IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)
e-ISSN: 2279-0853, p-ISSN: 2279-0861.Volume 24, Issue 6 Ser.4 (June 2025), PP 38-41
www.iosrjournals.org

Neurofibromatosis Type 1 Presenting with Spinal Deformity and Cutaneous Neurofibromas in a Middle-Aged Woman from Rural Bihar: A Clinico-Social Perspective

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Abstract
Background:
Neurofibromatosis Type 1 (NF1) is a complex and often misunderstood genetic disorder that presents with distinct skin features and a variety of systemic manifestations. In rural and resource-limited settings, such conditions frequently go unrecognized due to limited awareness and poor access to specialized healthcare.
Case Presentation:
We present the case of a 42-year-old woman from a remote village in Bihar, who had silently endured persistent back pain and noticeable skin changes for several years. These symptoms, though significant, had never been medically investigated. Upon clinical evaluation at our center, she was found to have multiple café-au-lait macules, numerous cutaneous neurofibromas, and marked thoracolumbar scoliosis. Based on the NIH diagnostic criteria, a clinical diagnosis of NF1 was made. Unfortunately, financial constraints prevented further genetic testing or advanced imaging.
Conclusion:
This case highlights the silent burden of genetic disorders in under-resourced populations. It underscores the importance of early clinical recognition and the need for culturally sensitive health education. Equally vital is the role of compassionate communication in helping patients understand their condition and navigate its implications.
Keywords: *Neurofibromatosis Type 1, café-au-lait macules, rural healthcare, spinal deformity, clinical diagnosis*

Date of Submission: 02-06-2025 Date of Acceptance: 12-06-2025

I. Introduction

Neurofibromatosis Type 1 (NF1), first described by Friedrich von Recklinghausen, remains one of the most common neurocutaneous disorders, with a global incidence of about 1 in 3,000 live births. It follows an autosomal dominant inheritance pattern, caused by mutations in the **NF1 gene** located on chromosome 17q11.2. This gene encodes **neurofibromin**, a tumor suppressor protein that plays a crucial role in regulating cell growth. Clinically, NF1 is a syndrome of great variability—ranging from innocuous skin lesions to disfiguring tumors and serious neurological complications. Diagnostic criteria developed by the NIH have simplified clinical diagnosis in many settings. However, in resource-constrained regions like rural India, awareness of these criteria is still lacking. This case highlights not only the classical signs of NF1 but also the socio-cultural and emotional consequences of undiagnosed visible illness in a traditional community.

II. Case Presentation

Patient Background and Complaints

The patient, a 42-year-old woman from a modest household in **Samastipur, Bihar**, walked into the outpatient department with a quiet strength that belied years of discomfort. Her primary complaint was a dull, aching lower back pain that had gradually intensified over the past five years. More troubling to her, however, was the progressive curvature of her spine and the mysterious bumps that had multiplied across her skin over two decades.

She had sought help from local healers, who offered herbal pastes and spiritual explanations, but no formal diagnosis was ever given. With three children to raise and household duties to manage, she had learned to live with the pain—and the stares.

Clinical Examination

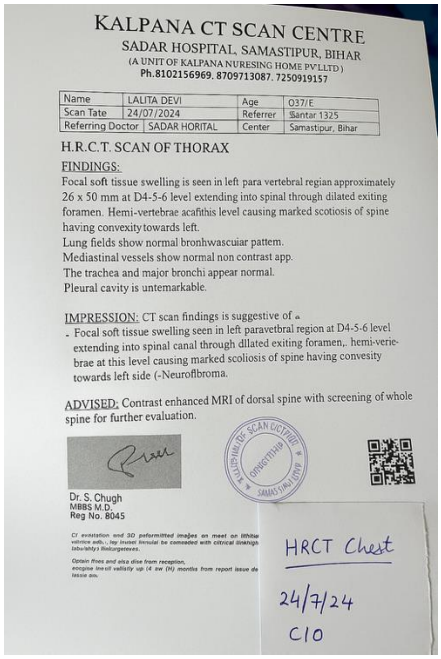
On examination, she appeared alert and cooperative. Her voice was soft, and her posture subtly protective, as if trying to hide parts of herself from the world. The following findings stood out:

- **More than 15 café-au-lait spots**, each measuring over 15 mm, were visible across her back, arms, and thighs.
- Numerous **soft, dome-shaped neurofibromas**—ranging from 0.5 to 2.5 cm—adorned her trunk and limbs.
- **Axillary freckling (Crowe's sign)** was distinctly present.
- A **left-sided thoracolumbar scoliosis** was visibly noticeable.
- **No Lisch nodules** were found on slit-lamp examination.
- Neurological assessment revealed **no motor or sensory deficits**.

