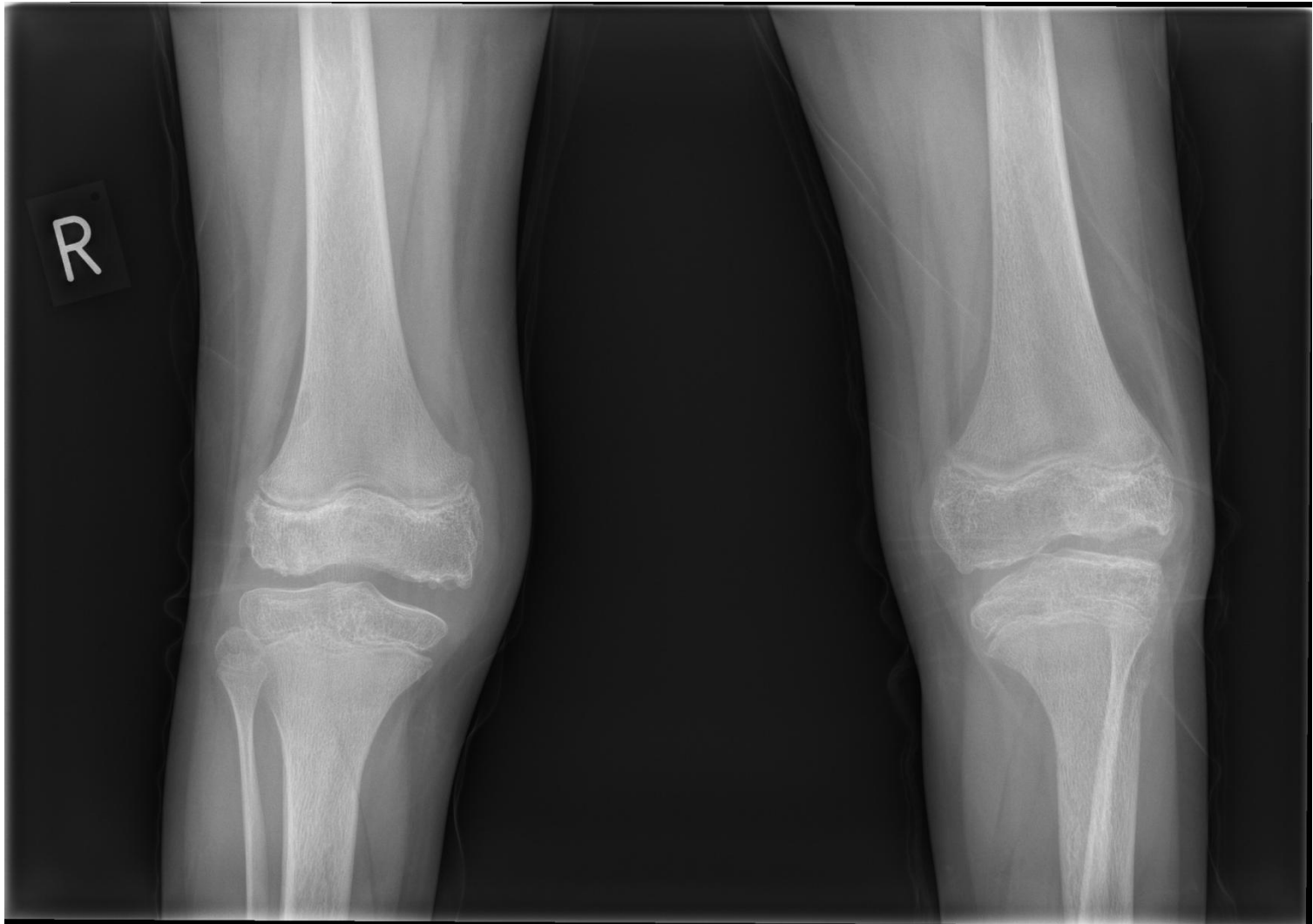
- Poor adherence with all prescribed therapy
 - Poor WBC cystine control
 - Continuous photophobia
 - Minor electrolyte abnormalities
 - However adequate PTH control and no radiological evidence bone disease
- Progressive deterioration in renal function
 - Rise in plasma creatinine from 2 years of age
 - Listed for transplantation July 2013
- Pre-emptive deceased donor transplant Nov 2013
 - 46y male. Subarachnoid haemorrhage. 1:1:0 mismatch. CMV D-/R-
 - TWIST immunosuppressive regimen – basiliximab x2, tacrolimus, MMF with corticosteroid withdrawal at D4.
 - Good primary transplant function
 - No early complications – discharged home D13 – plasma creatinine 53 μ mol/l



