THE HYPOCALCEMIA CONUNDRUM: WHEN LOW CALCIUM TELLS DIFFERENT STORIES

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Hypocalcemia is a frequently encountered biochemical abnormality with broad and often complex differentials encompassing endocrine, renal, and genetic disorders. We present a case series illustrating the varied etiologies and diagnostic challenges associated with hypocalcemia in a tertiary care setting. The series includes patients with Hypoparathyroidism, Pseudohypoparathyroidism, Familial Hypomagnesemia with Hypercalciuria and Nephrocalcinosis (FHHNC) Type 1 and 2. We describe four young patients who presented with diverse manifestations, each ultimately diagnosed with a different underlying cause. A 21-year-old man developed recurrent carpal spasms a few days after dengue fever; evaluation revealed hypocalcemia, hyperphosphatemia, low parathyroid hormone, and widespread intracranial calcifications, leading to a diagnosis of hypoparathyroidism. The second patient, a boy with recurrent renal stones since early childhood, had low calcium and magnesium, raised creatinine, elevated parathyroid hormone, hypercalciuria, hypermagnesuria, bilateral nephrolithiasis and nephrocalcinosis; genetic testing revealed a homozygous missense mutation, confirming FHHNC type 1. The third patient, a young girl with medullary nephrocalcinosis since age 5, also exhibited myopia, horizontal nystagmus, and bilateral macular atrophy; biochemical testing showed hypomagnesemia, hypocalcemia, secondary hyperparathyroidism, renal dysfunction, urinary calcium and magnesium wasting; whole-exome sequencing identified a novel mutation, establishing FHHNC type 2. The fourth patient, a 16-year-old boy with recurrent convulsions, round facies, short stature, brachydactyly, and subcutaneous ossifications, demonstrated hypocalcemia, hyperphosphatemia, elevated parathyroid hormone, and basal ganglia calcifications, consistent with pseudohypoparathyroidism. This case series underscores that hypocalcemia is not a singular entity but a manifestation of heterogenous disorders, and careful clinical and biochemical correlation is critical for accurate diagnosis and tailored care.

Keywords: Hypocalcemia , Endocrine Disorders , Diagnostic Challenges

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