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).

Recognizing the heterogeneity 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/PTHrP-related Peptide signaling have been described.

INTRODUCTION

• 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 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.

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 128cm (~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.
- During routine investigations: cCa: 1.18mmol/L, PO4:2.6mmol/L, iPTH:181.3pg/ml (15-32, 1196.66)
- Noted to have right eye cataract

RESULTS AND MANAGEMENT

12 years old

Hypocalcaemic seizures

Blood investigations:

- Se Potassium: 2.8-3.2mmol/L
- Se Magnesium: 0.6-0.7mmol/L
- Ur Potassium: 52mmol/L
- Renin: 54.2 mU/L (4.2-59.7)
- Aldosterone: 660.9 pmol/L (102.5-1196.66)

No metabolic alkalosis/acidosis and no hypocalcuria.

Medications:

- Added potassium and magnesium supplementation.
- Attained menarche
- Noted to have right eye cataract

15 years old

During routine investigations

Investigations:

- CECT brain: multifocal brain parenchymal calcifications
- EEG: generalized atypical spike wave discharges

Medications:

- Added lamotrigine and levetiracetam

18 years old

During routine investigations

- Recurrent seizures

- Investigations:

1. Pseudohypoparathyroidism on treatment
2. Persistent hypokalemia and hypomagnesemia on supplementation
3. Small speck of right eye cataract
4. Epilepsy

DISCUSSION

- A young lady with pseudohypoparathyroidism associated with Bartter-like syndrome being described in literature.

- YG is awaiting her genetic review for molecular diagnosis replacement.

- Whether these abnormalities are directly linked with her pseudohypoparathyroidism or a different entity is yet to be determined.

REFERENCES


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PSEUDOHYPOPARATHYROIDISM WITH HYPOKALEMIA AND HYPMAGNESEMA: ASSOCIATION OR SEPARATE ENTITY?

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