CASE REPORT

Leprosy with neurofibromatosis – a diagnostic dilemma

CHANDER GROVER, MANMOHAN LOHRA, SONI NANDA & B.S.N. REDDY
Department of Dermatology, Venereology and Leprology, Maulana Azad Medical College and Associated Lok Nayak Hospital, New Delhi, India

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Summary The coexistence of leprosy with neurofibromatosis is a rare finding and can pose a diagnostic dilemma. Neurofibromatosis coexisting with borderline tuberculoid leprosy has previously not been reported. We report such a case in a 13-year-old boy where biopsy of clinically uninvolved nerve revealed the presence of acid-fast bacilli. A careful diagnostic workup is needed in such cases to ensure proper treatment. Both disorders affect Schwann cells and their relationship merits further consideration.

Introduction

Involvement of nerve and skin occurs in leprosy and neurofibromatosis, with the Schwann cell being the primary target for both. However, the aetiology and pathophysiology of both these diseases is different, with leprosy being an infection with Mycobacterium leprae and neurofibromatosis being a genodermatosis. Neurofibromatosis has earlier been reported to coexist with lepromatous, histoid and pure neuritic types of leprosy, but not with borderline tuberculoid leprosy. We report the case of a boy affected by neurofibromatosis (type 1) and borderline tuberculoid leprosy.

Case report

A 13-year-old boy, resident of Bihar, presented with an asymptomatic, soft swelling in front of his right ankle present for the past 5 years, which had been gradually increasing in size. Over the past 3 years, similar, smaller, swellings had appeared over the trunk and arms. There was no history of seizure, deafness, visual complaints, or motor or sensory deficit. There was no family history of similar lesions or of leprosy. For the past 3 years, the patient had also
noticed a pale, numb patch progressively increasing in size over his right shin. He was developmentally normal with normal milestones.

On examination, there were 14 café-au-lait macules varying in size from 0.5 × 0.5 cm to 6 × 2 cm over abdomen, back, forearm, buttocks and legs. Axillary freckling was also present. There were no palmar freckles or Leish nodules. Multiple neurofibromas were present over the right ankle and trunk. There was a 5 × 2 cm oval, hypopigmented patch over the right shin with well-to-ill-defined margins and an atrophic, dry surface with loss of pain sensation and of skin appendages. There was a single enlarged nerve to the patch, but other peripheral nerves were not enlarged.

Slit skin smears showed no acid-fast bacilli in any of the lesions. Biopsy from a nodular lesion of neurofibromatosis revealed multiple spindle shaped cells with elongated nuclei, forming whorls with marked capillary proliferation and absence of acid-fast bacilli in Wade–Fite stained sections. Biopsy of the hypopigmented macule showed well-defined epithelioid cell granulomas with giant cells and lymphocytes in the upper dermis, encroaching onto the epidermis, with no acid-fast bacilli. Nerve conduction studies from clinically uninvolved nerves revealed delayed conduction in some. Biopsy of the left radial cutaneous nerve showed acid-fast bacilli on Wade–Fite staining with little inflammatory infiltrate.

The findings confirmed the diagnosis of neurofibromatosis-1 (Riccardi’s classification) and borderline tuberculoid leprosy. The patient was started on multibacillary treatment for leprosy and counselled for neurofibromatosis. The patient has been under regular follow-up for 9 months.

Discussion

Neurofibromatosis is one of the neurocutaneous syndromes, characterized by pigmented macules and tumors of skin and nerves. The majority of cases are inherited (autosomal dominant) and less than 10% arise from spontaneous mutations. The coexistence of neurofibromatosis with leprosy in the same patient is interesting. One observation has been that the onset of neurofibromatosis has preceded the onset of leprosy, both in our case and in previous cases. Ghosh et al. proposed that neurofibromatosis might predispose Schwann cells to invasion by *M. leprae*, but the pathogenetic relationship, if any, is far from clear as the association is rare.

Coexistence of these two disorders may pose a diagnostic dilemma. Nodules are a hallmark of both neurofibromatosis and leprosy. In a non-endemic area, lesions of leprosy have been mistaken for neurofibromatosis and appropriate treatment has been delayed. Conversely, in a population prone to leprosy, neurofibromatosis has been mistaken for leprosy. Hence, inappropriate treatment may be instituted. Chatterjee et al. proposed that demonstration of acid-fast bacilli (AFB) in a biopsy should be conclusive proof of the diagnosis of leprosy as opposed to neurofibromatosis, but this view was challenged by Johansen et al. The presence of *M. leprae* in neurofibromatosis had also been reported in other studies.

We report this case for its diagnostic and therapeutic implications. In addition, the possible relationship between these two seemingly unrelated disorders is worth exploration.
References