Bone problems

- Onset of left knee pain December 2013
- Progressive worsening with difficulty weight bearing
- Development of pain in right knee and left hip
- Orthopaedic review
 - Fixed flexion deformities of left hip (20°) and left knee (30°)
 - Decreased range of movement

Knee X-ray



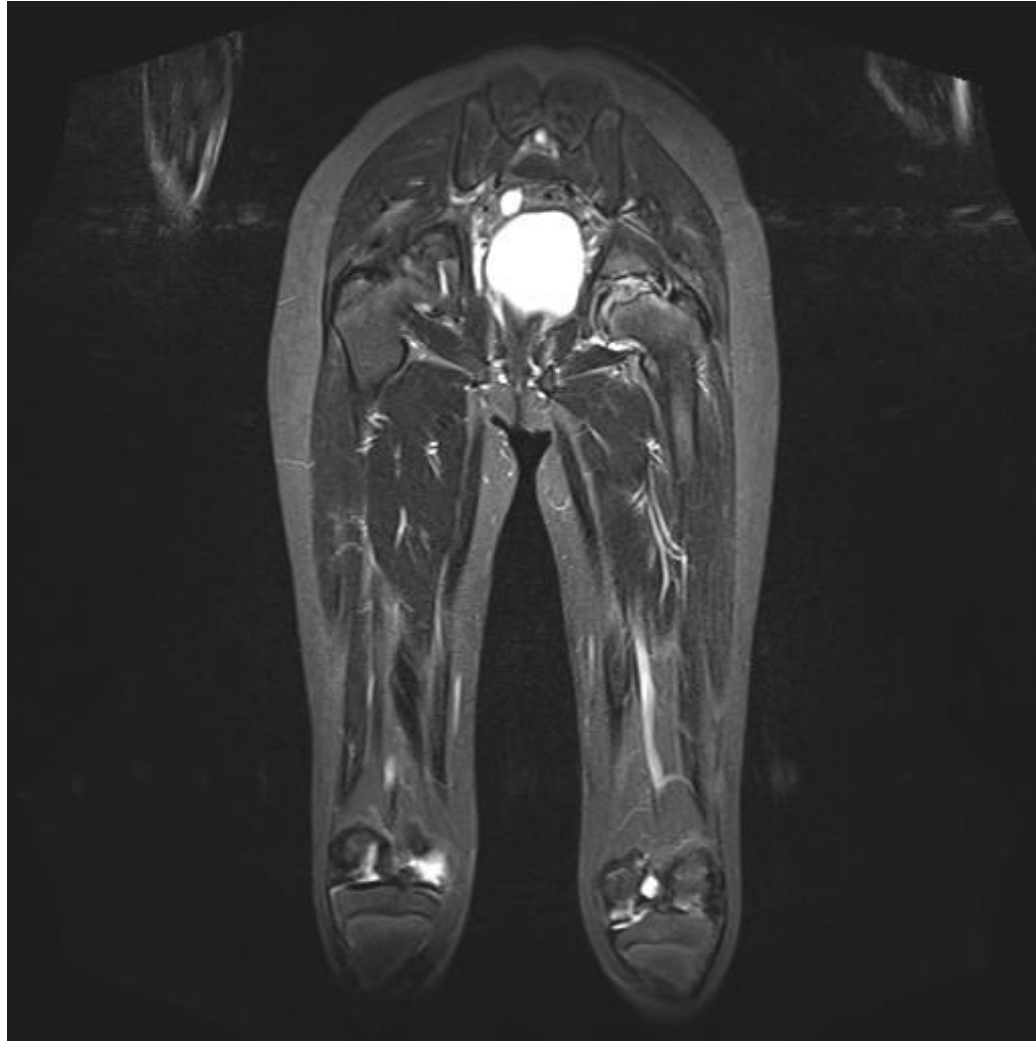
Changes consistent with avascular necrosis in left lateral femoral condyle and right medial femoral condyle. Significant chronicity of changes.

Hip X-ray



Changes consistent with avascular necrosis of both femoral heads, L>R

Hip MR coronal T2



High signal in left femoral head consistent with changes secondary to avascular necrosis. Other views show similar, less severe changes in right hip.

Treatment and progress

- Physiotherapy initially counter-productive
- Required escalating doses of analgesia
- Became wheelchair dependent
- Surgery April 2014
 - Bilateral knee arthroscopy
 - Extensive abnormality with degeneration of articular cartilage – removed
 - Microfracture performed using K-wire
 - Infusion of local anaesthetic
- Significant improvement in symptoms
 - More mobile, analgesia free
- Kidney function remains stable

Summary and introduction of expert speakers

- Significant ocular and orthopaedic symptoms both presenting following kidney transplantation
- Wide differential diagnosis in the patient with cystinosis, particularly with increasing age
- Introduction of expert speakers
 - Dr Martine Besouw
 - Mr Sus Biswas