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CLINICAL QUIZ

Gross hematuria in a case of Wilson disease: Questions

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Case report

An eight-year-old boy with the primary diagnosis of Wilson disease and maintained on penicillamine was referred to pediatric nephrology for recurrent episodes of gross hematuria. There was no history of vomiting or diarrhea, and physical examination was unremarkable apart from a slightly enlarged liver. Initial urine examination confirmed significant red blood cells (40–50 /hpf) even during periods when he was not having gross haematuria. Renal parameters were as follows: serum sodium (136 mmol/L), potassium (3.3 mmol/L), chloride (111 mmol/L), calcium (8.1 mg/dl), bicarbonate (16 mmo/L), albumin (4.1 g/dl), and creatinine (0.6 mg/dl). Serum complements (C3 and C4) were normal, and phase-contrast microscopy of the urine showed no significant dysmorphic red blood cell. His renal ultrasound scan was also reported as normal. Twenty-four-hour urine collection showed insignificant proteinuria (185 mg) but grossly elevated urinary calcium of 215 mg (normal 4 mg /kg/ 24 h=88 mg). The rest of his urinary electrolytes were as follows: sodium 50 mmol/L, potassium 61.3 mmol/L, and chloride 74.7 mmol /L.

1. What is the likely cause of hematuria?
2. What could be the possible correlation between the primary diagnosis of Wilson disease and hematuria?
3. What further investigations need to be done to confirm the diagnosis?

The answers to these questions can be found at http://dx.doi.org/10.1007/s00467-011-2011-x

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