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STUDY 0F OPHTHALMOLOGIC PLEXIFORM NEUROFIBROMATOSIS TYPE-1 IN AN ADULT MALE

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Abstract

Neurofibromatosis type one in its primitive age was known as von Recklighausens disease.Neurofibromatosis type 1 is an autosomal dominant disorder of chromosomal 17q that appear in 4000 to 5000 births. The patients who show mild symptoms in early age may develop more severe problems. The patient here shows plexiform neurofibromatosis on the left side of the eye, and was gone through surgery. The eye plexiform restarted growing gradually after few months of surgery, and covered almost half of the face, then the patient undergone second surgery. In plexiform of eye the proliferation of Schwann cells occur inside the nerve sheath, producing an irregularly thickened, distorted tortuous structure. The tumors develop when cells of peripheral nerves multiply out of control within the nerve sheath; the eye plexiform tumor was benign, confirmed by histopathology of tumors. The patient shows large size café-au- lait macules on the chest, freckling, tumors on both the palms are very well prominent. The case was sporadic. Keywords: Plexiform, von Recklinghausen's, café-au- lait macules. 1.

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1. Introduction

The autosomal dominant genetic disorder neurofibromatosis type one has been reported from all parts of the world, and it has no racial or gender preponderance, (Victor Zion 1983). As many as seven different forms of plexiform have been described by Riccardi, (1984). However, the present study is sufficient to clearly distinguish neurofibromatosis type one (previously known as, von Reckling hausens or peripheral neurofibromatosis. The bilateral acoustic or central neurofibromatosis); and further sub classification's are unjustified by the inadequate evidence of (NIH consciences conference 1988). The type neurofibromatosis one is the commonest form accounting for over 90%of all the cases (Husen1989) with a prevalence of 1/4000-. (Asdourian and Lewis 1980, Husen and Harper 1988) in a population. Its inheritance is autosomal dominant and approximately 50% of cases represent new mutations. The neurofibromatosis gene was mapped to chromosome 17q11.2 it was appositionally cloned by (Cawthon, *et al.*, 1990). The neurofibromatosis gene spans at 350kb of genomic DNA with 60 exons. (Danglot, *et al.*, 1995). The neurofibromin protein is responsible for it, and has a high similarity to ras-specific (GTPs) are activating proteins called GAP related domain. Ballester, *et al.*, 1990).

Lisch nodules (Iris hamartomas).

They are virtually pathognomic of the disease and over the last few years they have acquired a definite role to establish the diagnosis of neurofibromatosis, NF-1 (Lewis and Riccardi 1981). The involvement of other tissues and orbit of the eye is common (Boltshauser *et al.,* 1985; Asdourian and Lewis 1980). The present communication deals with ophthalmic evaluation, and its envolvement with other facial features. In this study we specified the neurofibromatosis type one with plexiform of eye. The patient also showing many other well developed signs, criteria of neurofibromatosis confirmation.

2. Methodology

Materials and methods: are mainly based on the initial diagnosis and clinical findings of neurofibromas and cafe-au-lait spots. Diagnostic criteria. The well established signs and symptoms according to the criteria of National Institute of Health Consensus Conference on Neurofibromatosis (July 1988). The patients and samples were provided by Nazar Mohammad Neurofibromatosis Foundation (2001). The NIH criteria are met by an individual, who shows two or more of the following are; First- degree relative (parent, sibling, child) with known NF1. Six

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or more cafe-lait-spots over 5mm in diameter in a prepubertal patient and 15mm in post pubertal patient. Two or more neurofibromas of any type. Only one plexiform also confirms neurofibromatosis.

Eye examination was used for Lisch nodules that were done on patients using slit lamp to confirm NF-1 by ophthalmologist. History of the patient in detail for the construction of pedigree was carefully taken. Photographs and X-Ray, of the patients were also saved. Tumors were taken out by surgery and were kept in 10% formalin for histopathology.

3. Results and Discussion

The NF-1 patients were identified from the two different civil hospitals of Sindh. The NF1 patient's number recorded from LUMHS hospitals Hyderabad and Jamshoro were fifty-four, while forty-two patients were recorded rom civil hospital Karachi. The NF was diagnosed according to the NIH consensus diagnostic criteria 1988.The frequency of the NIH diagnostic criteria signs become more prominent with progression of patient's age. Lisch nodules were identified in 47 cases. The reported case shows plexiform of eye on left side. The plexiform has covered almost left side of the face of patient. The patient was under gone through surgery two times, (Fig. a) as the tumors grow again and sprout from the sides of scars. Lisch nodules were also seen and they progress with the age of patient, they appear as small tumors on the iris of the eyes. Patient's age was 45 years; who was reported eye plexiform, while both the eyes were packed with Lisch nodules. The plexiform was extended into the temporal region and was associated with a palpable bone defect in the temporal fossa with awfully cosmetic disfigurement. **(Fig. a)**. Lisch nodules cause

