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Background/Introduction:

The genetic mutations of the GNAS gene and resistance of end-organs to parathyroid hormone comprise a spectrum of disease classified as pseudohypoparathyroidism. This disease has an unknown prevalence in the US and variable phenotypic presentation and clinical course. This case report describes the clinical diagnosis of pseudohypoparathyroidism and Albright's Hereditary Osteodystrophy, a phenotypic feature present in a subset of individuals affected by pseudohypoparathyroidism with unique physical findings. This diagnosis of a rare genetic disease initially presented during the routine workup for hypocalcemia in the outpatient setting.

Methodology:

A 19 year old woman presented to the outpatient clinic for a complaint of new onset muscle spasms and cramps. Blood work revealed moderate hypocalcemia, mild hypothyroidism, and a PTH level five times above normal. Additional lab work revealed significant vitamin D deficiency which was initially determined to be the likely etiology of her PTH elevation. After adequate supplementation of both vitamin D and calcium, these levels normalized while, on multiple checks, her PTH remained elevated. Due to limited resources, endocrinology referral was not feasible; after a review of available literature physical examination revealed unique sequelae of pseudohypoparathyroidism and Albright's Hereditary Osteodystrophy, characteristic brachydactly, blue sclerae, poor dentition, short stature, round facies, and obesity.

Results:

With very limited resources available for laboratory testing, imaging, and specialist care, her case was referred to the Undiagnosed Disease Network. After their review, her diagnosis of pseudohypoparathyroidism was affirmed, and confirmatory genetic tests were recommended for further specific genetic defect and pathologic classification.

Conclusion/Discussion:

Following a course of treatment aimed at correcting an elevated PTH, which was initially felt to be due to hypovitaminosis D with concomitant hypocalcemia, a persistently elevated PTH level warranted further investigation. Unique physical examination findings greatly aided diagnosis of this rare condition, in addition to expert review of her case. At the time of this case report, she has yet to have subcutaneous ossifications typical of Albright's Hereditary Osteodystrophy. This unusual case underlines the importance of maintaining a broad differential and careful examination, in addition to providing a summary of the current literature on the diagnosis and management of the specific pathology of Albright's Hereditary Osteodystrophy within the broad category of pseudohypoparathyroidism.

References:

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