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Case Report

A case of neurofibromatosis type1 with coexisting borderline tuberculoid leprosy

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Abstract Leprosy and neurofibromatosis are different entities. Both diseases present with nerve enlargement and skin lesions but the coexistence is rare. Nodules of neurofibromatosis may be mistaken for that of lepromatous leprosy and vice versa. Their simultaneous occurrence in same person may lead to diagnostic dilemma. We report a case which was at first mistaken as lepromatous leprosy. But proper history and examination aided by slit skin smear and skin biopsy for histopathological examination helped to reach the appropriate diagnosis of borderline tuberculoid leprosy associated with neurofibromatosis.

Key words
Leprosy, neurofibromatosis, Schwann cell.

Introduction

Leprosy is a tropical disease caused by *Mycobacterium leprae* that primarily affects the skin and peripheral nerve leading to significant disability. On the other hand, neurofibromatosis (NF) is a common genodermatosis of autosomal dominant variant with an incidence of 1 in 3000 births.[1] NF can be diagnosed clinically by the presence of café-au-lait macules, cutaneous and subcutaneous nodules, axillary freckling and peripheral nerve enlargement.

Though the etiology and pathogenesis are different, nerve involvement and skin lesions are the presenting features of both in leprosy and NF as Schwann cells are the primary target for both diseases.

We report a case of NF coexisting with borderline tuberculoid leprosy.

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Case report

A 26-year-old man was brought to local leprosy clinic for evaluation of skin patches with multiple skin nodules. The nodules were painless, skin colored, of varying size, present in trunk, extremities, forehead and submandibular area since his 19-year age. Over the past four months he noticed few hypopigmented and few erythematous patches on his face, shoulder and trunk. There was no history of seizure, deafness or visual problem. Family history was insignificant.

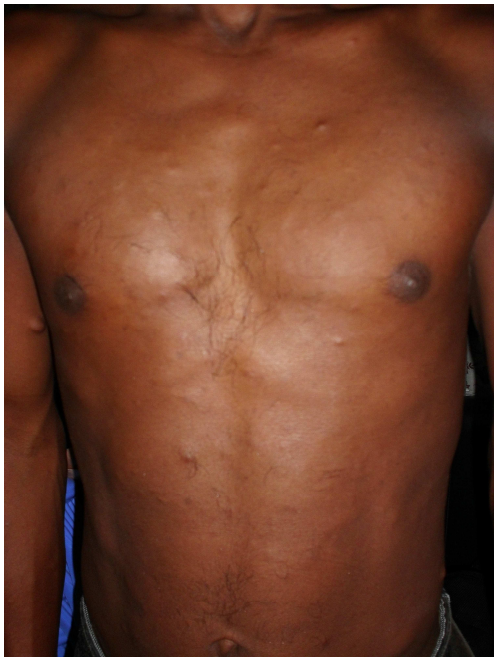

Figure 1 Multiple soft, non-tender, dome shaped nodules of variable size over trunk and extremity.

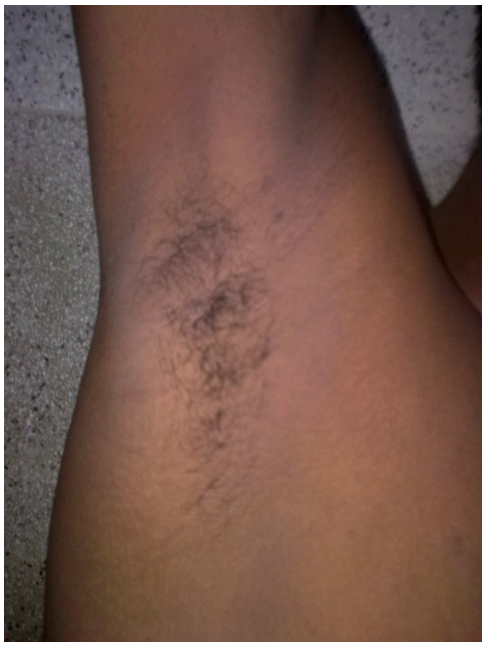

Figure 2: Axillary freckling (Cowe's sign).

