FROM MUSCLE CRAMPS TO ALBRIGHT'S HEREDITARY OSTEODYSTROPHY: A CASE REPORT

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INTRODUCTION

- Parathyroid hormone is a peptide hormone produced by the parathyroid glands, and plays an important role in calcium regulation [1,2].
- The genetic mutations of the GNAS gene and resistance of end-organs to parathyroid hormone comprise a spectrum of disease classified as pseudohypoparathyroidism. [1, 2]
- This disease classification shares a paradoxical relationship between elevated parathyroid hormone level and decreased calcium levels due to a decreased effective impact of the hormone in inciting reuptake of calcium into the blood and deposition into bone. [1-3]
- Pseudohypoparathyroidism has an unknown prevalence in the US and has a variable phenotypic presentation and clinical course. [3]
- 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.

CASE REPORT

A 19-year old woman presented to the outpatient clinic with new onset muscle spasms and cramps.

Evaluation with bloodwork revealed moderate hypocalcemia, mild hypothyroidism, and a PTH level five times above normal limits.

Additional lab work revealed significant vitamin D deficiency which was initially suspected to be the likely etiology of her PTH elevation.

After adequate supplementation of both vitamin D and calcium, these levels normalized. However the patient's PTH remained elevated despite multiple repeat testing.

Due to limited resources, an endocrinology referral was not feasible. After a review of available literature, physical examination revealed unique sequelae of pseudohypoparathyroidism and Albright's Hereditary Osteodystrophy. This included characteristic brachydactly, blue sclerae, poor dentition, short stature, round facies, and obesity (*Figure 1 A-F*).

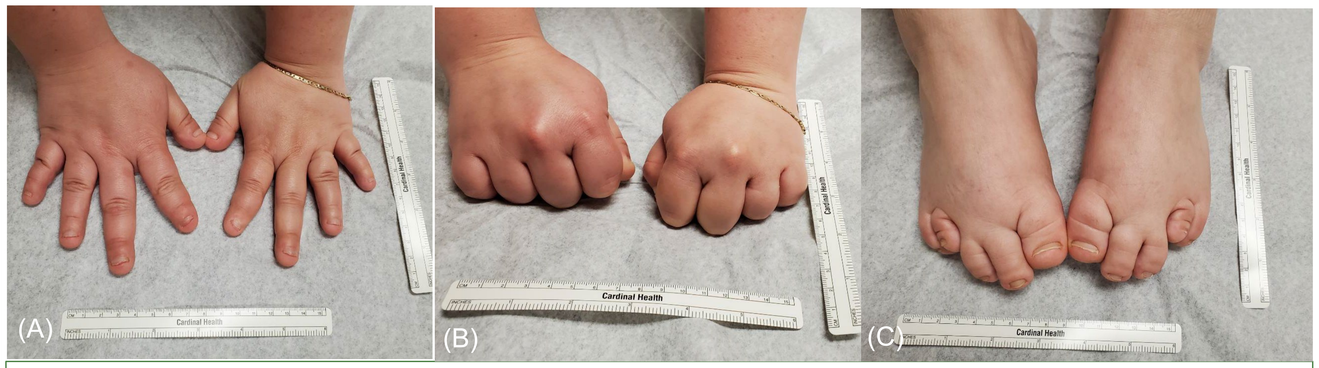
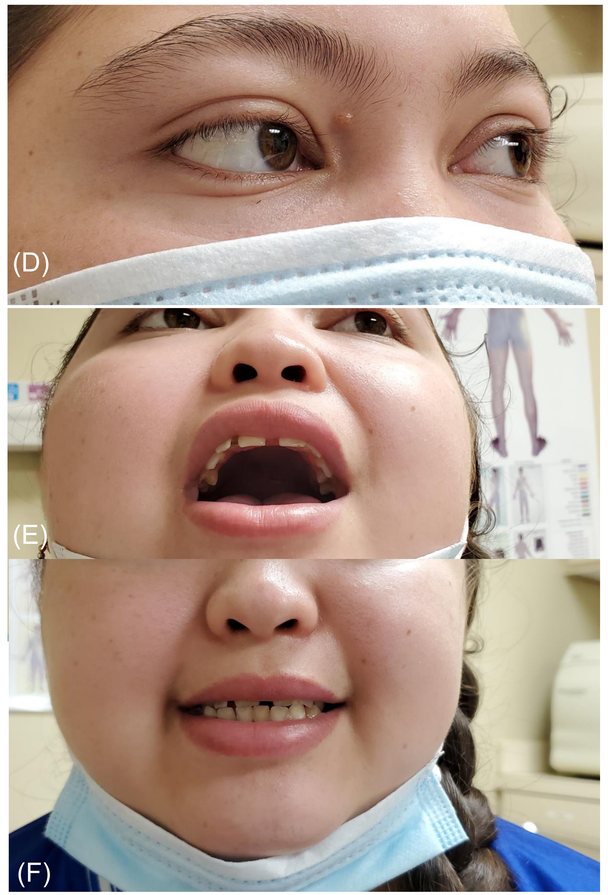


Fig 1: Albright's Hereditary Osteodystrophy characteristic clinical features in a 19-yo woman.
(A,C) Brachydactyly. (B) Hutchinson's sign. (D) Blue sclerae. (E-F) Poor dentition, round facies.



RESULTS

- With very limited resources available for laboratory testing, imaging, and specialist care, the patient's 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.

CONCLUSIONS

- A persistently elevated PTH level warranted further investigation after a course of treatment of vitamin D and calcium, as the elevated PTH was initially felt to be due to hypovitaminosis D with concomitant hypocalcemia.
- This unusual case underlines the importance of maintaining a broad differential and careful examination, and includes 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. [3,4]
- Management includes frequent monitoring of PTH and electrolyte levels, replacement of deficient calcium and vitamin D, monitoring for emergence of metabolic syndrome and hypothyroidism, genetic testing and counseling, and monitoring for skeletal changes and subcutaneous ossifications. [1]
- This case also highlights the use of resources such as the Undiagnosed Disease Network for clinical cases.

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