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Exploring Segmental Neurofibromatosis: Case Studies and Genetic Implications

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Abstract: ***Neurofibromatosis is a genetic disorder of neural crest - derived cells that primarily affect growth of neural tissues. It is broadly divided into three categories: (a) von Recklinghausen's neurofibromatosis or NF - 1, (b) bilateral acoustic neuroma (NF - 2), and (c) all other neurofibromatoses, including alternate or atypical forms of the disease. The patients with generalized form of NF1 are characterized by multiple café - au - lait spots and neurofibromas and diagnosed easily. But when an individual has small number of lesions in a limited region of the body it could be neglected by the patient or not be recognized by the clinicians as a segmental form of neurofibromatosis. We describe three cases of segmental neurofibromatosis (SNF). These cases have been classified as segmental NF according to Riccardi's definition of SNF and classification of neurofibromatosis. Segmental form of NF may evolve into a complete form over time.2 Also, this disorder may be transmitted to the offspring of these individuals. Hence genetic counselling of these individuals must include these facts.***

Keywords: Segmental neurofibromatosis, neurofibromatosis 1, café - au - lait spots, neurofibroma

1. Introduction

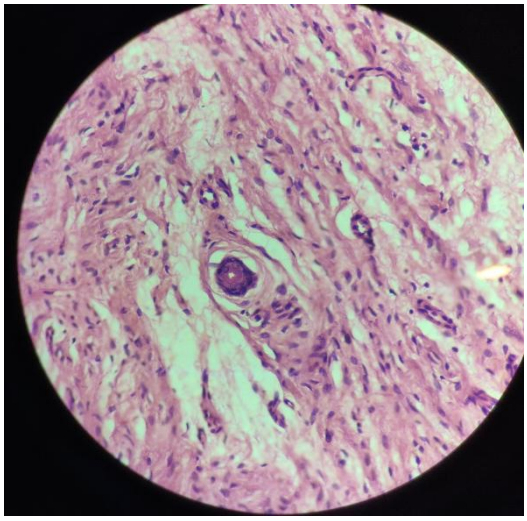
Neurofibromatoses are a set of inherited disorders designated as neurofibromatosis type 1 (NF1), neurofibromatosis type 2 (NF2), and schwannomatosis. They tend to result in the development of benign tumors of nerve sheath. The three clinical entities are distinguished by specific clinical features and are due to mutations in distinct genes.1 Neurofibromatosis type 1 is the most common of these disorders, affecting approximately 1 in 3500 individuals worldwide and with nearly 100% penetrance of the disease.

Segmental neurofibromatosis (SNF) was first described by Crowe *et al*. in 1956 and the authors termed it *Sectorial neurofibromatosis*. In 1977, Miller and Sparkesrenamed this term as SNF. Riccardi included SNF in his classification of neurofibromatosis (NF) as type V. The clinical features of SNF were also established by him as: Café - au - lait spots and/or neurofibromas in a single unilateral segment of the body, with no crossing of the median line, no family history, and no systemic involvement. Roth *et al*. has further subdivided the SNFs into four subtypes: True segmental, localized with deep involvement, hereditary, and bilateral.

2. Case Report

A 54 yr male, primary school teacher by profession presented to dermatout patient department with history of multiple skin colored, dome shaped, soft to firm nodules and papules over extensor aspect of right arm, right lateral aspect of upper back over C7 - C8 dermatome since 15 years not associated with any complaints. The lesions were gradually progressive in nature. Lisch nodules in iris, Café - au - lait spots & axillary freckling was not found. There was no history of seizures. There was no family history of such lesions. MRI showed no abnormal findings.

Skin punch biopsy was taken from one of the nodules which showed - findings concurrent with neurofibromatosis.



