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CASE REPORT

Parathyroid Hormone Resistance: An Uncommon Cause of Hypocalcaemia in an Adult Ghanaian Female

Résistance à l'hormone parathyroïdienne: Une cause peu commune d'hypocalcémie chez une femme adulte ghanéenne."

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ABSTRACT

BACKGROUND AND OBJECTIVE: Parathyroid hormone (PTH) resistance, the main biochemical feature of a rare group of disorders known as Pseudohypoparathyroidism (PHP) is an uncommon cause of hypocalcaemia. In addition to the biochemical abnormalities, some individuals with PHP may have features of Albright Hereditary Osteodystrophy (AHO). Being a rare disorder with a significant level of variation and overlap in its clinical presentation, diagnosis of PHP may be challenging in some clinical settings. This case report highlights the diagnosis of this rare disorder.

CASE REPORT: A 20-year-old Ghanaian female who had been involved in a road traffic accident (RTA) was referred to the endocrine clinic after a computer tomography (CT) scan of her head revealed an incidental finding of multiple basal ganglia calcifications. Investigations revealed hypocalcaemia, hyperphosphatemia, and elevated intact PTH in the presence of normal levels of 25-hydroxyvitamin D and magnesium, and a normal kidney function. She also had phenotypic features of AHO. Findings suggested a diagnosis of PHP however the type could not be identified due to the unavailability of further testing.

CONCLUSION: This report of a Ghanaian female with PTH resistance and features of AHO diagnosed at the age of 20 years, is expected to add to the existing literature and assist in increasing the level of awareness and facilitate the diagnosis of this disorder in our setting. **WAJM 2023; 40(10); 1131-1134.**

Keywords: Hypocalcaemia, Parathyroid hormone, Ghana

RÉSUMÉ

CONTEXTE ET OBJECTIF: La résistance à l'hormone parathyroïdienne (PTH), principale caractéristique biochimique d'un groupe rare de troubles connus sous le nom de pseudohypoparathyroïdie (PHP), est une cause rare d'hypocalcémie. En plus des anomalies biochimiques, certaines personnes atteintes de PHP peuvent présenter des caractéristiques d'ostéodystrophie héréditaire d'Albright (AHO). Étant un trouble rare avec un niveau significatif de variation et de chevauchement dans sa présentation clinique, le diagnostic de PHP peut être difficile dans certains contextes cliniques. Ce rapport de cas met en lumière le diagnostic de cette maladie rare.

RAPPORT DE CAS: Une femme ghanéenne de 20 ans qui avait été impliquée dans un accident de la circulation routière (RTA) a été référée à la clinique endocrinienne après qu'une tomodensitométrie (TDM) de sa tête a révélé la découverte fortuite de multiples calcifications des ganglions de la base. Les examens ont révélé une hypocalcémie, une hyperphosphatémie et une PTH intacte élevée en présence de taux normaux de 25 hydroxyvitamine D et de magnésium et d'une fonction rénale normale. Elle avait également des caractéristiques phénotypiques d'AHO. La découverte a suggéré un diagnostic de PHP, mais le type n'a pas pu être identifié en raison de l'indisponibilité de tests supplémentaires.

CONCLUSION: Ce rapport d'une femme ghanéenne présentant une résistance à la PTH et des caractéristiques d'AHO diagnostiquée à l'âge de 20 ans, devrait s'ajouter à la littérature existante et aider à accroître le niveau de sensibilisation et à faciliter le diagnostic de ce trouble dans notre contexte. **WAJM 2023; 40(10); 1131-1134.**

Mots-clés: Hypocalcémie, Hormone parathyroïdienne, Ghana

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ABBREVIATIONS: PTH – Parathyroid Hormone, PHP – Pseudohypoparathyroidism, AHO – Albright Hereditary Osteodystrophy, RTA – road traffic accident, PTHrP – Parathyroid Hormone receptor 1, CT scan – computerized tomography, iPPSD – inactivating PTH/PTH-related Peptide signalling disorders, cAMP – cyclic adenosine monophosphate, Gsα – subunit of the stimulatory G-protein, TSH – Thyrotropin Stimulating Hormone, FT3 – Free Triiodothyronine, FT4 – Free Thyroxine, LH – Luteinizing Hormone, FSH – Follicle Stimulating Hormone,