ORIGINAL RESEARCH PAPER
Volume - 15 | Issue - 03 | March - 2026 | PRINT ISSN No. 2277 - 8179 | DOI : 10.36106/ijsr
INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

SEGMENTAL NEUROFIBROMATOSIS: AN UNUSUAL CASE AT AN UNCOMMON SITE

Dermatology

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ABSTRACT

Background: Neurofibromatosis type 1 (NF-1) is a multisystem genetic disorder characterized by café-au-lait macules and neurofibromas. Segmental neurofibromatosis (SN), a mosaic and uncommon variant, is restricted to a limited body segment and occurs far less frequently than classic NF-1. **Objective:** To report a rare presentation of SN localized to the left mandibular region and ear pinna-an unusual and diagnostically challenging site. **Case Summary:** A 36-year-old woman presented with multiple, gradually progressive papules and nodules over the left jawline and ear pinna. Lesions were soft to firm and demonstrated button-hole sign. No systemic or ophthalmologic features of NF-1 were detected. Histopathology confirmed a dermal neurofibroma. Findings were consistent with segmental neurofibromatosis. She was counselled about genetic implications and offered surgical excision. **Conclusion:** SN may be subtle and localized, leading to delayed diagnosis. Recognition is important because patients may transmit generalized NF-1 due to gonadal mosaicism. Management includes excision and genetic counselling.

KEYWORDS

Segmental Neurofibromatosis; Neurofibroma; Mosaicism; Neurocutaneous Syndromes

INTRODUCTION

Neurofibromatosis type 1 (NF-1) is an autosomal dominant genodermatosis with a wide spectrum of cutaneous and systemic manifestations, including neurofibromas, café-au-lait macules, skeletal abnormalities, and ocular lesions.[1] With an incidence of approximately 1 in 3000 individuals, NF-1 is among the most common neurocutaneous syndromes.[4]

Segmental neurofibromatosis (SN), also termed mosaic-localized NF or Riccardi type V, is a rare variant characterized by lesions confined to a unilateral, sharply demarcated body segment.[5] Fewer than 100 cases haveI been documented worldwide. We report a case of SN involving the left mandibular region and pinna-an uncommon distribution that underscores the importance of recognizing this mosaic subtype.

Case Report

A 36-year old woman presented with a 4-year history of asymptomatic papules and nodules over the left jawline and ear pinna. Lesions began beneath the mandibular margin and gradually increased in number.

Cutaneous examination revealed multiple, soft well-defined, skin colored papules and nodules ranging from 3 mm to 1 cm noted over left ear pinna and left mandible. Some lesions exhibited a positive button-hole sign. No similar lesions were present elsewhere. Occular examination was normal.Family history was negative.Classical NF-1 findings like café-au-lait macules, axillary or inguinal freckling, Lisch nodules, plexiform neurofibromas, and skeletal defects were absent.

Histopathological examination showed a well-circumscribed dermal tumor composed of spindle cells arranged in bundles and sheets, consistent with a neurofibroma.

Based on the localized distribution and supportive histopathology, a diagnosis of segmental neurofibromatosis was established. The patient was counselled regarding genetic implications, including the risk of gonadal mosaicism, and offered surgical excision.

DISCUSSION

NF-1 is caused by NF1 gene mutations on chromosome 17 and demonstrates extensive clinical variability.[1] SN results from post-zygotic mutations leading to mosaicism and localized manifestations. Despite being mosaic, affected individuals may transmit generalized NF-1 if gonadal involvement occurs.[3]

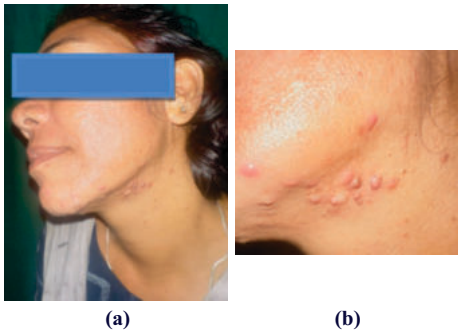
The prevalence of SN is low, estimated at 0.0014% -0.002%. [2,4,5] Clinical features vary and may include localized neurofibromas, café-au-lait macules, or axillary freckling. Because SN remains restricted to one segment, it is frequently underdiagnosed.

Differential diagnoses in our case included keloids, adnexal tumors, nodular acne, angiolymphoid hyperplasia with eosinophilia, and cutaneous sarcoidosis. Histopathology is essential for confirmation. On histopathology arborizing schwann cells and collagenous stroma can be seen.[1]

Management includes surgical excision, CO2 laser ablation, electrodessication, and counselling regarding genetic implications.

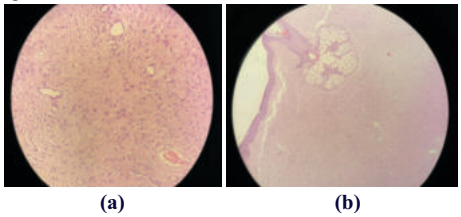
CONCLUSION

Segmental neurofibromatosis is a rare mosaic variant of NF-1 with lesions confined to one body segment. Its subtle presentation may delay diagnosis. Early identification is vital to address complications and counsel patients regarding the risk of transmitting generalized NF-1. Treatment primarily involves surgical removal and genetic counselling.



(a) (b)

Fig1(a) Multiple skin colored papules noted over ear pinna and below mandible. **(b)** Skin colred papules noted over mandle and below extending to neck



(a) (b)

Fig2 (a):spindle shaped nerve fibres noted in dermis. **(b)** Dermal tumor composed of spindle cells arranged in bundles and sheets

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