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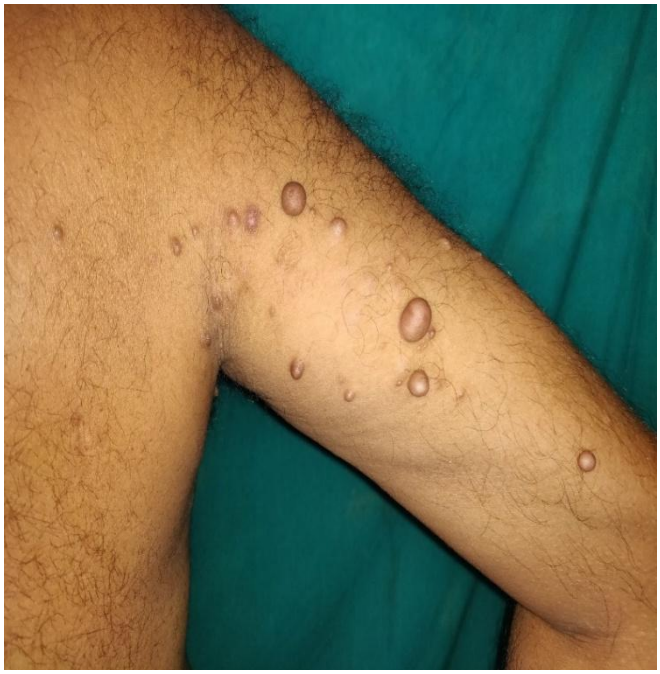
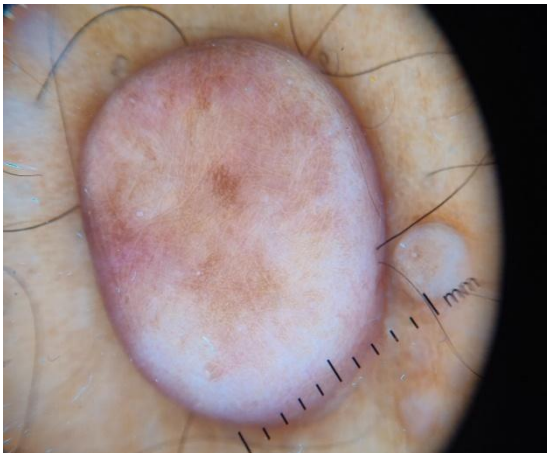


Clinical Image showing multiple well defined nodules over right arm, back.



Dermoscopic examination – shows Fingerprint - like structures, Pink - red structureless areas, Peripheral pigment network.

3. Discussion

Our patient had neurofibromatosis that was restricted to the dermatomal distribution of a specific nerve root and did not cross the midline. In addition, the patient had no family history of neurofibromatosis. As a result, we believe that this patient has a type of neurofibromatosis known as Segmental Neurofibromatosis according to Riccardi's classification.

Neurofibromatosis manifests itself in a wide range of ways. Although the disease manifests itself in a a generalized form for majority of cases (90%), certain patients appear with localised neurofibromas or café - au - lait spots.[3, 4] Crowe et al. first described this as sectorial NF. They argued that neurofibromatosis is caused by somatic mutation in a specific area of the body, with no hereditary transmission of the characteristic.[5]

Recognising the disease's variability, Riccardi proposed a useful system for categorising NF into eight subtypes (NF - I to NF - VIII). One of these forms (NF - V) is segmental NF, which is distinguished by the confinement of café - au - lait spots and neurofibromas to a specific region of the body. Riccardi considered that the most likely source of development was a postzygotic somatic mutation in primitive neural crest cells, and thus the lesion should be absolutely non - inherited. SNF is defined as café - au - lait macules or neurofibromas in a single, unilateral section of the body with no crossing of the midline, no family history, and no systemic involvement, according to him.[4]

SNF is an uncommon condition with an estimated frequency of 0.0014 to 0.002%. It is an example of mosaicism in which a postzygotic NF1 gene mutation on the proximal long arm of chromosome 17 causes localised illness. Mutations in genes can occur in both somatic and gonadal cell lines. It is suggested that gonadal mosaicism is to blame for instances of patients with localised sickness bearing offspring with generalised NF1.[6, 7]

In light of these characteristics, what approach should be taken for the patient who presents with localised NF lesions? To begin, a thorough physical examination should include a search for neurofibromas, Lisch nodules, or other café - au - lait spots. If the lesions are seen bilaterally, systemically, or in family members, the patient is not diagnosed with segmental NF. Even if the patient adheres to Riccardi's description, he may develop lesions systematically or in a specific location over time, and the disease would have to be classed as a separate subgroup of segmental NF. Patients with SNF have also been reported to pass on NF to their offspring.[6]

4. Conclusion

Segmental neurofibromatosis is a rare type of neurofibromatosis, and a clinician must be vigilant for its diagnosis. There are no specific management guidelines for segmental NF. The patient should be advised that they do not have generalised NF 1 and that their risk of disease - related consequences is minimal.

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