McCune Albright Syndrome in children- Case series from a tertiary care Centre- Sri Lanka

Dinendra Siriwardhane[1] Chamidri Naotunna[1] Chamila Lakmini[1] Senani Gamage[1] Dimarsha Desilva[1] Jananie Suntharesan[1] Buddhi Gunasekara[1] Raihana Hashim[1] Dilusha Premathilake[1] Navoda Atapattu[1]

1- Lady Ridgeway Hospital For Children, Sri Lanka

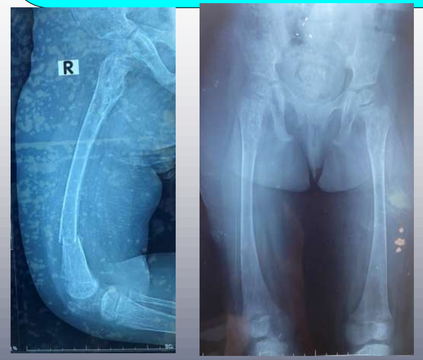
HUMAN GROWTH FOUNDATION

2022 PEDIATRIC BONE SYMPOSIUM

Introduction

McCune-Albright syndrome (MAS) is rare with a prevalence of 1 in 100,000 to 1 in 1,000,000., characterized by the triad of monostotic/polyostotic fibrous dysplasia (FD), café au lait skin pigmentation, and hyperfunctioning endocrinopathies caused by somatic activating mutations of the *GNAS1* gene encoding the α subunit of guanine nucleotide-binding protein. 1,2

Here we are reporting three cases of MAS who are actively being followed up in a leading Children's Hospital in Sri Lanka.



X rays showing fibrous dysplasia and fracture in patient 3

Patient 1 :

- 3 year old boy
- Presented with
  - Bilateral frontal prominence,
  - Multiple café au lait spots
  - Large hands and feet
  - Height >97th centile
  - Right leg deformity
  - Pubertal genitalia.
- Investigations
  - High testosterone with undetectable gonadotrophins
  - High Prolactin
  - High Growth hormone.
  - Hypophosphatemic rickets
  - X ray- fibrous dysplasia in the skull
- Management
  - started on Spironolactone, Cabergoline, Phosphate buffer, 1 alpha calcidol with regular intravenous bisphosphonate.
  - He continued to have high IGF-1 and GH level needing long acting Octreotide at 11 years.
  - *He's now 17 years and 2 months and his serum cortisol and thyroid function remained normal throughout with no effect on hearing and vision.*

Patient 2:

- 3 year old boy
- Presented with
  - Right leg deformity and fracture of the right humerus.
  - Multiple café-au-lait spots
  - Frontal prominence
  - Pre pubertal genitalia
- Investigations
  - X ray- fibrous dysplasia in the right femur and humerus with a fracture in the latter
  - At 5 years- Gonadotrophin independent precocious puberty (GIPP) with bone age of 10-11 years
  - Hypophosphatemic rickets
  - High Prolactin
- Management
  - Regular intravenous bisphosphonate and vitamin D supplements
  - Letrazole and Spironolactone started for GIPP
  - phosphate buffer with 1 alpha calcidol
  - Cabergoline for high Prolactin
  - *He's now 8 years and 10 months and other endocrinopathies are absent with normal hearing and vision*

Patient 3

- 2 year old girl
- Presented with
  - low impact right sided femur fracture
  - Multiple café-au-lait spots
  - Clinical features of thyrotoxicosis
- Investigations
  - X ray- fibrous dysplasia in the right femur with fracture
  - High T3 and T4 with low TSH
  - At 2 years and 9 months- Gonadotrophin independent precocious puberty (GIPP) with an ovarian cust on the left side
  - Management
  - Commenced on regular intravenous Bisphosphonate, anti-thyroid drugs and beta blockers.
  - Also on phosphate buffer and 1 alpha calcidol for hypophosphataemic rickets
  - Underwent thyroidectomy at 4 years due to uncontrolled thyrotoxicosis and she's currently on Thyroxine replacement.
  - For GIPP -started on oral Letrazole.
  - *Other endocrine functions are normal at 5 years*

Conclusions

MAS has various different presentations therefore it's important to follow them up regularly and actively look for developing endocrinopathies.

Bibilography

1.Dumitrescu, C. and Collins, M., 2008. McCune-Albright syndrome. *Orphanet Journal of Rare Diseases*, 3(1).

2.Zhai, X., Duan, L., Yao, Y., Xing, B., Deng, K., Wang, L., Feng, F., Liang, Z., You, H., Yang, H., Lu, L., Chen, S., Wang, R., Pan, H. and Zhu, H., 2021. Clinical Characteristics and Management of Patients With McCune-Albright Syndrome With GH Excess and Precocious Puberty: A Case Series and Literature Review. *Frontiers in Endocrinology*, 12.