Is there anything good in uric acid?

SIR,

This recent review gives an excellent overview of uric acid and its disease associations. We would like to add that low levels of uric acid may be a significant and pathological finding; and that there are other important inherited causes of hyperuricaemia.

As the reviewers noted, the kidney plays a significant role in uric acid handling. Idiopathic renal hypouricaemia is an inherited form of hypouricaemia that is characterized by excessive urinary wasting of uric acid leading to an increased fractional excretion of uric acid. While most patients are asymptomatic, some may present with uric acid nephrolithiasis or acute kidney injury following severe exercise.1 Mutations in SLC22A12 encoding the URAT1 transporter2 and SLC2A9 encoding the GLUT9 transporter3 underlie this condition. Initial reports of this disorder were limited to Japan, Korea and China, but we have recently reported SLC22A12 mutations in European patients with nephrolithiasis.4 In these patients serum uric acid was low, ranging between 0.72 and 2.02 mg/dl (normal reference range 2–7.5 mg/dl).

In addition to mutations in UMOD, there are other inherited renal disorders that may lead to hyperuricaemia and renal disease. These include the Renal Cysts and Diabetes syndrome where mutations in TCF2 encoding the transcription factor HNF1b, which regulates UMOD, may lead to hyperuricaemia, gout, tubulointerstitial nephropathy and end-stage renal disease.5 Also, mutations in REN may lead to early onset anaemia, hypouricosuric hyperuricaemia and progressive kidney failure.6 These hyperuricaemic conditions are inherited in an autosomal dominant manner and should be considered where there is a family history of gout and/or renal disease.

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doi:10.1093/qjmed/hcs006
Advance Access Publication 20 January 2012