ABSTRACTS OF E-POSTERS

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Hypertension, Hyperkalaemia and Metabolic Acidosis and Low Serum Renin Activity: A Case Report on Psuedohypoaldosteronism Type 2 in a Six-Year-Old Child

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Introduction

Pseudohypoaldosteronism type 2 (PHA II), also known as Gordon syndrome or familial hyperkalaemia and hypertension syndrome, is a rare cause of monogenic low renin hypertension. Its clinical profile commonly includes hyperkalaemia, metabolic acidosis, diminished serum renin and normal aldosterone levels. It typically follows an autosomal dominant inheritance pattern, involving mutations in *WNK1, WNK4, KLHL3*, and *CUL3* genes. Timely detection and management are crucial as these children are prone to complications arising from hypertension and hyperkalaemia. Here, we present a case where biochemical investigations played a pivotal role in arriving at the final diagnosis of PHAII.

Case Presentation

Apparently well 6 years and 2 months old girl presented with bilateral frontal headache persisting for three months. Upon examination, her blood pressure consistently exceeded the 99th percentile for age. Laboratory investigations revealed hyperkalaemia, mild hyperchloridemia and low-normal calcium level and metabolic acidosis, in the background of normal renal functions which raised suspicion on PHA II. Her plasma aldosterone concentration was markedly low, while plasma direct renin concentration fell within the low-normal range. Observation of low plasma renin activity provided further evidence of PHA II. Treatment with age-appropriate doses of thiazide diuretics was given in combination with prazosin. The normalization of blood pressure and serum potassium levels following treatment further supported the diagnosis.

Discussion and Conclusions

Patients with PHA II typically exhibit hyperkalaemia and metabolic acidosis (resembling renal tubular acidosis type IV), in the background of normal renal functions. Further they may present with mild hyperchloremia and hypocalcaemia, as observed in our case. In settings where genetic testing resources are limited, the combination of hyperkalaemia, metabolic acidosis, and low serum renin activity serves as a reliable diagnostic hallmark for PHA II.

Keywords

Aldosterone, Gordon syndrome, Pseudohypoaldosteronism, Hyperkalaemia, Metabolic acidosis