



# PSEUDOHYPOPARATHYROIDISM WITH HYPOKALEMIA AND HYPOMAGNESEMIA: ASSOCIATION OR SEPARATE ENTITY?

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## INTRODUCTION

- Pseudohypoparathyroidism (PHP) is a group of heterogenous disorders characterized by end organ resistance to parathyroid hormone (PTH) action.
- In 1942, Fuller Albright first described the phenotype of Albright Hereditary Osteodystrophy (AHO) associated with end organ hormone resistance (brachycephaly, rounded faces, short stature, central obesity, subcutaneous ossifications, and variable degrees of mental retardation).

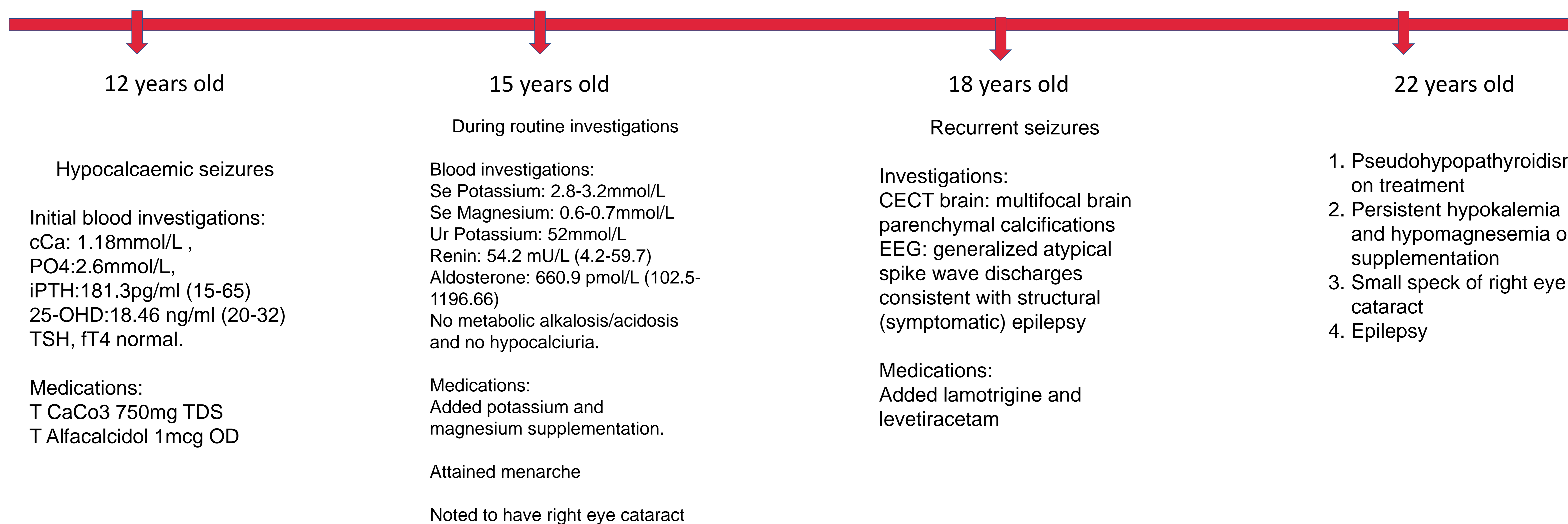
## RECENT DEVELOPMENT

- Recognizing the heterogenicity and challenges in the management of PHP, in 2018 EuroPHP network has proposed new classifications to minimize confusion and simplify this rare disease.
- Molecular defects affecting the PTH/PTH-related Peptide signaling have been described.

## CASE REPORT

- YG, currently 22 years old young lady
- First presented at 12 years of life with hypocalcemic seizures.
  - Examination revealed a short and thin girl. Height 129cm (-3.89 SD), weight 23.7kg (-4.09 SD). Normotensive, no goitre. No AHO features. Short fourth and fifth metacarpophalangeal bones.
  - Normal developmental milestone and described as an average student at school.
  - At 15 years old, noted to have asymptomatic hypokalemia and hypomagnesemia needing replacement.
  - Throughout the years she developed small speck of cataract on the right eye with normal visual acuity.
  - Diagnosed with epilepsy and started on lamotrigine and levetiracetam at 18 years of age.

## RESULTS AND MANAGEMENT



## DISCUSSION

- A young lady with pseudohypoparathyroidism accompanied by persistent hypokalemia and hypomagnesemia.
- Clinical features complicated by right eye cataract and symptomatic epilepsy.
- Only 2 case reports mentioned type 1b pseudohypoparathyroidism associated with Bartter like syndrome being described in literature.
- YG is awaiting her genetic review for molecular diagnosis
- Whether these abnormalities are directly linked with her pseudohypoparathyroidism or a different entity is yet to be determined.

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