Neurofibromatosis

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Introduction

Neurofibromatosis (NF) is a genetically-inherited disorder in which the nerve tissue grows tumours (neurofibromas) that may be benign and may cause serious damage by compressing nerves and other tissues. Neurofibromatosis is an autosomal dominant disorder, which means only one copy of the affected gene is needed for the disorder to develop[1]. Neurofibromatosis-1(Von Recklinghausen diseases) occurs following the mutation of neurofibromin 1 on chromosome 17q11[2]. It is characterized by multiple café au lait spots, axillary and inguinal freckling, multiple cutaneous neurofibromas, and iris Lisch nodules[3]. Learning disabilities are present in at least 50% of individuals with NF-1. Less common but potentially more serious manifestations include plexiform neurofibromas, optic nerve and other central nervous system gliomas, malignant peripheral nerve sheath tumours, scoliosis, tibial dysplasia, and vasculopathy[4]. We report a baby who was admitted with us in view of prematurity (34 weeks gestation) and low birth weight (1.32 Kg). Baby's mother was antenatally diagnosed with NF-1(Fig 1,2). Baby had multiple café au lait spots all over the bodies (Fig 3,4). Baby was discharged from nursery in well condition. Because there is no cure for the condition itself, the only therapy for patients with neurofibromatosis is a program of treatment by a team of specialists to manage symptoms or complications.

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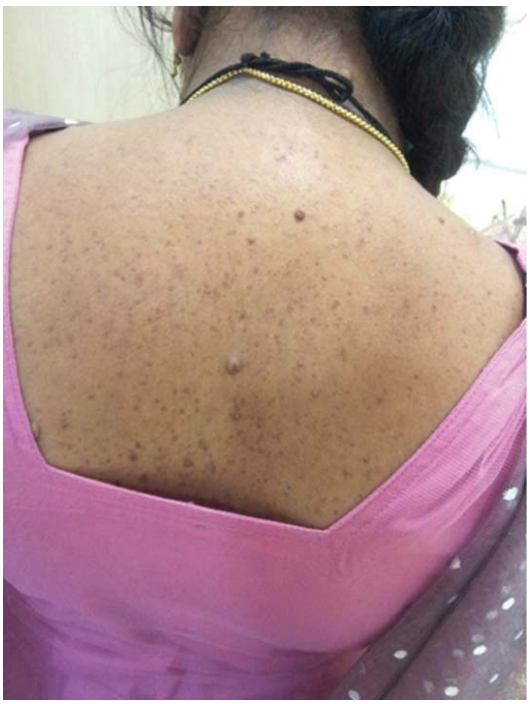

Fig 1: Showing Neurofibroma in mother

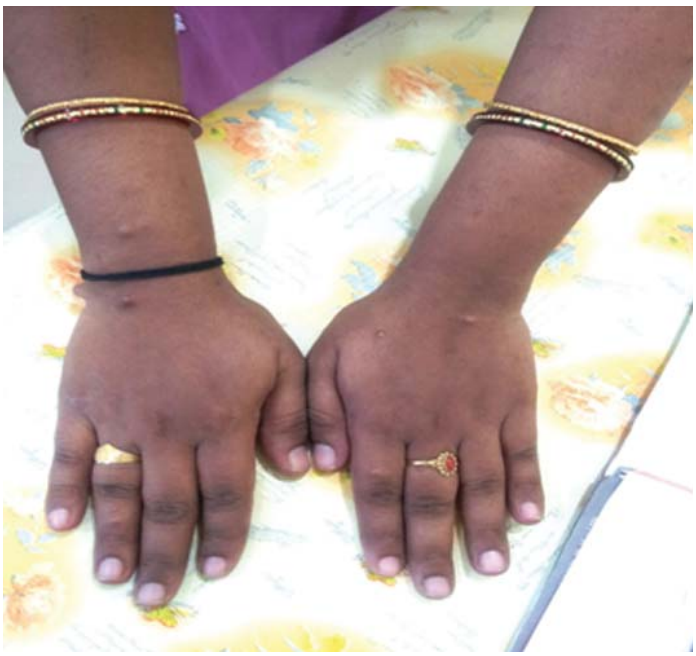

Fig 2: Showing Neurofibroma of hands in mother

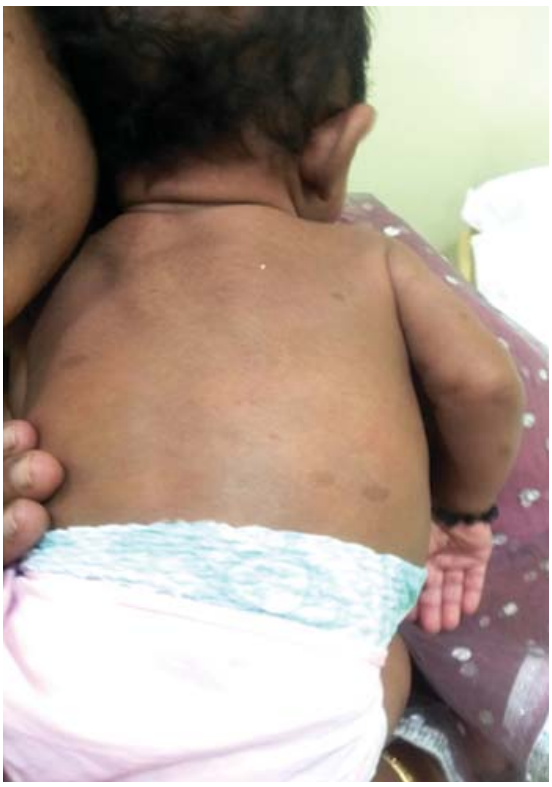

Fig 3: Showing Café au laits spots on baby back

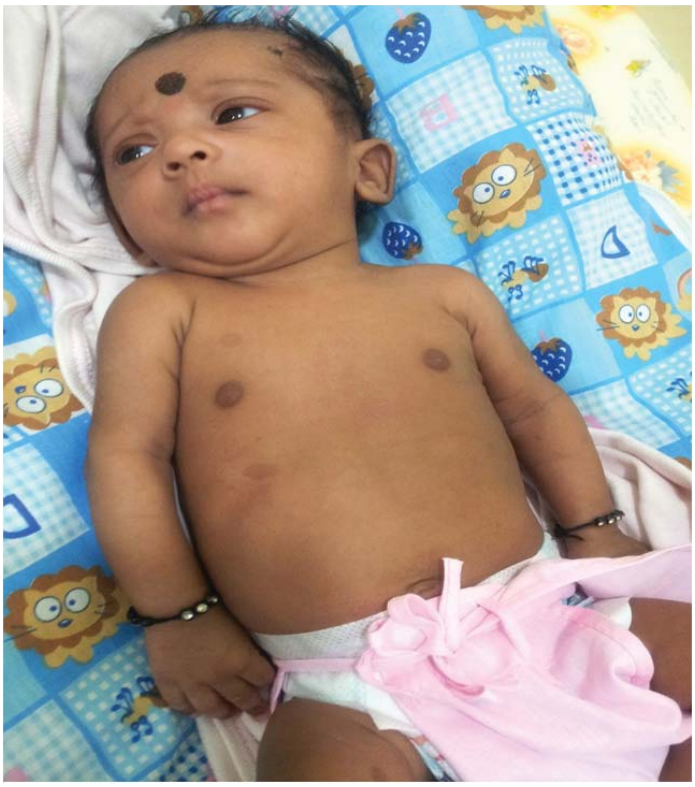

Fig 4: Showing Café au lait spots on baby abdomen

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