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CASE REPORT

Routine Excision, Rare Diagnosis: Solitary Neurofibroma Prompting NF1 Screening in an Adolescent

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Abstract: A solitary plexiform neurofibroma in a 15-year-old girl prompted an unexpected referral for neurofibromatosis type 1 (NF1) evaluation. Initially excised under the impression of a lipoma, the lesion's histopathology revealed neural origin features with strong S100 and CD34 positivity. Dermatologic examination uncovered multiple café-au-lait macules, and subsequent referral to neurology raised clinical suspicion for NF1. This case emphasizes how incidental histological findings in a benign-appearing lesion can serve as the first clinical clue of a genetic disorder, triggering appropriate multidisciplinary evaluation.

Plain Language Summary: A 15-year-old girl had a small, soft lump on her upper back that looked like a harmless fatty growth (lipoma). After removing it through a minor surgery, doctors found that it was actually a type of nerve tumor called a neurofibroma. This discovery led to further evaluations that showed she might have a genetic condition called neurofibromatosis type 1 (NF1), even though she had no other obvious symptoms. This case shows how something that seems simple can be the first clue to a bigger health issue and why it's important to look deeper when the results are unexpected.

Keywords: neurofibromatosis type 1, plexiform neurofibroma, solitary neurofibroma, genetic screening, adolescent, histopathology

Introduction

Neurofibromatosis type 1 (NF1) is a common autosomal dominant neurocutaneous disorder characterized by cutaneous, neurological, and skeletal manifestations. Though often diagnosed in early childhood, some individuals may present with subtle or overlooked signs, delaying recognition. Plexiform neurofibromas are considered pathognomonic for NF1, but may occasionally present in isolation. Identifying such tumors incidentally through histology can prompt genetic evaluation and early diagnosis of NF1, offering an opportunity for timely surveillance and family counseling.[1–3]

Case Presentation

A 15-year-old female presented with a 3×3 cm soft, mobile, painless upper back mass noted for one year. It was excised under local anesthesia due to cosmetic concerns. Histopathology confirmed plexiform neurofibroma, with characteristic spindle cell features (Figures 1–4) and supportive S100 and CD34 positivity (Figures 5 and 6).

Following histologic diagnosis, the patient was re-evaluated, revealing multiple café-au-lait macules. There was no personal or family history of NF1. She was referred to dermatology and neurology for NF1 evaluation and genetic counseling was initiated.

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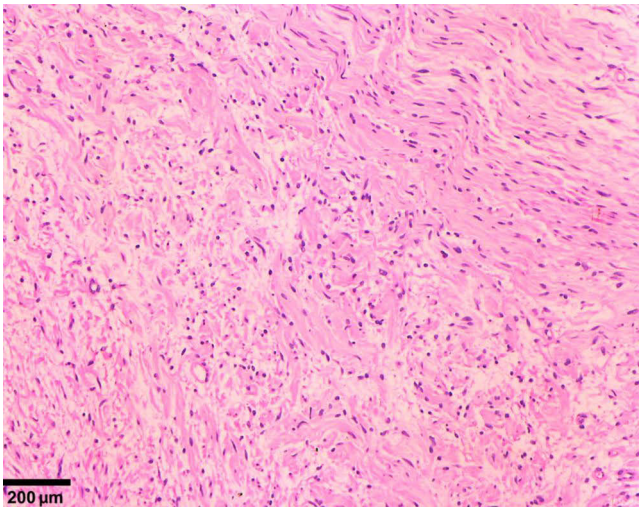

Figure 1 Low power magnification (10X) showing a moderately cellular spindle cell lesion with focal edematous stroma.

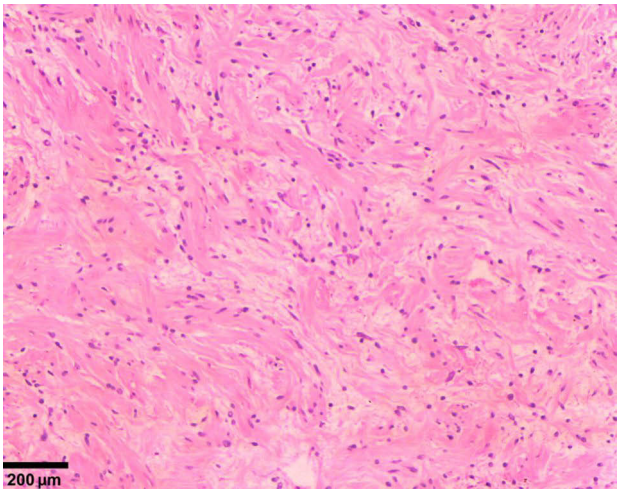

Figure 2 Low power magnification (10X) with another view of spindle cell areas.

