Case Report

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# Neurofibromatosis Type 1 (NF1): Case Report and Review of literature

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## Abstract

Neurofibromatosis type 1 (NF1) or Von Recklinghausen's disease is a rare genetic disorder characterized by the development of multiple noncancerous (benign) tumors of nerves and skin (neurofibromas) and areas of abnormally decreased or increased coloration of the skin. We present a case of 11-year-old boy with NF-1. The disease started in childhood with the appearance of multiple hyper pigmented skin macules. The boy presents excruciating lower back pain and lumbar scoliosis. The diagnosis NF-1 was made according to the presence of two or more diagnostic criteria of the National Institute of Health Consensus Development Conference. NF1 is a multisystem disorder requiring management by multiple disciplines, often coordinated through a primary care physician or a geneticist and dermatologist.

**Keywords:** Neurofibromatosis type 1; Recklinghausen's disease; Café-au-lait macules

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## Introduction

Neurofibromatosis type 1 (NF1) or Von Recklinghausen's disease is a rare genetic disorder characterized by the development of multiple noncancerous (benign) tumors of nerves and skin (neurofibromas) and areas of abnormally decreased or increased coloration of the skin. The earliest historical evidence first appeared in the 13th Century, but it was not until Friedrich Daniel von Recklinghausen published his landmark paper in 1882 [1]. Beginning in early childhood, patients with neurofibromatosis type 1 have multiple café-au-lait spots, which are flat patches on the skin that are darker than the surrounding area. These spots increase in size and number as the individual grows older. Freckles in the underarms and groin typically develop later in childhood.

## Case Report

### History

An 11-year-old boy is referred to our pediatric surgery department. The disease started in childhood with the appearance of multiple hyper pigmented skin macules. The boy presents excruciating lower back pain and lumbar scoliosis.

### Physical examination

Dermatological status: Hundreds of hyper pigmented skin macules on the chest and back; multiple café-au-lait spots with diameter >1.5 cm **(Figure 1)**; axillary and inguinal freckling **(Figure 2)**. The mucous membranes were not affected.

## Ophthalmological Status

Lisch's nodules on the iris of both eyes were without clinical visual involvement.

### Lab and imaging studies, histologic findings and consultations

The standard laboratory tests values were in the Normal range. X-ray photography was within the normal too. CT scan of spine and lumbar region: Lumbar scoliosis with an abscess of the left psoas, brain MRI without anomalies. Lumbar MRI: Neurofibroma left psoas muscle, with multidirectional extension with lumbar scoliosis and mega dural sheath. The neurologist did not detect alterations in the Central and peripheral nervous system. According to the otologist the acoustic nerve has not been damaged. The histological result confirmed the diagnosis of Neurofibromatosis (Psoas biopsy).

### Diagnosis

The diagnosis NF-1 was made according to the presence of two or more diagnostic criteria of the National Institute of Health Consensus Development Conference (**Table 1**) [2]:

a) Six or more café-au-lait spots measuring at least 5 mm before puberty or 15 mm after puberty.

 

**Table 1** Diagnostic criteria of the National Institute of Health Consensus Development Conference.

| Clinical diagnostic criteria for neurofibromatosis-1 (NF1) |
|---|
| Patients have two or more of the following symptoms:<br>a) 6 or more café-au-lait macules (>0.5 cm in children of >1.5 cm in adults)<br>b) 2 or more cutaneous or subcutaneous neurofibromas or one plexiform neurofibroma<br>c) Axillary or groin freckling<br>d) Optic glioma<br>e) 2 or more Lisch nodules (iris hamartomas visualized on slit lamp examination)<br>f) Sphenoid wing dysplasia or bowing of long bone (with or without pseudarthrosis)<br>g) First degree relative with an NF1 diagnosis |

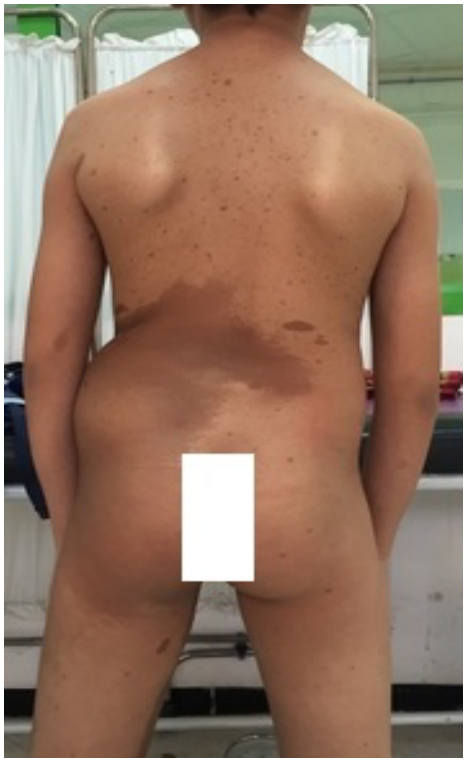

**Figure 1** Hyper pigmented skin macules with multiple café-au-lait spots.

b) Axillary or inguinal freckling.
c) Two or more Lisch's nodule.

