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Case Report

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A Case Presentation of Calcium in an Active Adolescent Male with CaSR Mutation Disorder

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Abstract

Patients with mutation in the calcium-sensing receptor (CaSR) present with hypocalcemia, hyperphosphatemia, with low or inappropriately normal parathyroid hormone. This presentation follows the fluctuating calcium levels of an active adolescent male (aged 15-16 years of age) during his involvement in high school athletics as well as during periods of recovery from various orthopedic injuries and medication adjustments needed to maintain a normal level of calcium to avoid emergent intervention.

Keywords: Calcium-Sensing Receptor; Hyperphosphatemia; Parathyroid Hormone; Orthopedic; Autosomal Dominant Hypocalcemia Type 1

Introduction

Autosomal dominant hypocalcemia type 1 (ADH1) is a rare genetic endocrine disorder caused by an inherited or de novo heterozygote activating mutation of the calcium-sensing receptor (CaSR) gene, located on chromosome 3q13.3–q21. In ADH1, the CaSR is activated, even in the presence of normal or low serum calcium levels, resulting in the suppression of PTH synthesis and release, a decrease in renal Ca+2 reabsorption, causing hypocalcemia with relative hypercalciuria. ADH1 prevalence was estimated at 3.9 per 100,000 in a recent study of 51,289 subjects from a single US healthcare system. Over 100 distinct germline-activating mutations of the CaSR gene have been described in association with ADH1. The mode of inheritance is usually autosomal dominant; however, sporadic mutations have also been reported and the diagnosis can be confirmed by DNA analysis. The patient in question in this case is one of the sporadic mutation variants. The clinical presentation of ADH1 is heterogeneous and may vary from mild hypocalcemia which is not diagnosed until late adulthood, to more severe presentations during infancy or childhood with recurrent seizures. This case is to present adjustments in medication management in maintaining close to normal calcium levels in an adolescent male whose activity level varied with high school sports during the two-year period followed.

Case Presentation

The patient has a disorder of an activating mutation of the calcium-sensing receptor (CaSR) gene which was initially presented at 3 weeks of age due to observed seizures resulting from severe hypocalcemia. Occasional admissions in an inpatient setting were needed to address and correct periods of both hypocalcemia and hypercalcemia. He has been treated with various doses of calcitriol, calcium carbonate, and hydrochlorothiazide. The calcitriol was a concentrated solution of 1mg/ml regimen. Calcium carbonate was given in the form of OTC TUMS. Hydrochlorothiazide was initially given in oral concentration of 50mg/cc and then to oral doses of 12.5mg tablets. The patient was followed over a 2-year period during his adolescence of ages 15 through 16 while attending a large, public high school in the Northern California area. During these years, the patient (MK) was noted to have a 2-inch growth and was involved in year-round sports with varying degrees of activity intensity [1-3]. Patient MK was also noted to have periods of inactivity when recovering from surgeries involving removal of pectus bar for repair of pectus xcavatum and from a broken right thumb needing casting (which ended his seen prematurely). His

calcium level was monitored weekly with adjustments in his medication regimen made to correct his calcium level with the goal of being in normal range. This was pursued to allow the patient to continue to participate in his high school athletic activities. What was observed was the expected utilization of calcium during periods of growth and increased activity resulting in instances of potential hypocalcemia as well as elevated calcium levels seen when there was a decrease in activity and when recovering from surgery. Calcium levels were checked weekly, and his medication regimen was adjusted accordingly to maintain low normal values. Changes were made for the sole target of the calcium level to be close normal levels allowing the patient MK to continue to participate in sport [4,5].

Data

[Left Y-axis (calcium level and number of days per week of practice), X-axis (date), Right Y-axis (calcitriol dose)]

(Figure 1-3).

