Case Report

THE ELEPHANT MAN*: HUMAN MONSTROSITY OR MARVEL?

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Abstract

Neurofibromatosis type 1 is an inherited neuro-cutaneous syndrome. Described by Friedrich von Recklinghausen for the first time in 1882, it is characterized by benign tumors (neurofibromas) arising from neural sheath cells, subcutaneous, visceral peripheral or cranial nerves and cutaneous manifestations like café-au-lait spots. Plexiform Neurofibromatosis (PNF) is seen exclusively in neurofibromatosis type - 1. PNF is common along trigeminal nerve though segmental plexiform neurofibromas in scalp, neck, chest, pelvis of abdomen have been reported. When present along the Trigeminal never, it can cause gross disfigurement & cosmetic as well as functional problems. We hereby report a case of facial plexiform neurofibromatosis.

Key Words: - Neurofibromatosis type 1, plexiform neurofibromatosis, café-au-lait spots

INTRODUCTION

Neurofibromatosis type1 (NF1) is an inherited neurocutaneous syndrome or phakomatosis characterized by benign tumors (neurofibromas) arising from neural sheath cells, subcutaneous, visceral peripheral or cranial nerves and cutaneous manifestations like café-au-lait spots (1). Plexiform Neurofibromatosis (PNF) occurs exclusively in NF1 in about 5-15% patients (2). PNF, especially along Trigeminal nerve cause gross disfigurement and pose cosmetic as well as psychological problems to patients. We report a case of facial Plexiform Neurofibromatosis.

CASE REPORT

A 33 year old male patient presented with history of slowly growing swellings over face since previous 22 years (Figure1). He had undergone surgical excision of swellings on the left side of neck (20 yrs back), left side of mandible (10 yrs back) and over left eyebrow (2 months back) for disfigurement and functional problems. His father had many small subcutaneous swellings all over the body (Figure 2). He had 3 siblings but none of them had similar complaints.

On examination, he had multiple, non-tender, pendulous swellings on both sides of face with irregular margins, hanging down in folds. The swellings had a characteristic 'bag of worms' consistency; overlying skin showed hyper-pigmentation (Figure 1). Scars of previous surgeries could be seen over left mandible and eyebrow. Patient also had numerous swellings over tongue causing considerable speech disturbance (Figure 2). He had axillary freckling and many café-au-lait spots >1.5 cm size (Figures 3, 4). His father had multiple neurofibromas of different sizes with characteristic 'button-hole' defects, and café-au-lait spots (Figure 5, 6) Thus a final diagnosis of facial Plexiform neurofibromatosis was made.

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* Joseph Merrick (1862-1889), an Englishman believed to be suffering from NF1 and Proteus Syndrome was described as 'The Elephant Man' by his biographer, Frederick Treves in 1923. Due to his grotesque physique and physical disabilities, he spent most of his adult life as a freak exhibit. Based on his life, award winning play and movie by the same name were staged in 1979 and 1980 respectively.