## Discussion

The symptoms of neurofibromatosis have been observed for a long time before Robert William Smith described it in 1849 [3]. The classic variety is the one which German pathologist, Friedrich Daniel von Recklinghausen reported, who precisely described the diverse findings as a single entity in 1882; so the condition is often directed to as von Recklinghausen's disease. Von Recklinghausen's neurofibromatosis (NF-1) is inherited in an autosomal-dominant fashion and has a prevalence of between 1 per 3000 and 1 per 5000 live births thus being one of the most common autosomal-dominant conditions in humans [4].

In childhood or early adolescence, the characteristic clinical features would be apparent. NF-1 which is the most common type accounts for 90% of cases and is characterized by multiple café-au-lait spots and the incidence of neurofibromas along peripheral nerves [5]. Café-au-lait discoloration generally emerge before the development of neurofibromas and their amounts increase with age [6]. Cutaneous neurofibromas are soft, fleshy -pink colored tumors, most enormous on the trunk and limbs as sessile or dome-shaped masses, other clinical aspects contain Lisch's nodules (melanocytic pigmented iris hamartomas) and oral lesions.

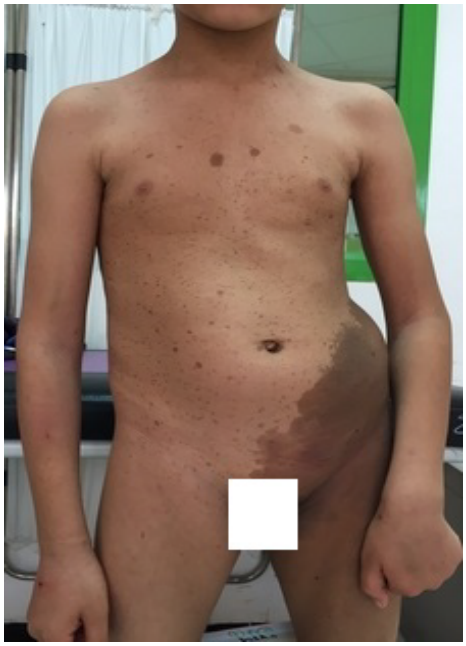

**Figure 2** Axillary and inguinal freckling.

Neurofibromatosis type 1 (NF1) represents a major risk factor for development of malignancy, particularly malignant peripheral nerve sheath tumors, optic gliomas, other gliomas, and leukemias, malignancy is an important component of the NF1 phenotype, and one of the few life-threatening complications [7].

Most children with NF1 followed a regular education, over one third of patients needed support, such as language therapy, extra-curricular educational support, or psychological support, Most parents reported learning difficulties, writing problems, and attention difficulties. Learning difficulties are the main concern of parents, and are generally present in 50% of cases [8]. There is a comorbidity of motor and cognitive deficit in developmental disorders. The principal complication during childhood is learning disabilities [8].

Predominance of visuospatial and motor difficulties in children with NF should be considered in rehabilitation programs aiming to reduce the learning and school difficulties of these children. Parental reports of specific difficulties should be considered independently of clinical severity and neuropsychological tests [9].

The patient, presented here, was referred to compatible center for specialized care (Cincinnati Neurofibromatosis Center). He did not respond to our calls for following up his treatment procedure.

## Conclusion

NF1 is a multisystem disorder requiring management by multiple disciplines, often coordinated through a primary care physician or a geneticist. The dermatologist has a role not only in the diagnosis of NF1 and differentiating it from other similar disorders but also in the recognition of rare but associated skin manifestations. There is a paucity of available medical treatments, but ongoing trials hold promise in treating both the cutaneous and non-cutaneous manifestations of NF1.

## Conflict of Interest

There is no conflict of interest among the authors.

## Funding Statement

There is no funding for the case report.

## Ethical Approval

Necessary approval was taken from the Institution and the patients for carrying out this work.

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