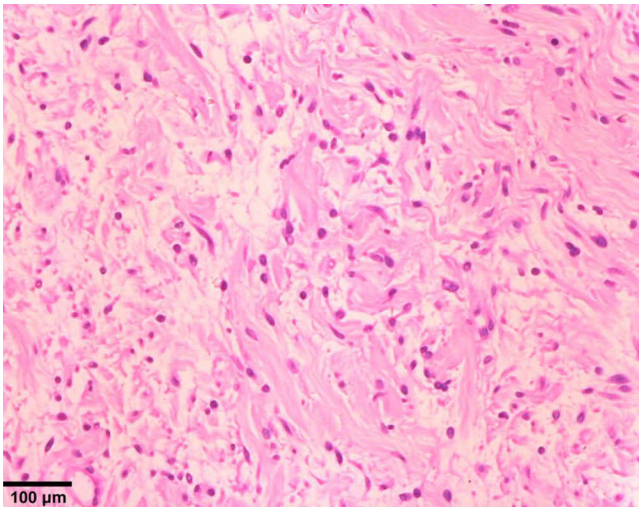

Figure 3 High power magnification (20X) showing spindle cells with wavy nuclei and eosinophilic cytoplasm.

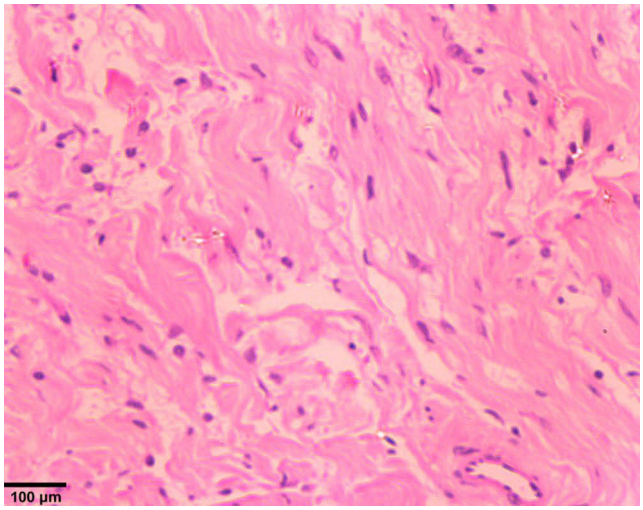

Figure 4 High power magnification (20X) confirming no atypia or necrosis.

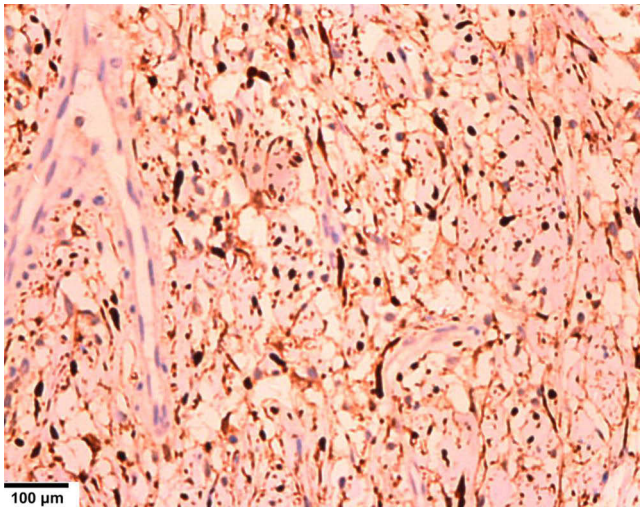

Figure 5 S100 immunohistochemistry showing diffuse nuclear positivity.

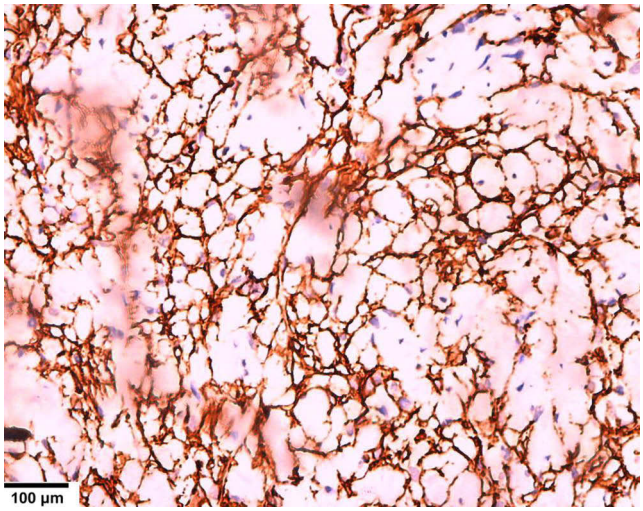

Figure 6 CD34 immunohistochemistry demonstrating a lattice-like network positivity.

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Discussion

This case illustrates how a routine excision of a lesion presumed to be benign led to the first clinical suspicion of NF1. The incidental histological finding of a plexiform neurofibroma in a child should prompt dermatological and neurological assessment for NF1, especially in the presence of pigmentary skin changes. While genetic testing was pending at the time of report, the presence of both a plexiform neurofibroma and café-au-lait macules is sufficient to meet clinical criteria for NF1.

Early identification is crucial in NF1, as complications may include optic gliomas, learning disabilities, skeletal abnormalities, and increased tumor risk. Early referral allows for proactive monitoring and intervention.[4,5]

Conclusion

Solitary plexiform neurofibroma can be an early or even the first clinical indicator of NF1. This case reinforces the importance of correlating unexpected histological diagnoses with dermatologic and neurologic signs, prompting timely referral and surveillance.

Data Sharing Statement

All data generated or analyzed during this study are included in this published article. Further inquiries can be directed to the corresponding author.

Ethics and Consent Statements

Institutional review board approval was not required for publication of this case report. Informed consent for publication was obtained from the patient's guardian.

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Disclosure

The author declares no conflicts of interest in this work.

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