Lepromatous leprosy with coexistent neurofibromatosis type 1

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Abstract

Neurofibromatosis type 1 (NF 1) and leprosy are two different disease entities as the former is a genodermatosis while the latter is an infectious disease mostly confined to tropical countries. Interestingly, among their many clinical manifestations, both conditions share the common features of involvement of the skin and Schwann cells of the peripheral nerves. The presence of lepromatous leprosy in a patient with NF 1 is relatively rare and may pose certain diagnostic dilemmas. In this case report we describe manifestation of lepromatous leprosy in a 31 years old patient with pre existing NF 1.

Introduction

Etiology and pathophysiology of NF 1 and leprosy are entirely different. However both of these conditions have cutaneous and neural manifestations and Schwann cells seem to be the primary target for both. NF 1 has previously been reported to coexist with borderline tuberculoid[1], borderline lepromatous[2], lepromatous[3] and histoid leprosy[4]. Most of them are isolated case reports and share one common feature that neurofibromatosis preceeded the onset of leprosy. A pathogenetic relationship between two conditions has been suggested since abundance of perineural tissue containing Schwann cells in NF 1 patients might make them susceptible to leprosy.

Case report

A thirty one years old male was referred from a medical ward with painful swelling of both lower limbs for which he was treated unsuccessfully with a course of intravenous antibiotics. He had a history of multiple asymptomatic nodules on the limbs and trunk over 12 year duration. He had recently noted a progressive numbness of both upper and lower extremities. Other family members were not affected with a similar kind of disease.

Clinical examination revealed multiple cutaneous neurofibromas, a plexiform neurofibroma, more than 6 café au lait macules each more than 15 mm in diameter and axillary freckling suggesting the diagnosis of NF 1. Slit lamp examination of the eyes failed to show the presence of Lisch nodules. Interestingly, patient's ear lobes and facial skin were infiltrated with supracilliary madarosis (Figure 1). In addition to neurofibromas which were soft nodules with positive buttonhole sign, there were multiple firm infiltrated nodules without "buttonholing". Some of them were tender. He was febrile with active arthritis over large joints. Both hands and feet were swollen, warm and tender. Feet were deformed with trophic ulcers at pressure points. He had uniformly thickened symmetrical non tender ulnar and common peronial nerves. Further examination revealed glove and stocking type peripheral anesthesia.

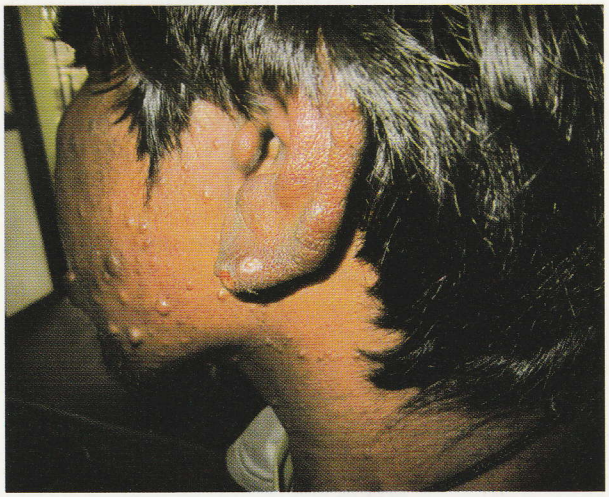

Figure 1.

Clinical diagnosis of lepromatous leprosy with type II lepra reaction in a patient with pre existing neurofibromatosis was made. Ear lobe smear was positive for acid fast bacilli with bacterial index of 6+. Morphological index showed 30% of bacilli in solid forms while granular and fragmented forms were 40% and 30% respectively. Biopsy from an infiltrated firm nodule showed a diffuse foamy

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histiocytic infiltration admixed with lymphocytes of the dermis with a Grenz zone, suggestive of lepromatous leprosy. Nerve conduction study showed severe axonal neuropathy of the main peripheral nerves. Other baseline investigations and imagings were normal.

Patient was started on multibacillary multi drug treatment for leprosy. Oral prednisolone 40 mg was started to control lepra reaction. Limb physiotherapy was started and special footwear were provided while trophic ulcers were managed with standard ulcer care. Family screening was negative for leprosy.

Discussion

NF is a common genodermatosis of autosomal dominant inheritance with the prevalence of 1 in 2500 - 3300 births[5]. Sporadic mutations account for up to 50% of all cases[5]. It is diagnosed clinically using a set of criteria which include the presence of several cutaneous manifestations (café-au-lait macules, neurofibromas, freckling etc), ocular manifestations, skeletal manifestations and positive family history. Its main clinical manifestations occur as a result of cutaneous and peripheral nervous system involvement.

Leprosy is a bacterial infection which is mostly confined to tropical countries where it is endemic. It has a wide variety of clinical manifestations, many of which are secondary to skin and peripheral nervous system involvement.

Both conditions, having entirely diverse pathogenetic mechanisms, target the Schwann cell although utilizing different methods. NF is associated with excess proliferation of perineural cells and Schwann cells whereas in leprosy, Schwann cells are the primary target for *mycobacterium leprae*[5]. Both conditions share same clinical features causing diagnostic dilemma while they can manifest in the same individual giving rise to the question whether one condition increases the susceptibility of the individual to the other.

Neurofibromata of the NF 1 can be mistaken for leprosy nodules and vice versa. Since the clinical features of NF 1 are much more prominent compared to the subtle changes of leprosy, the former can mask the presence of the latter. Hence a high degree of clinical suspicion is needed to identify the co-existence of leprosy among these individuals. Although this is a rare combination, it is important to identify every case of leprosy to achieve the target of eliminating leprosy. Both conditions can present with peripheral nerve thickening. In leprosy, thickened nerves are usually smooth and have cord like regular thickening unless a nerve abscess occurs as a part of type 1 lepra reaction. NF 1 usually presents with irregular asymmetrical nerve thickening. However Rao AG *et al* reported a patient with NF 1 presenting with generalized nerve thickening which could have been easily mistaken for leprosy[6]. To complicate the situation further there are case reports of NF 1 presenting with hypopigmented patches which closely mimic leprosy. Khandpur S *et al* reported a patient who fulfilled clinical criteria for NF 1, while having symmetrical peripheral nerve thickening and multiple hypopigmented patches at the same time[7].

Considering the striking similarity of the clinical features in both conditions, high degree of clinical suspicion is required. Ear lobe infiltration, supra-cilliary madarosis, diffuse cutaneous infiltration and nodules without buttonholing sign are some of the clinical features which may be helpful in identifying co existing leprosy in a patient with NF 1[3].

Finally we would like to consider the other aspect of this co existence where some authors have suggested that abundance of Schwann cells in NF 1 could make the patient more susceptible to leprosy[1]. However this concept is difficult to establish in the current context because of the rarity of this association.

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