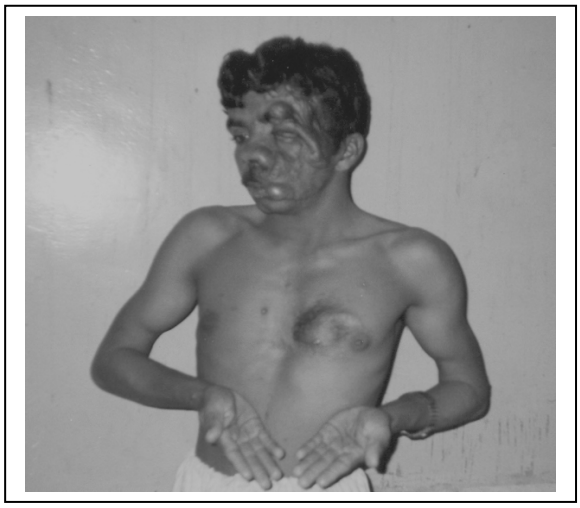

Fig. a. Neurofibromatosis on both palms and all over the body, large size café-au lait spots on the chest, and groin freckles are prominent.

complications in vision and weakness of optic system. Histopathology shows fibroblast, spindle nuclei, and mast cell **(Fig. b)**, the pedigree given in **(Fig. c)**, confirms that the case was sporadic; no other family

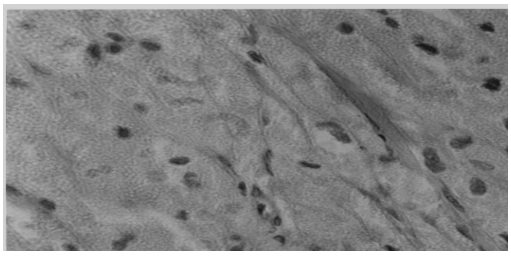

Fig. b Histopathology shows fibroblasts with spindle shaped nuclei, matrix, and inflammatory cells.

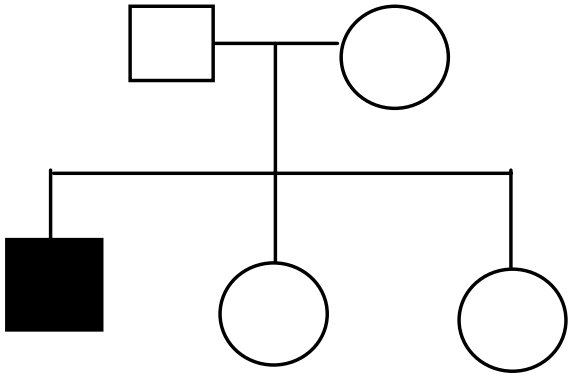

Fig. c. Shows pedigree of sporadic case. Proband age 45 years.

member was with NF1 signs. The patient shows neurofibromas, of different size, scattered all over the body, and well prominent freckled groins and skin. The plexiform neurofibromas are known to be associated with bone deficiencies involving the temporal region, occipital bone, sphenoid, orbital roof, and part of sella turcia and the general enlargement of the orbit, an observation which is in accordance with previous studies (Asdourian and Lewis 1980). Such deformities may be present as a co congenital defect or they may arise as a result of erosion by the tumor. The pulsatile proptosis represents the transmitted cerebral pulsation caused by the herniation of brain tissue into the orbit through such defects and is differentiated from the other causes of pulsatile proptosis especially carotico-cavernous fistula by the absence of the characteristic. The patients were searched with neurofibromatosis from Sindh, as this province is considered as a mine of genetic disorder, due to poverty, social, psychological, environmental reasons and pressures. The psychological negative effects are more flourished by poverty pressures, an observation

which is in accordance with many previous studies (Samuelsson 1981, Husen et al., 1989a, Riccardi 1992). Neurofibromatosis is a slowly progressive disorder with haemartomas lesions increasing in number and size with age, and the increasing appearance of diagnostic features in old age found in the present study supports this opinion, similar results have been reported by Huson et al., (1989a), the results of current study specify adding a few more symptoms which were not exhibited in previous studies. The additional symptom was a sevier photophobia, with its complications in fourth grades of its troubleness, and puts the patient in a very much distress, aggressiveness, irritation, and a psychic hate of light. All this collectively forces patient to experience a dreadful condition, additionally the patient become hypertensive. NF1 is a disorder.

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