Giant Café au Lait Spot in a Patient with Neurofibromatosis

Sir,

Neurofibromatosis is an autosomal dominant disorder with characteristic skin lesions, such as café au lait spots, axillary freckling and neurofibromas, and other congenital and hamartomatous lesions of the bone, endocrine glands and central nervous system (1, 2).

We describe here a patient with neurofibromatosis 1 (NF1) with a striking clinical appearance due to a massive café au lait spot.

CASE REPORT

A 24-year-old female presented with a brown patch on her back which had been present since birth. She was in good health and there was no history of mental retardation, learning disabilities or seizures. Physical examination revealed a 31×38 cm flat brown macula covering a great portion of her back, extending up to the nape of her neck and down to her waist (Fig. 1). The lesion had a uniform depth of colour, clearly defined margins, slightly irregular borders with skin markings the same as adjacent skin. A biopsy specimen taken from the lesion revealed findings consistent with café au lait spot. Numerous café au lait spots, 11 of which had a diameter greater than 1.5 cm, and neurofibromas were present on her trunk, upper arms and thighs (Fig. 2). Bilateral axillary freckling was present. Ophthalmological examination revealed no Lisch nodules. No skeletal or neurological abnormalities were noted and magnetic resonance imaging of the brain was normal. Her family history was negative for neurofibromatosis.

DISCUSSION

Neurofibromatosis is a relatively common autosomal dominant disorder with a high rate of spontaneous mutation. It is estimated to occur with a frequency of about 1 in 2500 – 3000 live births with no significant difference with respect to sex, race or colour (1, 2). Riccardi (3) has classified neurofibromatosis into 8 types due to its considerable variation in clinical presentation. NF1, the classic or the peripheral form of neurofibromatosis, accounts for about 85% of cases and is marked by the more classic features of the syndrome: multiple café au lait spots; axillary freckling; cutaneous neurofibromas; and Lisch nodules (1 – 6).

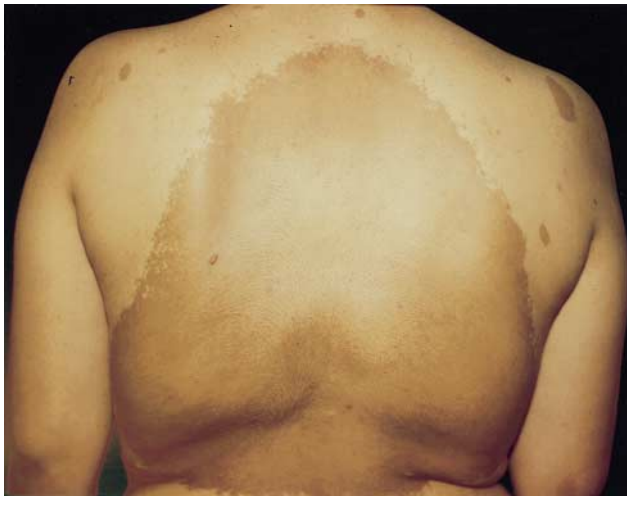

Fig. 1. A 31×38 cm café au lait macula covering a great portion of the patient's back.

Fig. 2. Numerous café au lait spots and neurofibromas on the patient's trunk, upper arms and thighs.

Typical café au lait spots are discrete, flat, light-brown in colour, with clearly defined margins, and skin margins the same as adjacent skin. The border must have a regular outline, but the larger café au lait spots may also have irregular borders. The lesions characteristically average 2 – 5 cm in diameter, although café au lait spots as large as 15 cm have been noted in patients with neurofibromatosis (2, 7 – 9). In our patient, the café au lait macula, being 31×38 cm in diameter, has a size that is rarely seen.

REFERENCES

1. Paole S, Fenske NA. Cutaneous markers of internal malignancy. I. Malignant involvement of the skin and the genodermatoses. J Am Acad Dermatol 1993; 28: 1 – 13.
2. Hurwitz S. Clinical pediatric dermatology. A textbook of skin disorders of childhood and adolescence. 2nd edn. Philadelphia: WB Saunders Co., 1993: 624 – 629.
3. Riccardi VM. Neurofibromatosis: clinical heterogeneity. Curr Probl Cancer 1982; 7: 1 – 34.
4. Niimura M. Aspects in neurofibromatosis from the viewpoint of dermatology. J Dermatol 1992; 19: 868 – 872.
5. National institutes of Health Consensus Development Conference (1988) Neurofibromatosis conference statement. Arch Neurol 1988; 45: 575 – 578.
6. Cohen PR. Genodermatoses with malignant potential. Am Fam Physician 1992; 46: 1479 – 1486.
7. Whitehouse D. Diagnostic value of the café au lait spot in children. Arch Dis Child 1966; 41: 316 – 319.
8. Burwell RG, James NJ, Johnston DI. Café au lait spots in schoolchildren. Arch Dis Child 1982; 57: 631 – 632.
9. Alper J, Holmes LB, Mihm MC. Birthmarks with serious medical significance: nevocellular nevi, sebaceous nevi, and multiple café au lait spots. J Pediatr 1979; 95: 696 – 700.

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