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# Hypomagnesemia as first clinical manifestation of 17q12 microdeletion including HNF1B

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**Case Study:** Background: Hepatocyte nuclear factor 1β encoded by the *HNF1B* is a member of the homeodomain-containing family of transcription factors and the most commonly identified genetic cause of renal malformations. Besides, *HNF1B* mutation is well known to associate with pancreatic dysfunction as diabetes, genitalia malformation, liver dysfunction, primary hyperparathyroidism and neuropsychiatric feature like epilepsy and autism. Chromosome 17q12 microdeletion syndrome encompasses several genes including *HNF1B* and is characterized by a diverse spectrum of phenotypic manifestations with variable expressivity.
Methods: We report here a case of hypomagnesemia as first clinical manifestation of 17q12 microdeletion including *HNF1B.*
Results: A 40-year-old woman with history of uterine didelphys presented with muscular weakness and paresthesia of both hands. Her serum creatinine and magnesium (Mg) was 0.76 (0.51-0.95 mg/dL) and 0.6 mg/dL (1.9-2.5 mg/dL), respectively. Hypocalcemia and hypokalemia were also noted as low as 6.4 mg/dL (8.1-10.6 mg/dL) and 3.1 mmol/L (3.5-5.1 mmol/L). Intact-PTH was 32.9 pg/mL. She received oral and intravenous calcium, potassium and magnesium replacement therapy; serum calcium and potassium values normalized but serum magnesium level remained low. Urine chemistry revealed pathologic hypermagnesuria with elevated fractional magnesium excretion at 10.9%. Previous abdominal and pelvic computed tomography (CT) revealed not only the female genitalia malformation but also a multiple renal cysts in both kidneys with calyceal stones in right kidney and a pancreatic cyst. Patient DNA was sent in for gene sequencing to find gene mutation related to hypomagnesemia. Array comparative genomic hybridization (array CGH) revealed a 17q12 microdeletion, which included the *HNF1B* gene.
Conclusion: This case reports the patient with 17q12 microdeletion including *HNF1B* in whom hypomagnesemia was the first and predominant symptom of disease and illustrates the impairment of renal Mg2+ handling can arise when renal function is preserved in the patient with *HNF1b* mutation.