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Pseudohypoparathyroidism Type 1B: A Medication Compliance Challenge

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Introduction: Pseudohypoparathyroidism (PHP) encompasses five metabolic disorders characterized by target organ PTH resistance. The focus of this paper is PHP Type 1B (PHP1B), a disease that typically presents in children with symptomatic hypocalcemia. Only 15-20% of cases are genetic, making our case of Familial PHP1B even rarer. **Case Presentation:** A 23-year-old African-American female with hypothyroidism and PHP1B diagnosed in 2005 presented to the emergency department with symptomatic hypocalcemia. As a child, she was evaluated for progressive femur, tibia, radius, and ulna bowing in the setting of short stature without phenotypic features of Albright's Hereditary Osteodystrophy. Her biochemical profile was consistent with pseudohypoparathyroidism and imaging showed costochondral junction enlargement. Genetic testing revealed STX mutation confirming PHP1B diagnosis. Extensive studies of her family revealed several affected members, including one of her two children.Patient has history of multiple hospitalizations for symptomatic hypocalcemia. During this admission, her laboratories demonstrated: calcium 4.4 mg/dL (8.6-10.3 mg/dL), phosphorus 7.3 mg/dL (2.5-4.5 mg/dL), PTH 574 pg/mL (16-65 pg/mL), 1,25-dihyroxyvitamin D < 8 ng/mL (18-78 ng/mL), and 25-hydroxyvitamin D 7.6 ng/mL (30-100 ng/mL).She has an extensive history of non-compliance with medications and endocrinology appointments. During the last visit, she complained of ostealgia, particularly in long bones. While discussing the importance of maintaining adequate calcium levels, the patient expressed feeling overwhelmed by her regimen (calcitriol 1 mcg QAM and 0.5 mcg QHS, calcium carbonate 1,200 mg TID). After a lengthy conversation regarding the need for long-term medical therapy, she became increasingly distraught. **Conclusion:** When normocalcemia is achieved with high doses of calcium and calcitriol, patients with PHP1B have an average life expectancy. Given that patients are typically diagnosed in childhood, taking several medications multiple times a day can be challenging and lead to poor compliance. When this occurs, PHP1B becomes a life-altering disease with a significant decrease in quality of life.In 2018 Wierenga et al. reported that being emotionally overwhelmed by chronic disease prevents a patient from appropriately caring for themselves. Their research shows that older males with higher levels of education have better emotional regulation than young females with low educational levels. Providing patients at high risk of

psychological stress with a multidisciplinary team including cognitive-behavioral therapists can improve their quality of life and compliance while decreasing the healthcare costs associated with poorly controlled chronic diseases.

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