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Case Report

Plexiform Neurofibroma of Lip Presenting as Macrochelia with Borderline Lepromatous Leprosy – A Rare Association of Two Unrelated Diseases: A Case Report

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The coexistence of leprosy and neurofibromatosis in the same patient is interesting as two diseases have multifarious etiology and pathology with the primary target being Schwann cells. Multiple nodules and pigmented macules are a hallmark of both diseases which may pose difficulty in diagnosing either. Thus, when both diseases occur simultaneously in the same patient, it may pose a diagnostic dilemma and therapeutic hindrance. Here, we report a case of a 52-year-old male who presented with manifestations of both these diseases. Proper history and histopathological examinations should be done so that dermatologists/ leprologists/ other clinicians do not miss such diagnosis, considering the rare existence of both diseases in same individual.

Keywords: Neurofibroma, Leprosy, Macrochelia, Co-existence

Introduction

Neurofibromatosis is a genodermatosis whereas, leprosy is an infectious disease caused by *Mycobacterium leprae* destroying nerves and other organs characterized by nodular thickening of nerves due to the proliferation of schwann cells (Chander et al 2005). The coexistence of leprosy and neurofibromatosis in the same patient is interesting as two diseases have multifarious etiology and pathology with the primary target being Schwann cells. Both diseases are characterized by multiple pigmented macules, swellings of the skin, with thickening of nerves which may pose difficulty in the diagnosis of either (Rao et al 2015). Neurofibromatosis has earlier been reported to coexist with lepromatous, histoid, pure neuritic, and borderline tuberculoid leprosy but not with borderline lepromatous leprosy (Lam & Harun Nor Rashid 2022). Here, we highlight a case of an elderly male who presented with manifestations of both neurofibromatosis and borderline lepromatous leprosy.

Case Report

A 52-year-old male presented to the dermatology OPD with complaints of multiple swellings over the lip and body since birth and multiple hypopigmented lesions over the back for 2 years. The swelling over the lip was initially pea-sized and gradually increased to present size of approximately 3x5 cm and has not increased in

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Fig. 1: (a)- Plexiform neurofibroma present over the upper lip and single well defined neurofibroma present over the forehead. (b) Madarosis present over the left eyebrow.

size for 20 years (Fig. 1a and b). The swelling was not associated with pain, redness, discharge, or any rapid increase in size. It is not associated with any difficulty in eating, drinking, and speaking. There was no history suggestive of mental retardation, convulsions, deafness, or visual impairment.

In the last 2 years, the patient started noticing multiple well to ill-defined hypopigmented lesions. They first appeared over the back and gradually progressed to involve bilateral upper limbs, and abdomen (Fig. 2). The lesions are associated with decreased sensation, and loss of sweating. They were not associated with itching. There was no history of blurring of vision, bone pain, hoarseness of voice, alopecia, anosmia, swelling of feet, slippage of chappals, epistaxis, difficulty in doing fine movements like buttoning of shirt, mental retardation, convulsions, deafness or bony deformities. There was no history of leprosy in the past, family and distant relatives.

On examination, a single well to ill-defined solitary fusiform swelling studded with multiple small pin-head-sized nodules was present just below the philtrum extending towards the left vermilion border of the upper lip. Two well-defined soft, pedunculated swellings of size 1 X 2 cm and 2 X 3 cm were present over the back and forehead respectively (Fig. 3). All the swellings were non-tender, soft to firm in consistency, and non-adherent to underlying structures and overlying skin. There were no changes in the skin overlying the swelling. Axillary and inguinal

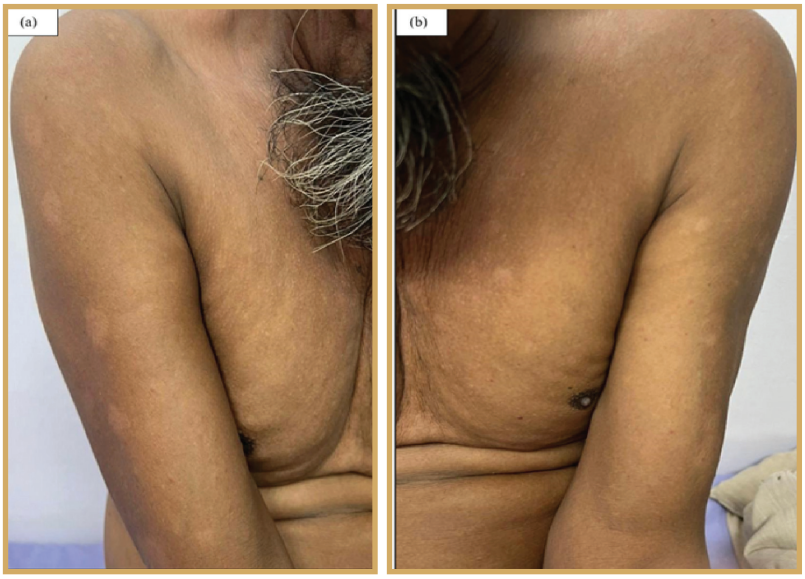

Fig. 2: Multiple well defined hypopigmented macules present over the bilateral arms and trunk.

freckling and café-au-lait macules were absent.