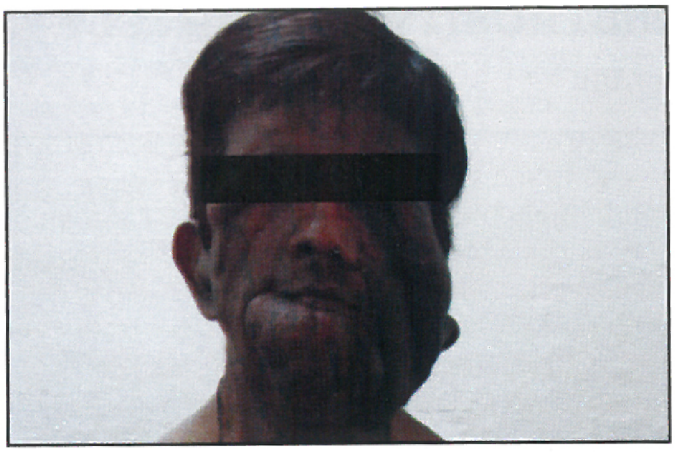

Figure 1

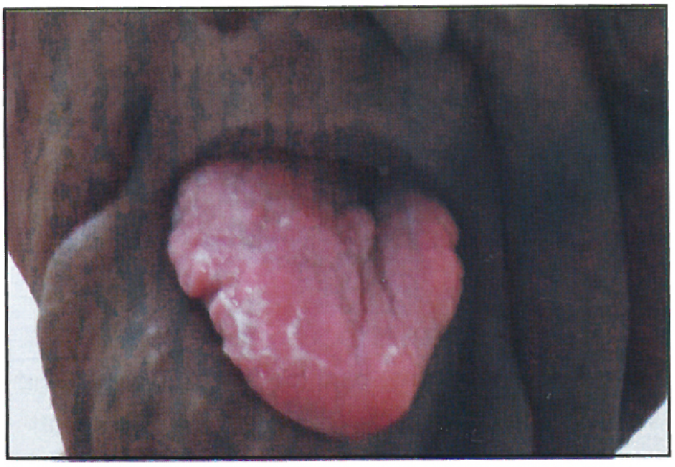

Figure 2

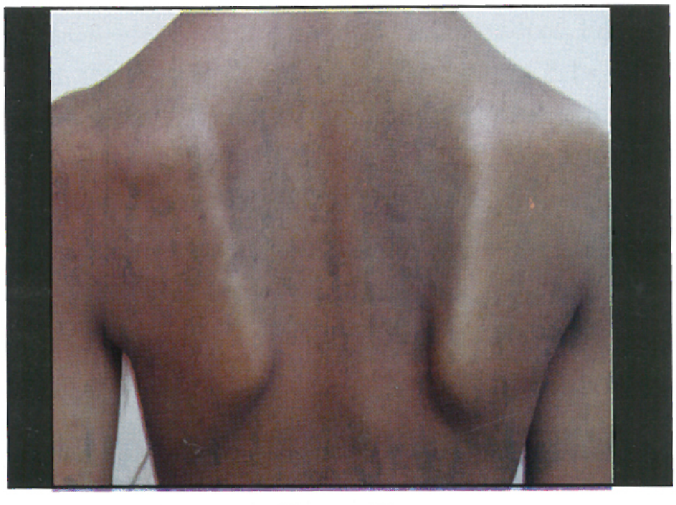

Figure 3

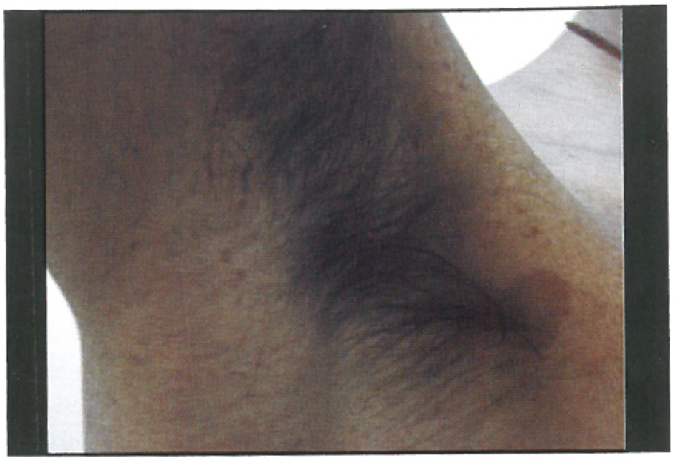

Figure 4

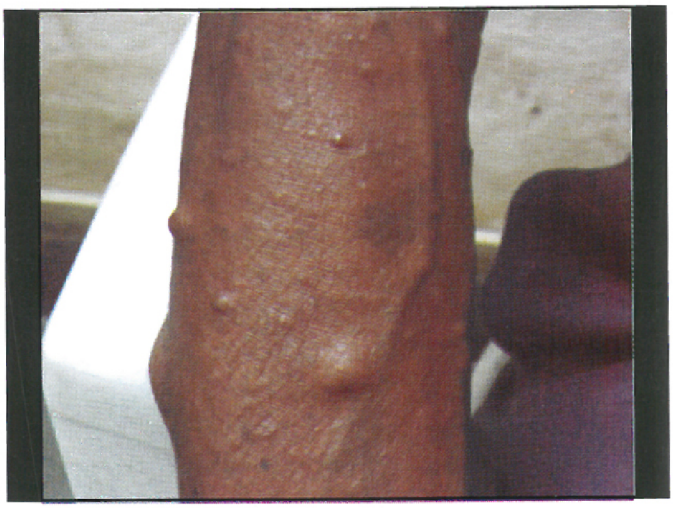

Figure 5

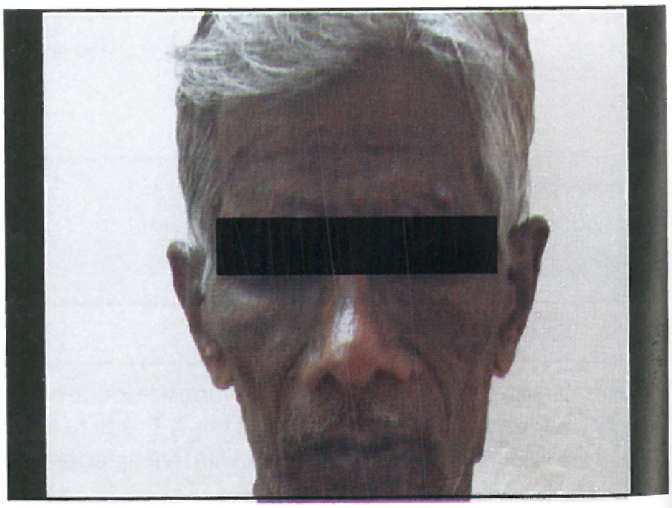

Figure 6

DISCUSSION

Neurofibromatoses (NF) are inherited neurocutaneous syndromes or phakomatosis, broadly classified as NF1 (von Recklinghausen'disease) and NF2; with distinct clinical manifestations and genetic abnormalities (defective genes on chromosome 17 and 22 in NF1and NF2 respectively). NF1, named after the German scientist, Friedrich von Recklinghausen who described it for the first time in1882 (3), is the commoner of the two. An autosomal dominant disorder, it is characterized by neurofibromas, café-au-lait spots(>6 in number, >1.5cm in size), freckling in axilla and groin(Crowe's sign),optic glioma, at least two Lisch nodules, sphenoid wing dysplasia or long bone cortical thinning and first degree relative with NF1. N1 is diagnosed if at least two criteria are fulfilled(1).

Pachydermatocele or Plexiform Neurofibromatosis (PNF) are uncommon tumors, occurring exclusively in patients with NF1. Plexiform neurofibromas are common along the distribution of Trigeminal nerve although isolated cases of segmental Plexiform neurofibromas have been reported in scalp, neck, chest, pelvis, abdomen or rarely gingiva (4,5).There are two types of Plexiform neurofibromatosis, nodular and diffuse. In diffuse type, the skin is thickened with overgrowth of epidermal and subcutaneous tissue giving a wrinkled and pendulous appearance (hence the name elephantiasis neurofibromatosa).

Facial PNF may be quite disfiguring and associated hemifacial hypertrophy can occur (6). The consistency of the tumor is described as "bag of worms" due to presence of soft areas interspersed with firm nodular areas. 4.6% patients with NF1, especially PNF have the risk of malignant transformation (7). Pain and rapid enlargement are the 'alarm'symptoms.

NF1 is a relentlessly progressive disease and due to the risk of malignant change, close and frequent monitoring is advisable. Surgical intervention is required for cosmetic or functional purpose, especially for facial PNF, or when malignancy is suspected. Excision of PNF may present problems for the surgeon as the tumor is entwined with normal tissue; there is increased risk of bleeding and tendency to recur.

The disease significantly affects the quality of life through alteration of health and appearance (8) and psychological as well as genetic counseling is needed

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