The child with haematuria and dysphagia

Markus J. Kemper, Rainer Ganschow¹, Knut Helmke² and Dirk E. Müller-Wiefel

Keywords: Alport syndrome; dysphagia; haematuria; leiomyomatosis

Case report

A 1.5-year-old girl was referred to our centre because of microscopic haematuria. This was discovered by chance at the age of 12 months by a general paediatrician during an upper respiratory tract infection.

Physical examination at presentation revealed no abnormalities. Her body weight was at the 50th and her length at the 97th percentile. Her blood pressure was 85/60 mmHg. The family history was negative regarding renal disease and deafness, and testing of the patient’s urine revealed no abnormality.

The serum creatinine was 0.4 mg/dl and repeated testing for serum C3, C4, AST, ANA, ds-DNA, p- and c-ANCA antibodies was normal. The urine contained 200 erythrocytes/µl (69% dysmorphic), red blood cell or white cell casts could not be demonstrated. There was no proteinuria initially. The urinary calcium/creatinine ratio (mg/mg) was 0.04. Renal ultrasound revealed no abnormality. A hearing test was normal and an ophthalmologic investigation showed no lenticonus anterior or cataracts.

During follow-up the patient developed macroscopic haematuria during infections and mild glomerular proteinuria (221 mg/m²/day). Renal biopsy was recommended but refused by the parents. At the age of 5 years she developed difficulties swallowing food but not liquids (one episode of dysphagia about every 3 weeks) for which she was investigated at another hospital. Achalasia of the oesophagus was diagnosed on barium swallow and a significant obstruction of the distal oesophagus was excluded by endoscopy.

As symptoms persisted, the parents agreed in a renal biopsy and endoscopy of the oesophagus at the age of 10 years. A repeated barium swallow at that time
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confirmed an achalasia-like picture but the presence of leiomyoma were also suspected (Figure 1). Ultrasound of the distal oesophagus (Figure 2) as well as endoscopy (Figure 3) then clearly revealed diffuse leiomyomatosis of the oesophagus. Furthermore, the renal biopsy showed typical features of Alport syndrome with splitting of the glomerular basement membrane.

The diagnosis of Alport syndrome with diffuse leiomyomatosis of the oesophagus was made. The typical molecular features (contiguous gene deletions of the COL4A5 and COL4A6 genes with COL4A6 breakpoints in intron 2) could also be demonstrated in our patient [1].

Teaching point

Alport syndrome with diffuse leiomyomatosis should be suspected in the patient with combined haematuria (and or proteinuria) and dysphagia—even in childhood [2–4]. Diffuse leiomyomatosis can mimic achalasia on barium swallow but has characteristic features on endoscopy and sonography.

Suggested reading