Multiple well-defined hypopigmented macules, ranging from 1 cm X 2 cm to 3 cm X 4 cm, were present over the back, abdomen, and upper limbs. These macules had a dry surface, decreased hairs, and well to ill-defined margins. Touch, pain, and temperature sensations over these macules were diminished over the lesions. Madarosis was present. On nerve examination (Brandsma 1981), bilateral ulnar nerves, right radial cutaneous nerve, and right common peroneal nerves were grade I thickened and had grade I neuritis. Book test, card test, pen test, oschner clasping test and beek test were normal (Brandsma 1981).

A slit skin smear (SSS) from ear lobules, eyebrows, and hypopigmented macules (Mahajan 2013) showed the presence of acid-fast bacilli. Average bacteriological index (BI) was 4 +. Patient denied SSS examination from the lip swelling. A punch biopsy was performed from the hypopigmented macule over the left upper arm which showed an epidermis lined by stratified squamous

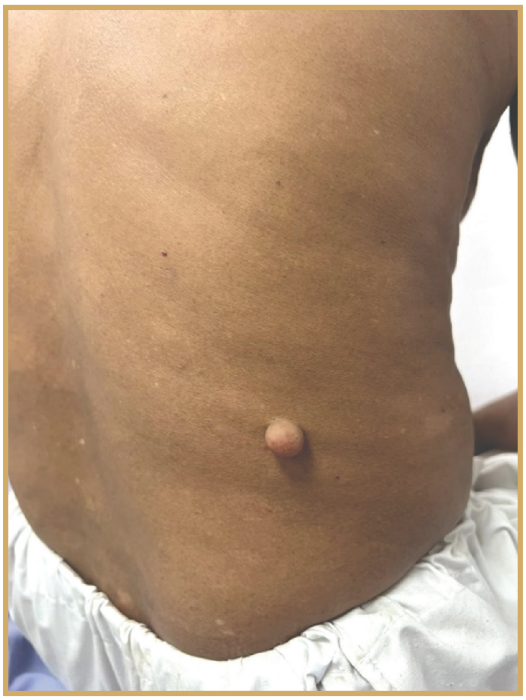

Fig. 3: Multiple well defined hypopigmented macules and solitary neurofibroma present over the back.

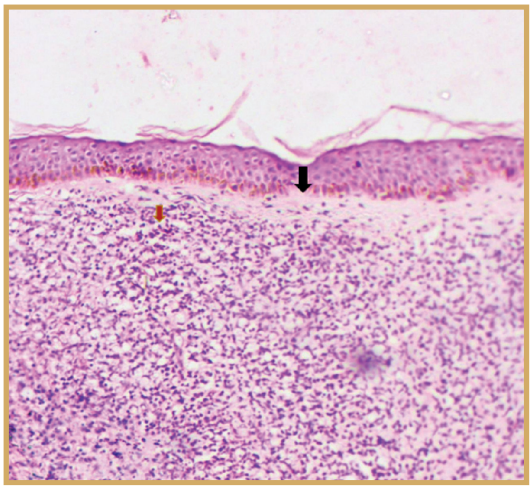

Fig. 4: Histopathology from hypopigmented macule showing grenz zone (black arrow) and lymphocytic infiltration present in the dermis (red arrow).

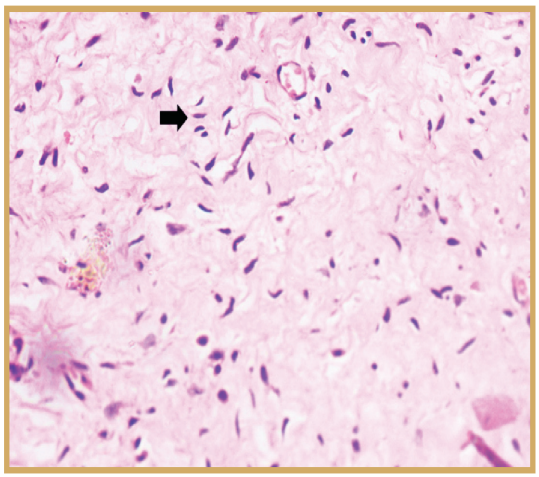

Fig. 5: Histopathology from nodular swelling of back showing spindle cells (black arrow) with interspersed collagen bundles.

