INTERNATIONAL JOURNAL OF MEDICAL AND APPLIED SCIENCES
E-ISSN:2320-3137

## Case Report

# NEUROFIBROMATOSIS TYPE – 1 (NF-1) OF HEAD AND NECK: A CASE REPORT

**Jain S.K., Gupta Harish, Jain Vandana, Garg K.M**

**D.C. Hospital, Jaipur and M.G. Medical College & Hospital, Jaipur**

**CORRESPONDING AUTHOR: Dr. S.K. Jain, 68, Vishveshvariya Nagar, Gopalpura By pass, Jaipur-18**

## ABSTRACT

Neurofibromatosis (NF) is one of the most common genetic disorders and it has been estimated that approximately 25% of all neurofibromatosis are found in head and neck. NF type 1 of the head and neck suffer from one of the most dramatic cosmetic disabilities, which may lead to psychological trouble and social segregation. There is no known cure for NF and there is a dilemma in the management of these cases.
**Keywords:** Neurofibromatosis, head, neck, case report

## INTRODUCTION

The Neurofibromatosis (NF) is the most prevalent neurological genetic disorder found in the population, affecting 1 in 3000-4000 individuals. It is autosomal dominant disease without predilection for sex, race or color (1). Frederich Von Recklinghausen in 1882, proposed the first systematic classification of nerve tumors (2). Briefly, NF 1 exposes a characteristical cutaneous phenotype including benign neurofibromas, which are mixed tumors composed of all cell types found in the pheripheral nerves, hyperpigmented macules, the so called axillary/Inguinal freckling, as well as pigmented hamartomas of the iris called Lisch nodules. NF-2 on the other hand is mainly restricted to tumor's of the central and pheripheral nervous system, which are only seldomly accompanied by cutaneous disorders (3).

In 1991, the gene causing NF-1 was found and cloned in March 1993. The gene causing NF-2 was found on chromosome 22, but variant forms may exist (4, 5). In general, patients with NF-1 and NF-2 should undergo yearly neurological and ophthalmological examinations (6). Treatment of severe and disfiguring tumors is usually performed by surgery. Although benign complete excision of tumors often remains a therapeutic endeavor due to their close association to nerves. Regarding NF-2, the total surgical resection of vestibular schwannomas is a suitable therapeutic option and may result in definite tumor control. However, due to the frequently multilobulating and infiltrating character of the tumors, consecutive damage to the cochlear nerve or facial nerves is associated with a high risk of permanent hearing loss and other malformations (7).

1

## CASE REPORT:-

A 17 yrs. old girl presented in the out patient department with c/o swelling upper part of left side neck and scalp, which gradually increased involving the left side of neck and upper third of scalp and back of pinna to reach its present size 20×15 cm. She had another soft tissue swelling dark brown in color over chest wall near midline 6×6 cm with no bony involvement. She had multiple coffee colored spots all over limbs and body (Café au lait spots). **(Photo- 1).**

Plastic surgery for this unsightly tumor was planned explaining the risk of recurrence and failure of graft uptake. Excision of the lesion up to muscles in neck and periosteum on scalp and perichondrium over pinna done and SSG applied. On 5th POD, dressing, graft was taken up well and patient was discharged from the hospital on 8th POD. On f/u visits few small non healing areas were seen, most of areas healed in subsequent dressings except one on neck, 3×3 cm size. Which required a repeat skin grafting at 3 months interval. Patient seen last month, at 6 months follow up, was perfectly well and cheerful. **(Photo -2).**

**HPE:** revealed diffuse dermal proliferation of cells with focal pacinian corpuscles and hypertrophied nerve trunks. The dermal adnexa displaced by neural tissue: neurofibromatoses.

