

LETTER TO THE EDITOR

A Suspected Case and Literature Review of McCune-Albright Syndrome

Kyung Eun Jung, Ji Hae Lee¹, Tae Yoon Kim¹

Department of Dermatology, Eulji University Hospital, Daejeon,

¹Department of Dermatology, The Catholic University of Korea, Seoul St. Mary's Hospital, Seoul, Korea

Dear Editor:

The McCune-Albright syndrome (MAS) is typically characterized by fibrous dysplasia (FD), endocrine abnormalities, and café au lait macules (CALMs). However, in some cases, MAS consists of a wide spectrum of symptoms.

A 15-year-old boy presented with an asymptomatic, brownish patch on the right cheek with facial asymmetry that had developed 100 days ago. On physical examination, a solitary, brown patch with an irregular border was seen covering the patient's right cheek and upper eyelid (Fig. 1A). He was otherwise healthy with no neurologic or visual defects. He had no significant medical or family history. Computed tomography (CT) scans of the brain showed polyostotic FD (Fig. 1B). The histopathologic features of the samples taken from the cheek and maxilla bone are shown in Fig. 2. On the basis of the findings, the boy was suspected of having MAS, accompanied with CALMs and polyostotic FD.

MAS is classically defined by the clinical triad of FD, CALMs, and precocious puberty (PP). It was later found that other endocrinopathies, namely hyperthyroidism, excess growth hormone (GH) production, renal phosphate wasting, and Cushing's syndrome, occasionally develop along with the original triad¹. Currently, a combination of

any of these findings is referred to as MAS². The involvement of a broad spectrum of tissues and the combined unpredictable manifestations of this disease are attributed to the molecular defects due to dominant mutations in the widely expressed signaling protein, $Gs\alpha$, and to the fact that these mutations arise sporadically, often early in development, prior to gastrulation, and are distributed across many or few tissues².

FD is a genetic, non-inheritable disease, characterized by bone pain, bone deformity, and fracture. The histological hallmark of the disease is the accumulation of fibrous tissue, containing immature spindle fibroblast-like cells, within the bone marrow and expanding from the medullary cavity to the cortical bone³.

Although a solitary, small CALM is common and con-

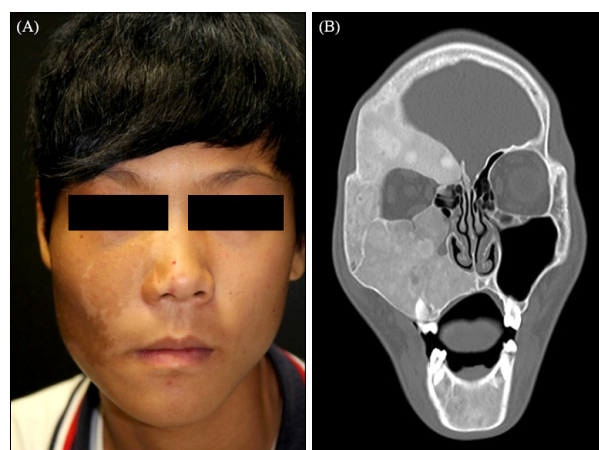


Fig. 1. (A) Solitary, brown patch with irregular border, throughout the patient's right cheek and upper eyelid with facial asymmetry. (B) Coronal computed tomography scan demonstrates heterogeneous ground glass appearance at the right anterior skull, consistent with polyostotic fibrous dysplasia of the skull, right maxilla and mandible and narrowing of superior and inferior orbital canal, and compressing the right optic nerve.

Received July 26, 2013, Revised September 11, 2013, Accepted for publication September 26, 2013

Corresponding author: Tae Yoon Kim, Department of Dermatology, The Catholic University of Korea, Seoul St. Mary's Hospital, 222 Banpo-daero, Seocho-gu, Seoul 137-701, Korea. Tel: 82-2-593-2626, Fax: 82-2-3482-8261, E-mail: tykimder@catholic.ac.kr

This is an Open Access article distributed under the terms of the Creative Commons Attribution Non-Commercial License (<http://creativecommons.org/licenses/by-nc/3.0>) which permits unrestricted non-commercial use, distribution, and reproduction in any medium, provided the original work is properly cited.

