Congenital, Segmental Pigmented Lesions

Jacklyn S. Stotts, MD, Howard K. Steinman, MD
University of California, San Diego, School of Medicine

REPORT OF A CASE

A 64-year-old woman was evaluated at the University of California, San Diego, Medical Center for a lifelong history of pigmented macules involving her left arm, the left side of her chest, and the left side of her back and a 40-year history of multiple soft, asymptomatic, pigmented cutaneous papules involving the same locations. She had hypertension, which had been controlled with diuretic therapy, but she had no history of headaches, hearing loss, speech problems, or seizures. There was a family history of facial "freckling," but no family members had similar skin lesions.

Physical examination revealed multiple 2- to 10-mm hyperpigmented macules distributed over the left side of the upper part of her back (Fig 1), the left side of her chest, her left breast, the medial aspect of her left arm and forearm, and her left axilla (Fig 2). In the same distribution there were multiple 3- to 8-mm hyperpigmented, soft, pedunculated nodules, which "buttonholed" on palpation (Fig 3). Results of ophthalmologic and neurologic examinations were normal. A skin biopsy was performed on a representative nodule (Fig 4).

What is your diagnosis?

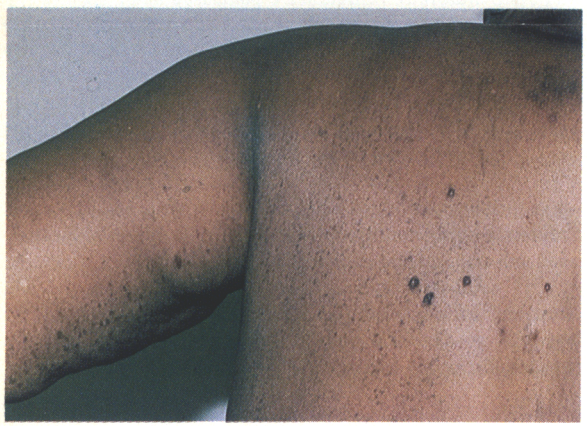

Figure 1.

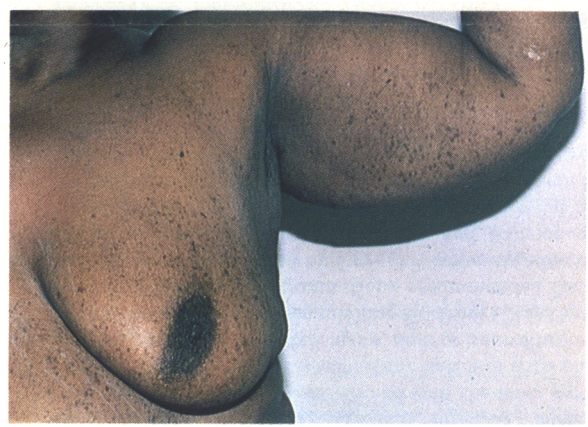

Figure 2.

Figure 3.

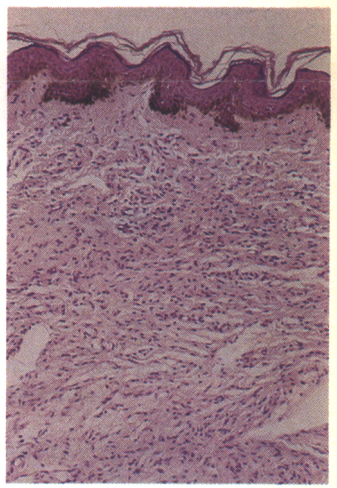

Figure 4.

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DIAGNOSIS: Segmental neurofibromatosis (SN).

DISCUSSION

Classic neurofibromatosis (CN), or von Recklinghausen's disease, is defined clinically by the presence of multiple cafe-au-lait macules, often with axillary freckling, multiple neurofibromas, and pigmented iris hamartomas (Lisch nodules). Associated features include central nervous system tumors, neurofibrosarcomas, malignant schwannomas, several forms of leukemias, and endocrine and musculoskeletal abnormalities.[1]

Our patient manifested the clinical and histologic features of SN. Segmental neurofibromatosis is a unique form of neurofibromatosis characterized by cutaneous neurofibromas and/or cafe-au-lait macules limited to a circumscribed body segment. This is a rare disorder, with only 13 cases reported (to our knowledge) in the world literature.[2-8] All reports have described a segmental anatomic distribution of neurofibromas, schwannomas, plexiform neurofibromas, and/or cafe-au-lait macules histopathologically identical to those found in CN. One case[7] also manifested iris hamartomas (Lisch nodules) on the same side as the cutaneous involvement. In addition, there has been one reported case described as bilateral dermatomal neurofibromatosis.[5] It has been suggested, however,[9] that this latter case did not represent SN and that the term *segmental neurofibromatosis* be reserved for patients with unilateral, segmental cafe-au-lait spots, and/or neurofibromas.

The importance of distinguishing SN from CN is that the extensive laboratory examinations recommended for CN, such as cranial computed tomographic scanning, electroencephalograms, skull roentgenograms, and 24-hour urine collections for catecholamines, are not necessary in SN.[5,10] To our knowledge, in no reported case of SN has there been the development of a malignant schwannoma, pheochromocytoma, or neurofibrosarcoma. Moreover, none of the patients have been neurologically affected.

While the inheritance of CN is autosomal dominant, SN is apparently not genetically transmissible. In only one case of the segmental form has there been a positive family history of neurofibromatosis. Uhlin[4] described a case of SN in which a maternal aunt, her son, and the child of her son had generalized neurofibromatosis, though the patient's parents and siblings were not affected. This most likely represented a spontaneous mutation in the maternal aunt.

The origin of SN is still unclear. Crowe et al[2] have suggested that it may be explained by a somatic mutation in early embryonic development that results in a limited pattern of involvement.

References

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