

# Neurofibromatosis Type I (von Recklinghausen) Presenting as Lower Limb Hemi hypertrophy : A Case Report

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## ABSTRACT

Neurofibromatosis type I is a rare genetic disorder with variable manifestations. We present a very unique case of this disorder with primarily skeletal manifestations resulting in lengthening of one lower limb and a limping gait.

Keywords: Neurofibromatosis type I; Hemihypertrophy; Café-au-lait spots

## INTRODUCTION

Neurofibromatosis type I (NF I) is an autosomal dominant genetic disorder with an incidence of approximately 1 in 2600 to 1 in 3000 individuals [1]. Approximately one-half of the cases are familial, the remainder are new mutations [2,3]. For reasons that are not well understood, the de novo mutations occur primarily in paternally derived chromosomes [4]. The neurofibromatosis type I gene has been mapped to chromosome 17q11.2 and cloned [5, 6]. Neurofibromatosis type I results in a variety of abnormalities of variable severity. To make the clinical diagnosis two or more of following are required [7]:

- 6 cafe au lait spots evident during 1 year
- two or more neurofibromas or one plexiform neurofibroma
- optic nerve glioma
- distinctive osseous lesion
- sphenoid wing dysplasia
- two or more iris hamartomas (Lisch nodules)
- axillary or inguinal freckling
- primary relative with NF 1 with above criteria

The orthopedic complications of neurofibromatosis, which usually appear early, include spinal deformities such as scoliosis and kyphoscoliosis, congenital bowing and pseudoarthrosis of the tibia and the forearm, overgrowth phenomenon of the extremity, and soft tissue tumors [8, 9]. In view of the rarity of

neurofibromatosis type I presenting as lower limb hemi hypertrophy, a case with such features is found worth reporting.

## CASE REPORT

An eight year old boy presented to the out-patient department(OPD) of Orthopaedics & traumatology Unit A, Lady Reading Hospital Peshawar, KPK, Pakistan with limping of right lower limb since 3 years of age. His parents noted lengthening of right lower limb, increase size of the leg and Coffee-coloured spots on his limbs, abdomen and back.. He had fracture of radius and ulna 1 year ago and treated in plaster of paris cast. His birth history, developmental history and family history were normal. On examination the right lower limb was externally rotated with limb length discrepancy. There was hypertrophy of the right leg and ankle, Café au lait spot on right thigh (more than 5mm in diameter), right heel valgus with flat foot. The left lower limb was normal (except Café au lait spot on thigh).We found a large spot on right wrist and forearm with swelling (neurofibroma). Spine examination was normal except multiple cafe au lait spots on the back. The patient was examined by ophthalmologist and otolaryngologist and eyes and ears of the patient were normal (no optic glioma/ Lisch nodules on the iris, no acoustic neuroma) The right lower limb was 67cm and left lower limb was 61cm in length (6cm discrepancy lower limb). Both

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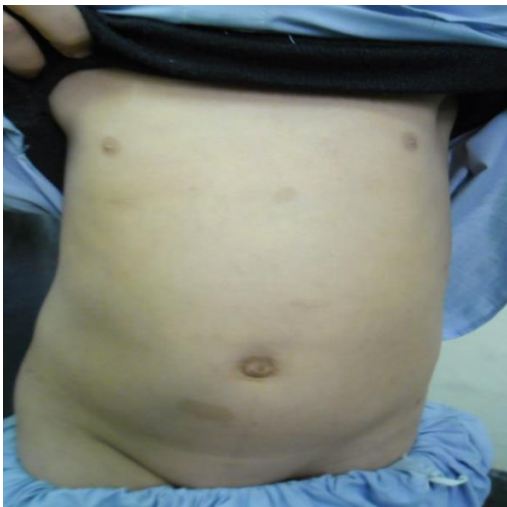
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femurs were 36cm in length. The right tibia was 37cm while left tibia was 31cm in length and was 6cm longer than the left one. Both the thighs were equal in circumference(29cm). Right leg circumference was 20cm while left leg circumference was 17cm. At ankle level the right leg was 4cm greater in circumference than the left. All relevant laboratory investigations were normal. Abdominal ultrasound was normal. X-ray chest and spine were normal. Radiographs of the limbs showed anterolateral bowing and hypertrophy of right tibia.

The parents were counseled about the disease and its management and requested for regular follow up. The child was given a special shoe raise on the normal side to compensate for the contralateral lengthening and a foot arch support and tibial splint on the disease side to correct foot deformity and prevent fracture tibia.

**Figure 1:** Photograph showing Café-au-lait spots on abdomen of the patient



**Figure 2:** Photograph showing Café-au-lait spots on the back of the patient



Epiphysodesis of the distal femur on the affected side was planned at appropriate age (probably 10-12 years) to overcome further lengthening of the limb. As the neurofibroma was symptomless at presentation, regular follow up was advised.

## DISCUSSION

Approximately 2% of individuals with neurofibromatosis type I develop bowing of the long bones, particularly the tibia and/or pseudarthrosis (a false joint in a long bone) [10]. Infants should be examined for the presence of bowing of the long bone and clinicians should consider the possibility of pseudarthrosis when assessing young children for possible non accidental injury. Scoliosis affects 10% to 26% of patients with neurofibromatosis type I, and children should undergo yearly spinal examination [11]. There is an 8–13% lifetime risk of developing malignant peripheral nerve sheath tumours in neurofibromatosis type I, predominantly in individuals aged 20-35 years [12]. These cancers are hard to detect, metastasize widely and often augur a poor prognosis [13]. The mainstay of management is age specific monitoring of disease manifestations and patient education. At all ages it is possible that severe disease complications such as malignant peripheral nerve sheath tumors will become symptomatic between appointments.

Neurofibromatosis type I individuals need to be encouraged to seek review of any unusual symptoms and ask if they are related to neurofibromatosis type I. All children with uncomplicated disease need to be assessed once a year [14]. The current management of neurofibromatosis type I focuses on genetic counseling and symptomatic treatment of specific complications. Despite early encouraging results from potential pharmacologic and biological-based therapies, new modes of therapeutic development are needed to move the field forward [15].

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**Figure 3:** Photograph showing neurofibroma right forearm



**Figure 4:** Photographs showing right lower limb hypertrophy and heel valgus



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**Figure 5:** Xray right tibia and fibula showing anterolateral bowing