Cutaneous examination revealed multiple soft, non-tender, dome shaped nodules of variable size, ranging from 1 to 1.5 cm in diameter, over the above mentioned sites (**Figure 1**). These nodules demonstrated characteristic 'buttonholing' i.e. application of pressure to these nodules caused them to herniate into

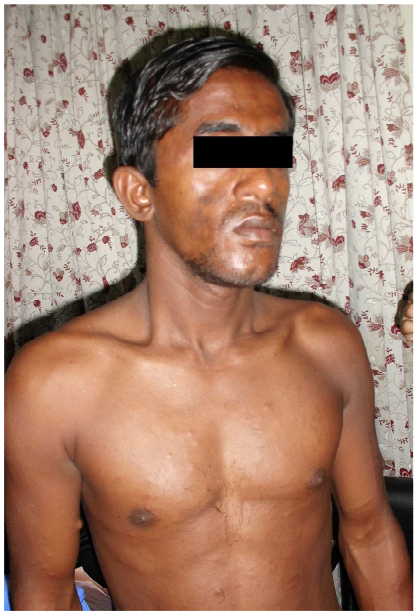

Figure 3 Erythematous patches with sensory deficit on the forehead

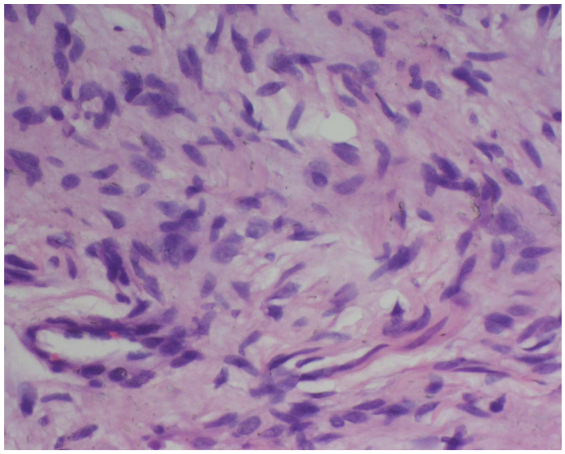

Figure 4 Histopathology showing features of neurofibroma

dermis. There were nine hyperpigmented patches with intact sensation (café-au-lait spot) on trunk and extremities, which were 1-2 cm in diameter. Both axillae showed freckling (Crowe's sign), (**Figure 2**).

In addition to these lesions there were also few hypopigmented and erythematous patches with sensory deficit on the face (**Figure 3**), shoulder and trunk. He had bilateral symmetrical thickening of great auricular, common peroneal

and posterior tibial nerves, which were non-tender. Quick muscle test of both hands and feet was normal. Systemic examination did not reveal any abnormalities. Slit-lamp examination showed Lisch nodules in the iris.

Though the initial impression was suggestive of lepromatous leprosy with erythema nodosum leprosum, after careful history and examination a clinical diagnosis of NF 1 with coexisting borderline tuberculoid (BT) leprosy was made. A slit-skin smear from earlobes, forehead and hypopigmented patches showed presence of *M. leprae*. The bacteriological index was 2+. Skin biopsy from nodules showed features of neurofibroma (**Figure 4**).

These findings confirmed the diagnosis of NF1 and BT leprosy. The patient received treatment for multibacillary leprosy and was counseled about NF. He was under regular follow-up during the treatment period of 12 months and showed gradual disappearance of leprosy lesions.

Discussion

NF is one of the neurocutaneous syndrome manifested by developmental changes in nervous system, bone and skin lesions like neurofibroma, café-au-lait spot, axillary freckling etc.[2] The majority of cases are inherited (autosomal dominant) and less than 10% arise from spontaneous mutation.[3]

Schwann cell is the primary target for both NF and leprosy. Electron microscopic studies have demonstrated that in NF most cells are derived from Schwann cells and in leprosy Schwann cells are predominantly invaded by *M. leprae*. Bacilli are present in cytoplasm of the Schwann cells and not in axon.[4,5] Ghosh *et al*.[6] proposed that NF might predispose Schwann cells to invasion by *M. leprae*, but the pathogenic relationship, as well as, association is rare.

Coexistence of these two disorders may lead to diagnostic dilemma as nodules and nerve involvement is common both in leprosy and NF.[7] Leprosy may be mistaken for NF in nonendemic area and thus appropriate treatment may be delayed. Conversely in endemic area NF may be mistaken for leprosy which occurred in our case at first sight. Hence, inappropriate treatment may be instituted.[8] Several instances of such diagnostic dilemma had been reported. Mittal *et al.*[9] reported a case of NF with gross enlargement of all peripheral nerve trunk simulating leprosy.[9] On the other hand, Khandpur *et al.*[10] reported a case of NF where an unusual hypopigmentation with symmetrical nerve enlargement raised the strong clinical suspicion of coexistent lepromatous leprosy. But leprosy was ruled out by microbiological, histopathological and electrophysiological studies. Swift has reported presence of lepra bacilli within neurofibroma in two cases of NF with lepromatous leprosy.[11] NF has been reported to be associated with borderline tuberculoid,[7] borderline lepromatous,[12] lepromatous,[13,14] histoid[15] and pure neuritic[16] leprosy.

Conclusion

NF and leprosy, two different pathologies of overlapping presentation, may sometimes cause diagnostic dilemma. Co-existence of both the disorders may need to be considered in appropriate clinical settings.

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