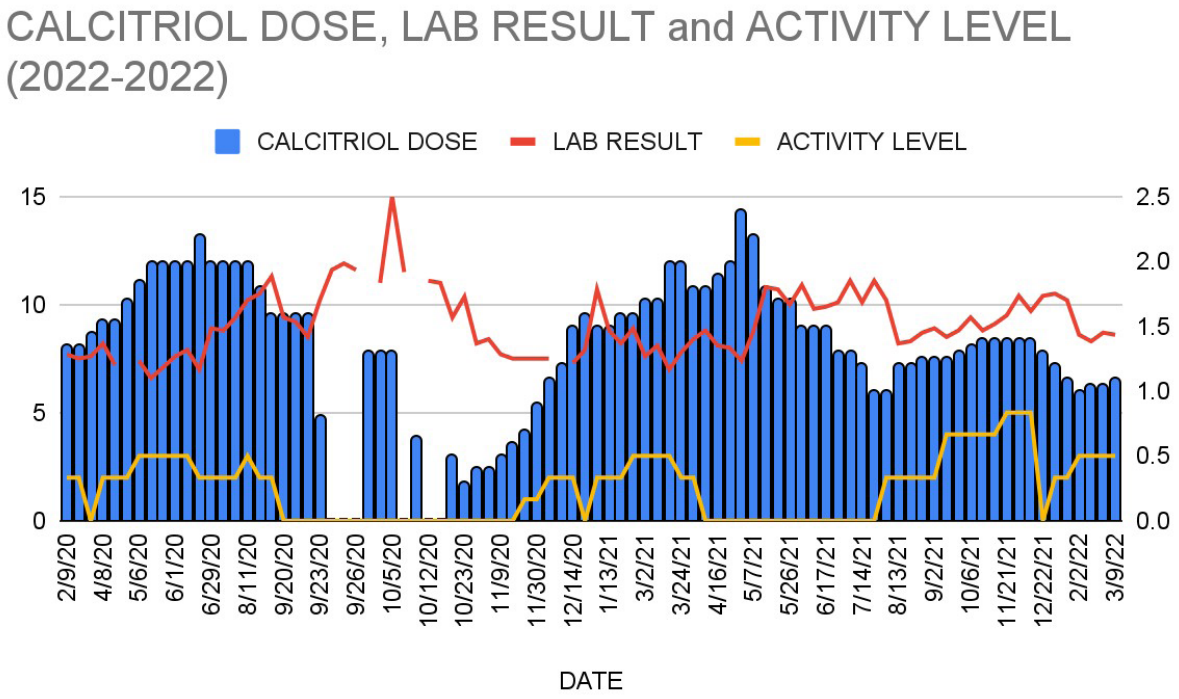

Figure 1: Calcitriol Dose, Lab Result and Activity Level (2020-2022).

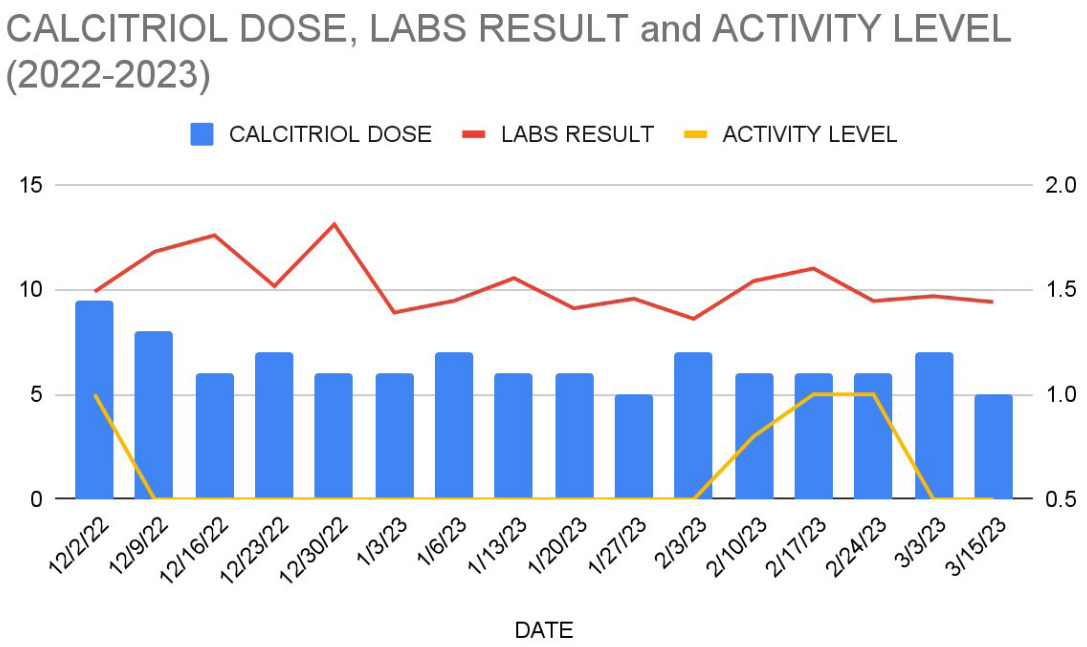

Figure 2: Calcitriol Dose, Lab Result and Activity Level (2022-2023).

