

Acquired oral mucosal pigmentation in a patient with McCune-Albright syndrome: A case report.

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ABSTRACT :

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McCune-Albright syndrome is a very rare disease characterized by the triad of polyostotic fibrous dysplasia, café-au-lait macules, and at least one of hyperfunctioning endocrinopathies. We present a case of a 19-year-old female with McCune-Albright syndrome who presented with acquired asymptomatic multiple hyperpigmented macules on the lips and the oral mucosae without acral and anogenital pigmentation, which are not a common finding of this syndrome.

Key words: McCune-Albright syndrome, oral mucosal pigmentation, gastrointestinal polyps, Peutz-Jeghers syndrome

บทคัดย่อ :

คงศักดิ์ สุนทรภา ประวีตร อัศวานนท์ รายงานการเกิดผื่นราบสีน้ำตาลบริเวณเยื่อปากที่เกิดขึ้นภายหลังในผู้ป่วยแมคคูน อัลไบรท์ ซินโดรม วารสารโรคผิวหนัง 2555; 28: 150-153.

สาขาวิชาตจวิทยา ภาควิชาอายุรศาสตร์ คณะแพทยศาสตร์ จุฬาลงกรณ์มหาวิทยาลัย

แมคคูน อัลไบรท์ ซินโดรม (McCune-Albright syndrome) เป็นกลุ่มโรคที่พบบได้น้อยมาก มีลักษณะสำคัญ 3 ประการ ได้แก่ ความผิดปกติของกระดูกเป็นโพลิออสโตติก ไฟบรัส ดิสเพลเซีย (polyostotic fibrous dysplasia), มีปานดำ (café-au-lait macules) และมีความผิดปกติอย่างใดอย่างหนึ่งที่เป็นการทำงานที่เพิ่มขึ้นผิดปกติของระบบต่อมไร้ท่อ (hyperfunctioning endocrinopathies) รายงานฉบับนี้นำเสนอผู้ป่วยหญิงอายุ 19 ปีที่เป็นแมคคูน อัลไบรท์ ซินโดรม และมีผื่นราบสีเข้มขึ้นบริเวณริมฝีปาก และในเชื่อกุช่องปากในระยะต่อมา ไม่พบมีผื่นลักษณะเดียวกันนี้บริเวณมือและเท้า บริเวณรอบทวารหนักและอวัยวะเพศ ซึ่งลักษณะผื่นดังกล่าวนี้ไม่ได้เป็นผื่นที่พบบ่อยนักในแมคคูน อัลไบรท์ ซินโดรม

คำสำคัญ : แมคคูน อัลไบรท์ ซินโดรม, ผื่นราบสีเข้มบริเวณริมฝีปาก, เนื้องอกในลำไส้, พอยซ์-เจเกอร์ ซินโดรม

Introduction

McCune-Albright syndrome (MAS) is a rare disease with prevalence of 1/100,000 – 1/1,000,000.¹ The syndrome results from postzygotic activating mutations of GNAS1 gene. It is characterized by the triad of polyostotic fibrous dysplasia, café-au-lait macules following lines of Blaschko (more specifically with irregular border and located on the same side with bone lesions), and endocrine hyperfunction.¹⁻³

In this report, we present a patient with MAS which has all of the typical features except that pigmented lesions developed later in life on her lips and oral mucosae mimicking those found in other syndromes, especially Peutz-Jeghers.

Case report

A 19-year-old female from Bangkok presented with asymptomatic multiple hyperpigmented macules on the lips and the oral mucosae which have been progressive for the last 5 years. These lesions were first noted on the tongue and then spreaded to other parts of the mouth. She had no acral or anogenital involvement and denied previous implantation of dental amalgam or dental

braces. Furthermore, she had no history of intussusception or gastrointestinal bleeding. None of her family members was affected with a similar disorder.

The patient was diagnosed with MAS since her childhood by following presentations: early menarche at the age of 3, treated by hormonal suppression; a café-au-lait macule on the right buttock; and multiple episodes of pathologic right femoral fractures which was the same side as the café-au-lait macule.

The skin examination reveals multiple and diffuse discrete bluish-grey hyperpigmented macules on the lower lips, gingivae, and the buccal mucosae. (figure 1-3) There is also a solitary hyperpigmented patch, sized 3x5 cm. with an irregular border on the right buttock. (figure 4)

We treated these oral lesions with Q-switched 532 nm Nd : YAG laser (Revlite[®], HOYA Conbio, Fremont, California), 3mm spot size with the fluence in the range of 1.5-1.8 J/cm². The lesions faded after 3 sessions.

**Figure 1****Figure 2****Figure 3**

Figure 1-3 Clinical photographs reveal multiple and diffuse discrete hyperpigmented macules on the lower lips, gingivae, and both buccal mucosae.



Figure 4 : A hyperpigmented patch with an irregular border on the right buttock, and also a few linear surgical scars were noted.

Discussion:

Orificial pigmented lesions are more commonly found in other syndromes, such as Peutz-Jeghers (PJS)^{3,4}, Laugier-Hunziker⁵, and Carney complex.³ In PJS, the well-established association is gastrointestinal polyposis. However, in MAS, there are just a few reports which document the findings of oral melanotic pigmentation.^{6,7} None of them was investigated for the gastrointestinal polyps. Recently, Zacharin et al.⁴ performed an observational study in four patients with confirmed MAS by DNA sequencing. Two patients had perioral pigmentary lesions, and the other two did not. Three of them were documented to possess GNAS mutations in the peripheral blood. Also, genetic testings for STK11 and PRKAR1A were done to exclude coexistent PJS, and Carney complex. They all

underwent gastrointestinal endoscopy, which showed hamartomatous gastrointestinal polyps with histological features similar to those in PJS in all patients. Zacharin et al. concluded that gastrointestinal polyps might also be one of manifestations of MAS, and indicated an overlap between the two entities. Therefore, we believe that the presence of oral hyperpigmented macules in this patient may be the manifestation of MAS, and she might have a risk of gastrointestinal polyps. Nonetheless, her parents did not give consent for a gastrointestinal endoscopic examination to confirm the presence of such polyps.

In conclusion, we report a case of McCune-Albright syndrome who presented with oral mucosal

pigmentation in her adolescent years which is a rare manifestation in this syndrome, and may be associated with the risk for gastrointestinal polyposis.

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