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## CASE STUDY REVIEW: MCCUNE- ALBRIGHT SYNDROME (MAS)

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### ABSTRACT

A 17 year old female presented with the complaints of pain in right lower thigh and unable to weight bear for past 10 days, got admitted in a private hospital at Coimbatore, Tamil Nadu, India. She had the similar complaints to left thigh 5yrs back and treated for pathological fracture in local hospital. Her postoperative period was eventful. Fracture healing was appropriate but her unable to weight bear on affected limb. History of menstruation at 4yrs of age. While performing physical assessment, found that she had, Cafe-Au-Lait Spots over right side face and thigh. Normal vital signs noted. A provisional diagnosis of McCune- Albright syndrome ((MAS) and bone scan findings, Histo pathological report on femur tissue shows fibrous dysplasia. Mirel's criteria scored as 11/12 in right femur. Patient was deliberately treated for the pathological fracture surgically and physiotherapy measures were implemented.

### KEYWORDS

Albright syndrome, MAS and Bone maturity.

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### INTRODUCTION

McCune-Albright syndrome is a disorder that affects the bones, skin and several hormone-producing (endocrine) tissues. It is a rare genetic disorder, occurs among 1/100,000 people.

#### Definition

McCune Albright syndrome is a, rare mosaic disorder, classically defined by the clinical trial of fibrous dysplasia of bone (FD), *café-au-lait* skin spots and precocious puberty (PP).

#### Etiopathogenesis

- It is not inherited disease.

- It can be formed during early or late embryogenesis.
- It is caused by mutated GNAS gene affecting the alpha subunit of a chromosome.

Due to any cause - unknown, physical, genetic and environmental factors



Abnormal mutation of GNAS gene



Influencing the GTPase activity of G protein in the cell membrane



Continuous stimulation of adenylyl cyclase



Producing cyclic AMP –influence on target cells

### Clinical Manifestations

#### Polyostotic Fibrous Dysplasia

Abnormal scar like, fibrous tissue in the bones, which leads to pathological fractures, deformity and asymmetrical growth occurs in single or multiple skeletal sites affects one side of the body.

- If the fibrous tissue lesions occur in the face, it may cause asymmetrical facial contour.
- Asymmetry may occur in the long bones, cause limping.
- Abnormal curvature of the spine, scoliosis may occur
- The proximal femur is one of the most commonly involved sites and may develop a characteristic coxa vara (“shepherd’s crook”) deformity

#### Cafe-Au-Lait Spots

Cafe-au-lait macules are often the first clinically apparent manifestations of MAS, presenting at or shortly after birth. Light brown colour patches of the skin with jagged irregular borders, which may present from birth. Spots ranging from light brown to dark brown in colour, often displaying a segmental distribution and frequently predominating on one

side of the body without crossing the midline; appears on only one side of the body.

### Hormonal Abnormalities

#### Precocious Puberty

It is usually vaginal bleeding or spotting, accompanied by development of breast tissue, usually without the development of pubic hair. Precocious puberty may occur at the age of 2. It is not influence of gonadotrophins. In this, Estrogen level is increasing due to the lesion in the ovaries. Less commonly, in boys, it can be bilateral, testicular enlargement with penile enlargement, scrotal rugae, body odour, pubic and axillary hair and precocious sexual behavior.

### Other Endocrine Problems

#### Thyroid gland

Thyroid gland may become enlarged or develop masses called nodules, characterized by macrocephaly, vision loss and hearing loss.

#### Pituitary gland

Excessive growth hormone may result in acromegaly, characterized by large hands and feet, arthritis and asymmetrical facial features.

#### Adrenal gland

Rarely, Cushing syndrome, an excess of the hormone cortisol may occur. In McCune, Cushing syndrome occurs only before age 2.

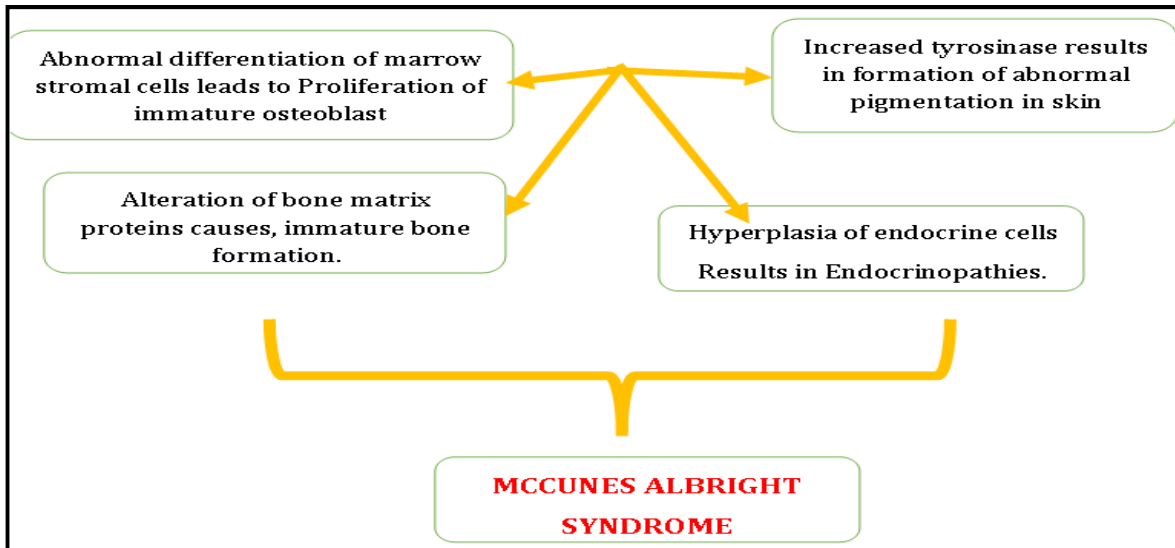
### Non-cancerous gastro intestinal growths called polyps may occur

Less common features of MAS may include

- Hepatitis
- Gastroesophageal reflux
- Pancreatic complications such as pancreatitis
- Intramuscular myxomas (benign tumors)
- Cancers

### Treatment Options

There is no specific treatment for MAS. Specific pharmacological agents will be prescribed for endocrinal abnormalities. For benign tumors in MAS will be treated with surgical interventions. Bisphosphonates are frequently used to treat fibrous dysplasia. Strengthening exercises are recommended to help maintain strength around the bones and minimize the risk of fractures.



## CONCLUSION

McCune-Albright syndrome is a disorder that affects the bones, skin and several hormone producing (endocrine) tissues. It is a rare genetic disorder, patients should receive close follow up with measurement of bone maturity and hormone levels.

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## CONFLICT OF INTEREST

We declare that we have no conflict of interest.

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