Table 1: Previously published cases of patients having neurofibromatosis and leprosy.

S.NO.	AUTHOR, YEAR	AGE/SEX	COUNTRY	DIAGNOSIS
1.	Quyum et al (2015)	26/M	Bangladesh	NF1 with borderline tuberculoid leprosy
2.	Mallawaarachchi, Gunasekera (2012-2014)	31/M	Srilanka	NF1 with lepromatous leprosy
3.	Angoori et al (2010)	60/M	India	NF1 with borderline lepromatous leprosy
4.	Chander et al (2005)	13/M	India	NF1 with borderline tuberculoid leprosy
5.	Rao et al (2015)	25/M	India	NF1 with pure neuritic leprosy
6.	Swift (1971)	42/M	USA	NF1 with lepromatous leprosy
		59/F	USA	NF1 with lepromatous leprosy

epithelium, loss of rete ridges, presence of grenz zone and ill-formed granuloma with lymphocytic infiltrate in the dermis. These findings were suggestive of borderline lepromatous Hansen disease (Fig. 4).

The patient denied biopsy from the lip swelling, so a biopsy was done from the back swelling which showed the presence of spindle cells with interspersed collagen bodies which confirmed the diagnosis of neurofibroma (Fig. 5).

Based on history, clinical findings, and histopathology the diagnosis of borderline lepromatous leprosy with neurofibromatosis was made. The patient was started on multibacillary multidrug treatment (MBMDT) for leprosy. He was also advised surgical excision of neurofibromas and was counselled regarding the disease.

Discussion

Neurofibromatosis (NF) is a genodermatosis, a mutation in the NF-1 gene, characterized by neurocutaneous manifestations. NF is classified into 8 types by Riccardi, namely NF-1 (von Recklinghausen disease), NF-2 (acoustic form), NF-3 (mixed form), NF-4 (the variant with diffuse Café au lait macules and neurofibromas with or without CNS tumours), NF-5 (the segmental type with Café au lait macules or neurofibromas limited to a unilateral, segmental distribution), NF-6 (Café au lait spots without neurofibromas), NF-7 (the late-onset type) and NF-8 (the unspecified type) (Tiwary 2016).

It is classified into three types- localized (most common), diffuse, and plexiform. The plexiform type is pathognomonic for neurofibromatosis type 1 (NF 1) (Messersmith & Krauland 2024). Plexiform neurofibroma is a rare variant of NF-1 arising from nerve sheath cells or peripheral nerves as nodular thickenings and deformed masses involving skin folds and connective tissues giving the appearance of a “bag of worms” (Alrehaili 2023) .

Neurofibromas over head and neck regions are seen only in 25% of cases out of which only 6.5% cases were present with oral lesions. Lesions are most commonly seen over the tongue, with rare presentation over the lips. This location can be easily confused with other lesions of the lip, like leprous macrochelia due to Hansen’s, mucocele, and traumatic fibroma (Alrehaili 2023). In our case, an unusual solitary fusiform swelling was present over the upper lip.

Leprosy also known as Hansen’s disease is a chronic granulomatous disease caused by a rod-shaped, acid-fast bacillus known as mycobacterium lepra clinically characterized by the presence of hypo and hyperpigmented anaesthetic skin lesions. It is classified by Ridley and Jopling based on immunological and histological features into five types: tuberculoid (TT), borderline tuberculoid (BT), mid borderline (BB), borderline lepromatous (BL), and lepromatous leprosy (LL) (Brandsma 1981).

Hence, the etiology and pathophysiology of both diseases neurofibromatosis and leprosy are different but clinically both are characterized by multiple nodules and pigmented macules (Lam & Harun Nor Rashid 2022). It was difficult to diagnose such case and for which biopsy was done from different nodules and by histopathological examination both the diagnosis were made in the same individual.

In the review of literature only a total of 6 case reports with both entities occurring in the same patient simultaneously across the world were found (Table 1). Of a total of 3 cases reported from India only a single case of borderline lepromatous leprosy with NF has been documented (Angoori et al 2010) with similar findings to our case. These cases are summarized in the Table 1.

Conclusion

Neurofibromatosis associated with Hansen’s disease is rarely reported in the literature. Therefore, in leprosy endemic areas lesions of neurofibromatosis can be mistaken for lesions of leprosy and vice versa. Extensive workup including history, clinical examination and investigations are required to differentiate both these entities this may pose a diagnostic dilemma and therapeutic hindrance., Hence, physicians should be mindful and investigate the coexistence of other diseases and should not be blinded by primary diagnosis.

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