NEUROFIBROMATOSIS PRESENTING AS A SOCIAL PROBLEM — A CASE REPORT

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INTRODUCTION

Neurofibromatosis (NF) type 1 (NF-1) is an autosomal dominant disorder with almost half cases as new mutation. NF-2 accounts for a very small percentage of the total cases of NF. The clinical diagnosis of NF-1 is always certain by ten years of age despite its very variable presentation. It can present in different ways and similarly causes different complications depending on its location and any neoplastic change.[1,2,3,8] We report an unusual social problem caused by NF-1 in a conservative society like that of Pakistan.

CASE REPORT

A two days old female neonate was admitted in the Special Case Baby Unit for observation. Her physical examination was normal except an extensive soft tissue left sided facial mass (Plaxiform neuroma) and a faint cafe u lait spot (2 cm x 3 cm) on Back. Her parents never turned up and their address was found fake as well. As the face of the baby was affected extensively therefore her parents decided to abandon her which is a very unusual phenomenon in this society. This child was adopted by a social worker and she underwent stage corrective surgery later on.

DISCUSSION

Neurofibromatosis (NF) is the result of an abnormal differentiation and migration of neural crest during early phases of embryogenesis.[1,8] The diagnostic criteria for NF-1 and NF-2 are described Table-1. NF-1 is very common with an incidence of 1/4000.

Clinical Criteria for the diagnosis of neurofibromatosis

Neurofibromatosis Type-1

If any two of the following signs are present

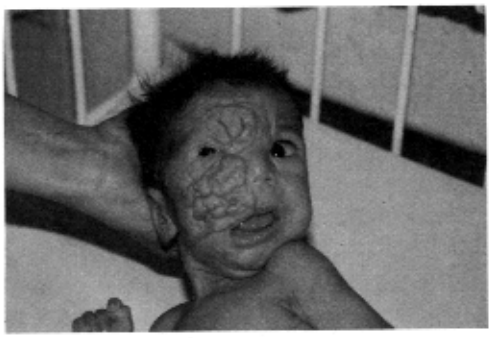
1. Six or more café-an-lait macules ver 5mm
2. Axillary or inguinal freckling
3. Two or more Iris Lish nodules
4. Two or more neurofibromas or on plexiform neuroma
5. A distinctive osseous lesion e.g. sphenoid dysplasia
6. Optic glioma
7. A first degree relative with NF-1

Neurofibromatosis Type-2

One of the following

1. Bilateral acoustic neuroma (by CT or MRI)
2. A parent, sibling or child with NF-21 with unilateral acoustic neuroma or two of following; neurofibroma, astrocytoma, neurolemmomas.

TABLE-1



Child at the age of three months

Plexiform neurofibroma (PN) is one of the special presentation of the NF-1. Plexiform Neurofibroma is specific to NF-1. These are irregular, thickened and noncircumscribed and results form diffuse thickening of nerve trunks that are frequently located in the orbital or temporal region of the face. The overlying skin may be hyperpigmented more than a Cafe-au-lait spot.[4,5]

There are a number of complication reported with plexiform neuroma per se. PN, which involve the inferior branch of the trigeminal nerve, causes disfigurement in particular. PN may compress the external auditory canal and cause compromised hearing. If plexiform neuroma is arising form the orbit in that case intracranial extension should be suspected and investigated accordingly. PN has been reported for causing compression of the celiac and mesenteric arteries and causing abdominal angina. Rapid growth of PN like other neurofibroma should suggest development of malignant change.[6,7,8] provided for a such challenging by a multidisciplinary team only.[2,6]

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