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Case Report

McCune-Albright Syndrome: A Rare Case

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Abstract: Pubertal changes before the age of 8 years are regarded as precocious puberty. McCune Albright Syndrome is a genetic disorder and accounts for 5% of female precocity. It consists of multiple, disseminated, cystic bone lesions, café au lait skin spots and sexual precocity. Sexual precocity is due to autonomous early production of estrogen by the ovary. We present a case of McCune Albright Syndrome in a 4 years old girl presented with premature thelarche.
Keywords: Precocious puberty, McCune Albright Syndrome, café au lait spots

INTRODUCTION

Pubertal changes before the age of 8 years are regarded as precocious puberty. In peripheral or incomplete precocity, also known as precocious pseudopuberty, there is no stimulation from hypothalamopituitary gonadotropin. It may be due to Adrenal Feminizing or masculinizing tumors, ovarian cysts or tumors or McCune Albright Syndrome. McCune Albright Syndrome is a genetic disorder and accounts for 5% of female precocity. It consists of multiple, disseminated, cystic bone lesions, café au lait skin spots and sexual precocity. Sexual precocity is due to autonomous early production of estrogen by the ovary. Here, we are presenting a case of McCune Albright Syndrome in a 4 years old girl who presented with premature thelarche.

CASE REPORT

A 4 year old Hindu, female child presented with complaints of gradual enlargement of the breasts and external genitalia since 6 months. In addition, she had white discharge but no bleeding per vaginum since six months. There was no history of fever, convulsions, headache, visual disturbances or sexual assault.

The girl was the firstborn of a full term, vaginal twin delivery, with immediate cry, born of a non-consanguineous marriage. Her twin sister and a 10 year old male sibling were normal and healthy. Her mother had attained menarche at the age of 13 years.

On examination: The child weighed 15.5 Kg (normal-18+/-3.6 kgms). Her height was 106 cms (normal-104+/-12.5 cms). There were brown, pigmented non pruritic macular lesions on the neck, chest, abdomen and back. There were no papules or vesicles. There was no evidence of acne or hirsutism. There was no goitre, no lymphadenopathy or organomegaly. Cardivascular and respiratory system were normal. There was no neurological deficit. Opthalmological examination including perimetry and fundoscopy were normal.

Axillary and pubic hair had not yet developed. Bilateral breasts were developed (SMR stage 2), there was no lump or discharge from them (Fig-1). Labia minora and majora were enlarged. There was discharge from the vagina which was white in colour with no foul odour.

On investigations: Hb, TLC, DLC, ESR, urine microscopy were normal. Vaginal discharge was normal on microscopy and culture. Liver and renal function tests and T3, T4 TSH were normal. Serum FSH, LH and Prolactin were normal for the age. Estradiol levels were 77.80 pg/ml.

X-Ray wrist showed presence of epiphyseal centres of 7 carpal bones (consistent with bone age-6 years-advanced) (Fig. 2). X-Ray Skull lateral view showed normal sellaturcica. X-Ray Femur –there was multiple, disseminated, cystic bone lesions.USG whole abdomen was normal. USG Pelvis showed uterus size enlarged to 31x25x30mm, both ovaries was normal.

CT Scan of head showed thickening of bones of base of skull with ground glass increase in bone density with obliteration of foraminae of skull (except foramen ovale) & reduction of size of maxillary & ethamoid sinuses, with thickening of periorbital region bones &

skull vault with normal sella region & normal brain parenchyma. Findings were suggestive of fibrous dysplasia of skull bone and periorbital region, normal parenchyma.

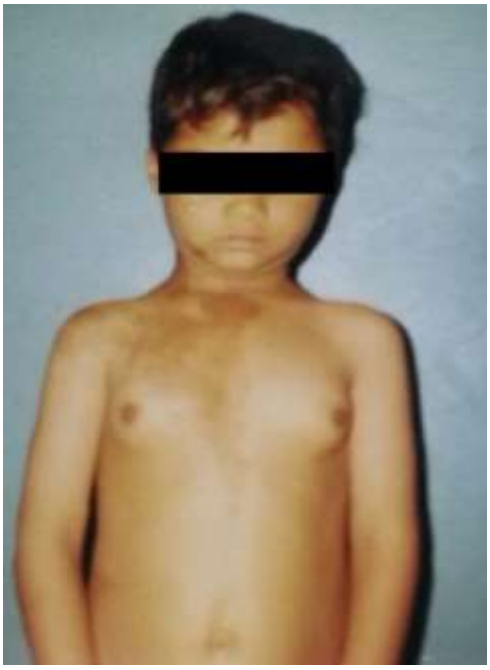

Fig.1: Precocious Puberty with café au lait spots

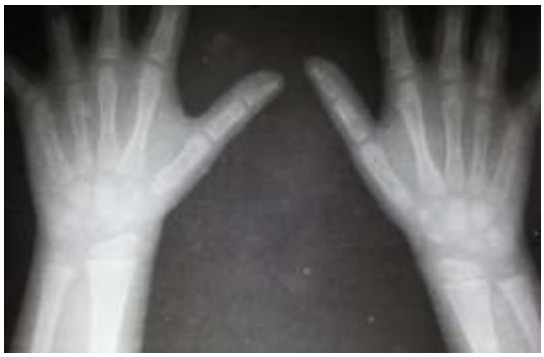

Fig. 2: X-ray of the wrists

DISCUSSION

The classical triad of McCune-Albright syndrome (MAS) consists of polyostotic fibrous dysplasia (FD), skin hyperpigmentation (café-au-lait spots), and endocrine dysfunction, frequently seen in females as precocious puberty [1]. Bone lesions may be solitary (monostotic) or multiple (polyostotic). MAS is a rare disease (the estimated prevalence ranges between 1/100,000 and 1/1,000,000).The skeletal aspect of the disease, Fibrous dysplasia, especially monostotic disease, is not rare [2].

Clinical picture in MAS is related to its mosaic nature, ranging from one or two mild clinical signs with excellent long-term prognosis to a severe life-threatening multiorgan disease [1]. MAS is most commonly confused with neurofibromatosis (NF). The location and shape of the spots usually can help to distinguish between the MAS and NF. The spots in MAS have jagged borders (coast of Maine), whereas those in NF are smooth (coast of California). Café-au-lait pigmentation in case of McCune-Albright syndrome does not cross midline. Frequent locations are the nape of the neck and the crease at the apex of the buttocks. Bisphosphonates are frequently used in the treatment of FD. Strengthening exercises are recommended to help maintaining the musculature around the FD bone and minimize the risk for fracture. Malignancies associated with MAS are distinctly rare occurrences. Malignant transformation of FD lesions occurs in probably less than 1% of the cases of MAS [2].

Other endocrinopathies, including hyperthyroidism, growth hormone excess, renal phosphate wasting with or without rickets/osteomalacia and Cushing syndrome may be found in association with the original triad. Rarely, other organ systems may be involved (liver, cardiac, parathyroid, pancreas). Treatment of all endocrinopathies is required. Ultrasonography is very useful in ovarian cyst follow-up and in the detection of thyroid and adrenal nodules [3].

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