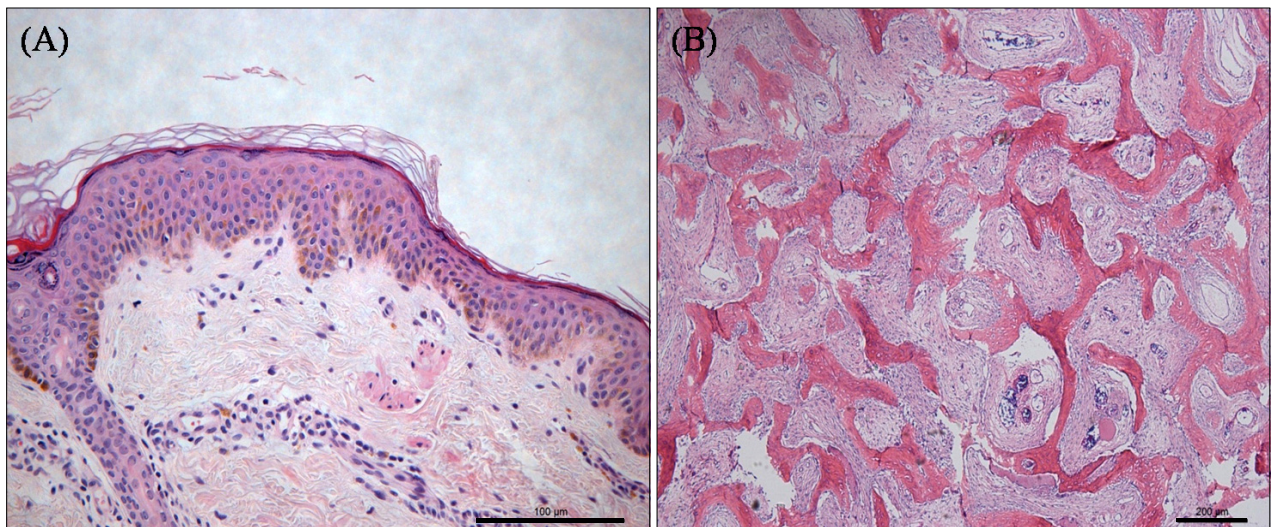


Fig. 2. (A) Hyperkeratosis, epidermal acanthosis, basal layer hyperpigmentation, and perivascular inflammatory infiltrates in the dermis (H&E, $\times 100$). (B) Curvilinear trabeculae of woven bone admixed with fibrous tissue without nuclear atypia and mitoses (H&E, $\times 40$).

sidered a normal finding, some CALMs are considered a diagnostic criterion or a characteristic trait of various syndromes, such as neurofibromatosis, tuberous sclerosis, Turner and Noonan syndromes as well as MAS⁴. The CALMs seen in MAS patients are typically the first manifestations of the disease, usually appearing either at or shortly after birth. Therefore, this manifestation could be used as a clue to the early diagnosis of MAS.

The symptoms associated with endocrinopathies of hyperfunction of the endocrine glands are PP, hyperthyroidism, excess production of GH, etc. Regarding PP, our patient showed no evidence of PP on physical examination; PP is very rare in boys affected with MAS. The timely diagnosis of hyperthyroidism in MAS patients is important to avoid exacerbation of osteoporosis. Non-suppressible serum GH secretion during oral glucose tolerance test (OGTT) is another characteristic feature of MAS. However, in subtle

disease, the results of the OGTT can be equivocal, especially in the cases of young children; therefore, more detailed examination is required in such cases.

REFERENCES

1. Dumitrescu CE, Collins MT. McCune-Albright syndrome. *Orphanet J Rare Dis* 2008;3:12.
2. Collins MT, Singer FR, Eugster E. McCune-Albright syndrome and the extraskeletal manifestations of fibrous dysplasia. *Orphanet J Rare Dis* 2012;7(Suppl 1):S4.
3. Chapurlat RD, Orcel P. Fibrous dysplasia of bone and McCune-Albright syndrome. *Best Pract Res Clin Rheumatol* 2008;22:55-69.
4. Landau M, Krafchik BR. The diagnostic value of café-au-lait macules. *J Am Acad Dermatol* 1999;40:877-890; quiz 891-892.