CASE REPORT

Café au lait spot: Case report

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Abstract

Café au lait spots (CALS) are pigmented lesions found in various genetic disorders. They also occur in normal populations. Identifying and having knowledge about CALS may help in early detection of associated diseases and genetic disorders, thereby facilitating the prevention of their late manifestations. The article reviews CALS with an example of a case that occurred in an adult patient.

Keywords

Café au lait spots, macule, neurofibromatosis, pigmentation

Introduction

Café au lait spots (CALS) or café au lait macules are birthmarks that are pigmented.[1] The word café au lait which has its origin from French, literally means “coffee with milk” because of their light-brown color. If these pigmentations have oval and smooth borders they are called as “coast of Maine spots,” alternatively if they exhibit jagged borders they are referred to as “coast of California spots.” Clinically they are evidenced by well-circumscribed, regularly pigmented macules or patches that have a diameter ranging from 1-2 mm to >20 cm in longest diameter. Smooth borders are typical of neurofibromatosis-1 (NF1) and jagged borders may point out at McCune-Albright syndrome. Histologically CALS shows increased melanin content of melanocytes and basal keratinocytes.[2]

Case Report

A 35-year-old patient with diagnosed NF1 presented with multiple CALS and neurofibromas throughout the body [Figure 1]. No history of seizures or mental retardation. The lesions were around 1 cm × 2 cm [Figure 2]. No skeletal and neurological abnormalities were present. Macules were flat with the color of brownish-white. They exhibited well-defined borders, and color was uniform throughout the lesion.

Discussion

Café au lait pigmentations are observed clinically in genetic diseases like NF1 Type I, McCune-Albright syndrome and Noonan’s syndrome. They typically are of tan or brown color with irregularly shaped macules of different sizes. They may occur in any region of the skin.[3] 85% of neurofibromas present with CALS. In general, CALS are flat, discrete, and light-brown in color, with clearly defined margins. The color of margins resembles that of the immediate adjacent skin. CALS usually exhibit borders with regular outline, though greater sized CALS may have irregular borders. Typical lesions characteristically are of 2-5 cm diameter, although it can be as high as 15 cm in patients with neurofibromatosis.[4]

Solitary CALS are common in infants and children, but multiple such spots are very rare, and 6 or more spots >0.5 cm may be typical of Type 1 NF1.[5] Diagnostic criteria for NF1 other than CALS includes freckling of skin folds, Lisch nodules, neurofibromas, skeletal dysplasia, optic glioma and affected first degree relative with NF1.[6] Reaching an early final diagnosis of NF1, which is the most common disorder with multiple CALS is crucial. Patients especially children have high risk of developing malignant diseases such as malignant schwannoma, myeloid disorders, rhabdomyosarcoma and Wilms’ tumor. Significant amount of cases with CALS are within 2 cm in diameter. CALS is more prevalent on unexposed
skin and are found on the trunk, buttocks, and lower limbs. Head, neck, and upper extremities are usually spared. Multiple smaller “freckle-like” CALS in the axillary or inguinal region if found are known as Crowe’s sign. Multiple small CALS in the perioral region, Torok’s sign, are observed less frequently. Differential diagnosis includes McCune-Albright syndrome, Silver-Russell dwarfism, Watson’s syndrome, and multiple lentigines syndrome.[7]

Crowe’s sign and Lisch nodules are highly characteristic of NF1 Type I. Lisch nodules are nothing, but pigmented lesions of the iris. Noonan’s syndrome and allelic LEOPARD syndrome, which are autosomal dominant disorders, also show pigmented mucocutaneous macules. The classic appearing CALS are characterized seen in Noonan’s phenotype. LEOPARD phenotype is typically associated with numerous small freckles like macules often involving the skin of the face.[3] Therapy are not usually indicated. However, a variety of options are available, including laser surgery.[7]

The significance of CALS, especially in children should be well appreciated. A normal healthy child below the age of five rarely exhibits multiple CALS. Hence, presence of such multiple spots may strongly hint the presence of an associated/underlying neuroectodermal disorder, for example-NF1. A diagnosis of neuroectodermal dysplasia should be confidently formulated if there are more than five such spots. In the first 5-6 years of a child, skeletal and visceral manifestations of NF1 are occasional before the appearance of classical tumors. Hence one can vehemently conclude that observance of occasional pigmentation may be the only clue to accurate diagnosis and appropriate treatment in such child patient with no other obvious manifestation of this disease.[6,9]

Johnson et al. reported more 3,4-dihydroxyphenylalanine (DOPA) positive melanocytes in CALS of NF1 patients than the surrounding skin. Also, these melanocytes contained giant pigmented granules. CALS in normal patients showed fewer DOPA positive melanocytes than the surrounding skin. But these melanocytes did not contain giant pigmented granules. They concluded that presence of giant granules and increased melanocytes in CALS is helpful in the diagnosis of NF1 when pigmented macules are the only manifestation.[10]

Conclusion

Every medical, dental and health practitioners should be thoroughly aware of CALS and be able to diagnose this pigmented lesion and the diseases associated with them. This will have a great impact on prognosis and prevention of the development of complications of lesions/diseases associated with CALS.

References
