Editorial

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McCune-Albright Syndrome

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Editorial

McCune-Albright syndrome (MAS) is a rare disorder attributed to a somatic mutation of the gene encoding the guanine nucleotide binding protein, alpha stimulating (GNAS) gene [1]. It was first described in 1937 by McCune and Bruch, and Albright and associates separately, as a rare syndrome characterised by the clinical triad of café-au-lait skin pigmentation, polyostotic fibrous dysplasia and Gonadotropin-independent sex steroid production resulting in precocious puberty [2-4]. The diagnostic gold standard is a specific form of polymerase chain reaction (PCR) that can detect activating mutations of *GNAS1* gene in peripheral blood cells, however, it is only recommended when the diagnosis is uncertain, as in isolated-monostotic lesions in the skull [1,5]. Additionally, MAS has been associated with multiple endocrine alterations like precocious puberty, thyrotoxicosis, Cushing syndrome, growth hormone excess, and non-endocrine disorders such as cardiac arrhythmias, cholestasis, hepatitis, and intestinal polyps [4,5]. All patients with suspected MAS should be screened to have fibrous dysplasia. Therefore, long bones and skull base radiographs should be performed to rule out the characteristic findings described as lytic lesions in the metaphysis or diaphysis with a "ground glass" matrix, thinning of the cortical bone, and endosteal scalloping without periosteal reaction. The most commonly affected sites include proximal femur, tibia, ribs and skull [6]. Also, it is important to remember that in peripheral precocious puberty -like occurs in MAS, the sequence of pubertal progression may be abnormal, e.g., with vaginal bleeding previous to breast development [7]. On the other hand, in boys with MAS is unusual to present sexual precocity and there is a high prevalence of ultrasonographic testicular pathology such as focal lesions representing Leydig cell hyperplasia and microlithiasis, so a further radiological screening with sequential testicular ultrasound should be performed [7,8]. Summarly, we recommend considering MAS in every child with early development of secondary sexual characteristics -before 8 years in girls and 9 years in boys- and café-au-lait skin pigmentation.

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