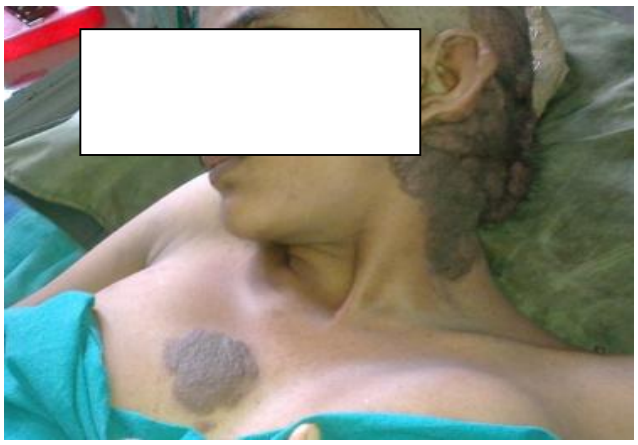

**Photo- 1: Pre Operative Photograph showing lesions on neck, scalp and chest wall**

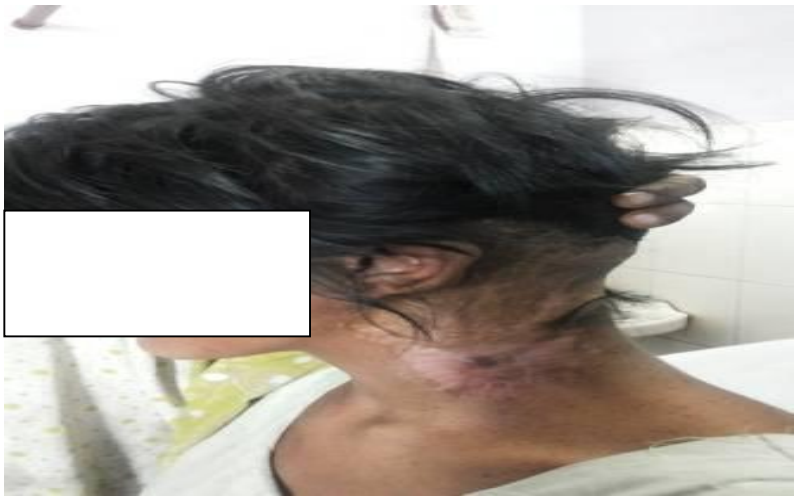

**Photo- 2 :Post Operative photograph at 7 months follow up**

## COMMENTS:-

Patients with NF type -1 of the head and neck suffer from one of the most dramatic cosmetic disabilities, which may lead to psychological trouble and social segregation. Our patient was a 17 yrs female underwent excision of the lesion with SSG with average outcome, requiring regrafting at residual areas 3 months later. Patient last seen at 6 months follow up was much cheerful and perfectly well with good cosmetic outcome. Hence excision and split skin grafting as treatment of NF type-1 of head and neck should be offered to these cases and to establish role of this type of surgery, more and more, of its use on larger number of patients is recommended.

2

## REFERENCES:-

1. Ghareeb F.M., Makeen K, Fawzy A.T., et al: Facial neurofibromatosis: Cosmetic considerations. Egypt. J. Plast. Reconstr. Surg., 25(2):139-146, 2001.
2. Jackson I.T., Carbonnel A., Potparic Z., et al.: Orbitotemporal neurofibromatosis: Classification and treatment. Plast. Reconstr. Surg., 92(1):1-11, 1993.
3. Baser ME, Evans DG, Gutmann DH. Neurofibromatosis 2. Curr Opin Neurol 16, 27-33 (2003).
4. Repado F., Simo R. and Small M.: Neurofibromatosis type 1 of the head and neck: dilemmas in management. J. Laryngol. Otol., 115:151-154, 2001.
5. Friedman J.M.: Neurofibromatosis 1: clinical manifestations and diagnostic criteria. J. child. Neurol., 17 (8): 548-554, 2002.
6. Gutmann DH, Aylsworth A, Carey JC, Korf B, Marks J, Pyeritz RE, Rubenstein A, Viskochil D. The diagnostic evaluation and multidisciplinary management of neurofibromatosis 1 and neurofibromatosis 2. Jama 278, 51-7 (1997).
7. Kramer K, Hasel C, Aschoff AJ, Henne-Bruns D, Wuerl P. Multiple gastrointestinal stromal tumors and bilateral pheochromocytoma in neurofibromatosis. World J Gastroenterol 13, 3384-7 (2007).