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Diagnostic approach to Hereditary renal hypouricemia

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Hereditary renal hypouricemia is a new genetic disorder affecting renal transport of uric acid (UA). This disorder predispose the patients to exercise-induced acute renal failure and/or nephrolithiasis. The known causes are defects in the *SLC22A12* gene, encoding the human urate transporter 1 (hURAT1), and impairment of *SLC2A9* gene, which encodes GLUT9 transporter. Diagnosis is based on hypouricemia (<119 μmol/l) and increased fractional excretion of UA (>10%). To date, this disorder have been reported in East Asia mainly. More than one hundred Japanese patients have been described. Hypouricemia is sometimes overlooked, therefore we have set up the flowchart. The patients were selected for molecular analysis from 660 hypouricemic patients. Other secondary causes of hypouricemia such as Fanconi syndrome or drug-induced tubulopathy were excluded. The estimations of : 1) serum UA, 2) excretion fraction of UA, 3) and analysis of *SLC22A12* and *SLC2A9* genes followed. We have found 3 transition, 4 deletions in *SLC22A12* gene and two insertions in *SLC2A9* gene in 9 Czech patients, which is the second group worldwide, in terms of number of patients. Three patients had acute renal failure and urate nephrolithiasis. In addition, we have detected two transitions in the *SLC2A9* gene in two boys from UK.

Conclusions: Hereditary renal hypouricemia is still unrecognized disorder and probably not wide spread in East Asia only. Asymptomatic hypouricemia may be a risk factor for kidney injury. Patients with unexplained hypouricemia need detailed purine metabolic investigations.

Biography

Ivan Sebesta, MD has completed his PhD from First Medical Faculty, Charles University. He worked in Purine Research Laboratory, University of London. He has published more than 25 papers in reputed journals. He received Award of the Ministry of Health of Czech Republic for Science and Research in 1999. During the period 2004-07, he was the President of Purine & Pyrimidine Society. In 2005, he organized 10th Symposium of this Society in Prague. He works as deputy of head for teaching and is involved in research and clinical care of patients with inborn errors of metabolism.

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