She quietly confessed that she often wore extra layers, even in the harsh Bihari summer, just to avoid questions or glances from neighbors. Her children were unaware of any medical significance of her condition, and she herself believed it to be a "curse" or punishment from a previous life.

Investigations

- **CT Thorax and Spine** revealed marked **left thoracolumbar scoliosis**, with displacement around L2–L4 vertebrae. Soft tissue masses consistent with **nerve sheath tumors** were observed.
- **Routine blood work** was within normal limits.
- **MRI and genetic testing** were recommended, but declined due to financial constraints.
- Slit-lamp exam: **No ocular involvement** noted.



KALPANA CT SCAN CENTRE
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Referring Doctor	SADAR HORITAL	Center	Samastipur, Bihar

H.R.C.T. SCAN OF THORAX

FINDINGS:
Focal soft tissue swelling is seen in left para vertebral regian approximately 26 x 50 mm at D4-5-6 level extending into spinal through dilated exiting foramen. Hemi-vertebrae acafithis level causing marked scotiosis of spine having convexity towards left.
Lung fields show normal bronhwascuiar pattern.
Mediastinal vessels show normal non contrast app.
The trachea and major bronchi appear normal.
Pleural cavity is untemarkable.

IMPRESSION: CT scan findings is suggestive of -
- Focal soft tissue swelling seen in left paravetbral region at D4-5-6 level extending into spinal canal through dllated exiting foramen,. hemi-verie-brae at this level causing marked scoliosis of spine having convesity towards left side (-Neurofibroma.

ADVISED: Contrast enhanced MRI of dorsal spine with screening of whole spine for further evaluation.

Dr. S. Chugh
MBBS M.D.
Reg No. 8045

HRCT Chest
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Diagnosis
Using the **NIH diagnostic criteria**, she met at least three points:
1. More than 6 café-au-lait macules (>15 mm)
2. Multiple cutaneous neurofibromas
3. Axillary freckling

A **clinical diagnosis of NF1** was confirmed.

Management and Follow-Up
Without curative options, management focused on **relief, reassurance, and rehabilitation**:
- **NSAIDs** were prescribed for symptomatic back pain.
- Referral to **physiotherapy** was made for postural correction and spine-strengthening exercises.
- **Dermatology** confirmed benign cutaneous neurofibromas.
- **Orthopedics** recommended conservative bracing and six-month follow-up.
- Most crucially, a **psychosocial counseling session** was held.

The counselor explained to her—in her language, with empathy—that her condition was genetic, not contagious or divine punishment. A look of relief crossed her face. For the first time, she understood what was happening to her body.

Educational material in Hindi was shared with her and her family, encouraging regular check-ups, monitoring of symptoms, and early screening for her children.

III. Discussion

1. Clinical Delay and Variability
Despite visible signs, this woman lived over two decades without a diagnosis. Her case reflects a pervasive lack of awareness, both at the community level and sometimes among frontline health workers.

2. Psychosocial Impact
Stigma surrounding disfigurement can be as painful as the condition itself. Her avoidance of public spaces and reluctance to discuss her symptoms were evidence of a deep emotional toll.

3. Financial and Structural Barriers
Advanced diagnostics and genetic testing remain out of reach for many rural patients. In such contexts, clinical diagnostic acumen becomes a powerful tool.

4. The Power of Empathy
Perhaps the most transformative moment for this patient was not medical, but human—a conversation that reframed her understanding and restored a measure of self-worth.

IV. Conclusion

This case sheds light on the silent suffering of patients with NF1 in rural India. It calls upon clinicians not only to diagnose but also to educate, advocate, and comfort. NF1, while incurable, need not be unendurable—especially when dignity and understanding become part of the treatment plan.

References

[1]. Wang ZC, et al. *Community-boosted neurofibromatosis research in China*. Lancet Neurol. 2022;21(9):773–774.
[2]. Acar S, et al. *Systematic Review of Clinical Trials in Neurofibromatoses*. Pediatr Neurol. 2022;134:1–6.
[3]. Friedman JM. *Epidemiology of NF1*. Am J Med Genet. 1999;89(1):1–6.
[4]. Ferner RE. *Neurofibromatosis in the 21st Century*. Lancet Neurol. 2007;6(4):340–351.
[5]. Korf BR. *Diagnosis in Children with Café-au-lait Spots*. Pediatrics. 1992;90(6):924–927.