Nephroquiz for the Beginner
(Section Editor: M. G. Zeier)

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Renal anaemia of an unusual origin

Case

A 23-year-old nurse was admitted because of iron deficiency anaemia. The history indicated that she had had gross haematuria since early childhood. The episodes had regularly been diagnosed as ‘cystitis’ and she had been given antibiotics on numerous occasions. Frequent cystoscopies were negative. One urologist prescribed an ‘oil’ to be instilled in the bladder about which we obtained no further information. She occasionally noted right-sided loin pain and dysuria but denied fever, bleeding disorders, passage of clots, or stones. Iron deficiency anaemia had been diagnosed in the past, although uterine bleeding was not excessive and no gastrointestinal losses were apparent. On physical examination she was normotensive and, except for pale mucous membranes, otherwise completely normal. Her haematocrit was 22 vol%, the iron was 2.2 μmol/l, transferrin 0.03 g/l, transferrin saturation 3%, ferritin 2.3 μg/l. The reticulocyte count was inappropriately low while the LDH and haptoglobin levels were normal. The urinalysis showed a monotonous sea of erythrocytes with some dysmorphic forms. The protein excretion was 600 mg/24 h. A renal biopsy was performed which was entirely normal by light microscopy (Figure 1). Iron substitution, first parenterally and then orally, was begun which resulted in a prompt reticulocytosis. An electron microscopic analysis (Figure 2) revealed a basement membrane that was variable in thickness, frequently decreasing to 150 nM. The normal thickness is 300–400 nM.

Question

What is your diagnosis?

Fig. 1. Light microscopy section showing normal renal architecture (PAS; ×250). An immunofluorescent study (not shown) showed no staining.

Fig. 2. Electron microscopy showing thinning of the basement membrane (×15000). The upper arrow indicates a portion of basement membrane with reduced width averaging approximately 140 nM. The lower arrow indicates a normal portion with a width of approximately 300 nm.
Thin basement membrane disease is relatively common and the only finding is the diffuse or irregularly thinned basement membrane observed with transmission electron microscopy [1–3]. The disorder is probably familial with autosomal-dominant inheritance. Our patient is an only child and the parents are not available for study. Most patients are asymptomatic although gross haematuria, as in our patient, is not infrequent [4]. Proteinuria is usually less than 1.0 g/24 h. Hypercalciuria and hyperuricosuria are associated with haematuria and have also been described in thin basement membrane disease [5]. Thin basement membrane disease must be distinguished from other renal disease, including IgA nephropathy and other hereditary nephritides [6]. Renal biopsy is not routinely warranted in patients with isolated haematuria. We elected to perform the procedure to relieve the patient’s anxiety and to dissuade further urological diagnostic procedures. The prognosis is generally good. Angiotensin-converting enzyme inhibition may be beneficial, perhaps by lowering intraglomerular pressure. To our knowledge, thin basement membrane disease presenting as iron deficiency anaemia has not been previously reported. We found no other cause for haematuria or iron deficiency.

References