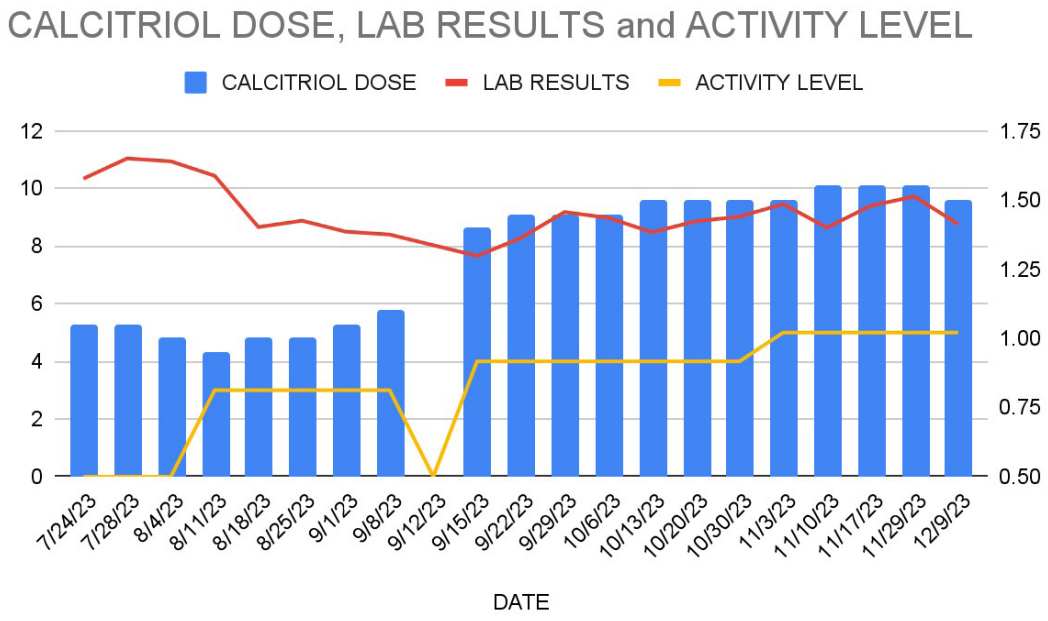

Figure 3: Calcitriol Dose, Lab Result and Activity Level.

Discussion

The clinical presentation of ADH1 is heterogeneous and may vary from mild hypocalcemia which may note present until late adulthood to more severe presentations during infancy or childhood with recurrent seizures. This disorder can have a great impact on the quality of life in children and their transition into adulthood. Being able to participate in various activities allows a sense of belonging, normalcy, and improvement in overall mental health, all of which may be difficult for many children with chronic medical issues. The tables above show the influence varying activity levels play in calcium levels and the need for medication dose adjustments during these times. Specifically, an increased need for calcitriol and calcium carbonate will likely be needed during times of intense exercise. This case is an example of the need for frequent monitoring of calcium and adjustments in treatment in this adolescent age group with a desire to participate in sports or various activities along with their peer group.

References

1. Kohrt WM, Wherry SJ, Wolfe P, Sherk VD, Wellington T, et al. (2018) Maintenance of Serum Ionized Calcium During Exercise Attenuates Parathyroid Hormone and Bone Resorption Responses. J Bone Miner Res 33(7): 1326–1334.
2. Barry DW, Kohrt WM (2007) Acute effects of 2 hours of moderate-intensity cycling on serum parathyroid hormone and calcium. Calcif Tissue Int 80(6):359–65.
3. Barry DW, Hansen KC, van Pelt RE, Witten M, Wolfe P, et al. (2011) Acute calcium ingestion attenuates exercise-induced disruption of calcium homeostasis. Med Sci Sports Exerc. 43(4):617–23.
4. Suzuki M, Aso T, Sato T, Michimata M, Kazama I, et al. (2005) A Case of gain-of-function mutation in calcium-sensing receptor: supplemental hydration is required for renal protection. Clin Nephrol. 63(6):481-6.
5. Zung A, Barash G, Banne E, Levine MA (2023) Novel Calcium-Sensing Receptor(CASR) Mutation in a Family with Autosomal Dominant Hypocalcemia Type 1 (ADH1): Genetic Study over Three Generations and Clinical Characcteristics. Horm Res Paediatr 96 (5):473-482.