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Liddles Syndrome: A Case Report of a Rare Monogenic Form of Hypertension

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Case Study : Liddle's Syndrome is a rare autosomal dominant disorder which occurs due to a gain of function of ENaC channel, and is characterized by early onset hypertension and hypokalemic metabolic alkalosis. It is most commonly seen in teenage children. We present an isolated case of Liddle's syndrome in a 15-year-old teenager, who presented to us with a diagnosis of hypertension since 1 year. His parents were consanguineous(parents being first cousins). His father and paternal grandfather were hypertensive. On examination, patient had a normal heart rate. His blood pressure was 142/72 mm of Hg in right upper limb despite taking T.Losartan 50mg twice daily. Systemic examination was normal with all peripheral pulses well felt. There was no renal artery bruit on auscultation. Routine investigations showed recurrent hypokalemia requiring potassium supplementation. Urine routine showed trace albumin. Urine microscopy showed 3-7 pus cells. CECT abdomen showed normal sized bilateral kidneys and adrenals and no evidence of tumors. CT renal angiogram ruled out renal artery stenosis. 8am serum cortisol was was normal which ruled out Cushing syndrome. 24 hour urine metanephrites was normal which ruled out pheochromocytoma. Serum aldosterone (<0.97ng/ml) and renin levels (1.87) were low. Aldosterone renin ratio was low (0.51). Hence hyperaldosteronism was ruled out. He did not have ACTH suppression and urinary free cortisol to cortisone levels were normal. With the above evaluation, a provisional diagnosis of Liddle's syndrome was made. Patient was started on potassium supplements and ENaC inhibitor. Blood pressure reduced to 117/72mm of Hg and serum potassium improved to normal levels on follow up. Patient is currently asymptomatic. The typical symptoms, investigation findings, and response to amiloride therapy confirms the diagnosis of Liddle's syndrome for our patient. Further genetic testing confirmed the diagnosis of Liddle's syndrome with an autosomal dominant homozygous mutation in exon 13 of SCNN1B gene.