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.

RESULTS AND MANAGEMENT

12 years old

Hypocalcaemic seizures

Initial blood investigations:
cCa: 1.18mmol/L ,
PO4:2.6mmol/L,
iPTH:181.3pg/ml (15-65)
25-OHD:18.46 ng/ml (20-32)
TSH, fT4 normal.

Medications:
T CaCo3 750mg TDS
T Alfacalcidol 1mcg OD

15 years old

During routine investigations

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 hypocalciuria.

Medications:
Added potassium and magnesium supplementation.

Attained menarche

Noted to have right eye cataract

18 years old

Recurrent seizures

Investigations:
CECT brain: multifocal brain parenchymal calcifications
EEG: generalized atypical spike wave discharges consistent with structural (symptomatic) epilepsy

Medications:
Added lamotrigine and levetiracetam

22 years old

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

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.

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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