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P041 -Adult presentation of Gitelman syndrome – don't ignore persistent hypokalemia

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A 65 year old gentleman was referred to the department of nephrology with a 15 year history of lethargy, restless legs and nocturia every few hours.

He denied salt cravings, had no history of nephrolithiasis and no family history of either nephrolithiasis or tubulopathies.

His serum potassium had been documented as low with a metabolic alkalosis. His total magnesium had always been within the normal range and his serum calcium normal with a low urinary calcium excretion. His hypokaleamia had been treated symptomatically with ACE inhibitors and NSAIDS which were poorly tolerated and stopped. His hypokalaemia had been treated with high doses of oral potassium. His blood pressure was low at 114/72 with no significant postural deficit.

Urinalysis was negative for blood, protein and glucose and abdominal ultrasound scan showed normal sized kidneys with no hydronephrosis and no structural abnormalities.

Genetic investigations identified biallelic pathogenic variants in SLC12A3 confirming a diagnosis of Gitelman syndrome. Despite supplements leading to normal serum levels of potassium the patient remained symptomatic with continued lethargy.

Oral magnesium supplementation was added and an improvement in the patient's energy levels were noted. The addition of slow sodium further improved clinical symptoms.

Though patients typically present with Gitelman syndrome in adolescence or early adulthood, this case demonstrates that presentation much later in life is possible.

In this case the diagnosis was also delayed. Clinical diagnosis can be very challenging in patients with Gitelman Syndrome yet serum and urine biochemistry and molecular genetics can allow a precise diagnosis. Treatment can be challenging and is often trial and error.