MedPeer Publishers
Empowering Evidence-Based Medicine Through Peer-Reviewed Excellence

Open Access Original Research Article

MedPeer Publisher
Abbreviated Key Title: MedPeer
ISSN : 3066-2737
homepage: https://www.medpeerpublishers.com

The Skin Was Only the Beginning: An Incidental Neurofibroma Uncovers a Silent Pheochromocytoma

DOI: 10.70780/medpeer.000QGT6

AUTHORS AND AFFILIATION

Mehdi Salmane[1], Alia Yassine Kassab[1], Lina Lasri[1], Omar El Aoufir[1], Leila Jroundi[1]

Emergency department of Radiology, Avicenne Hospital, Rabat.

Corresponding author : Mehdi Salmane.

ABSTRACT

Neurofibromatosis Type 1 (NF1) is a multisystem genetic disorder characterized by cutaneous neurofibromas and an increased risk of internal tumors, including pheochromocytoma. We report the case of a 19-year-old male patient presenting with multiple cutaneous neurofibromas and episodic adrenergic symptoms, including headaches, palpitations, and intermittent hypertension. Abdominal ultrasound revealed an unsuspected heterogeneous right adrenal mass, highly suggestive of pheochromocytoma, later confirmed by biochemical testing. The coexistence of visible cutaneous neurofibromas and a deep adrenal lesion underscored an underlying syndromic association, transforming a seemingly superficial dermatologic presentation into the diagnosis of a potentially life-threatening endocrine tumor. This case highlights the pivotal role of ultrasound as an initial, accessible imaging modality capable of revealing occult adrenal pathology and triggering timely diagnosis in patients with NF1.

KEYWORDS :

Neurofibromatosis type 1, Pheochromocytoma, Neurofibroma, Adrenal mass, Ultrasound.

MAIN ARTICLE

INTRODUCTION

Neurofibromatosis Type 1 (NF1) is a common autosomal dominant disorder caused by mutations in the NF1 gene. It is characterized by multiple café-au-lait macules, cutaneous neurofibromas, axillary freckling, and a predisposition to various benign and malignant tumors [1,2].

Pheochromocytoma is a rare neuroendocrine tumor arising from chromaffin cells of the adrenal medulla. Although uncommon in the general population, its prevalence is increased in patients with hereditary syndromes including NF1 [2,3].

Early diagnosis is crucial because untreated pheochromocytoma may lead to life-threatening hypertensive crises and cardiovascular complications [3].

CLINICAL PRESENTATION

A 19-year-old male patient with a history of multiple cutaneous neurofibromas presented with recurrent episodes of headache, palpitations, excessive sweating, and intermittent hypertension.

Physical examination revealed numerous soft cutaneous nodules distributed over the trunk and upper limbs, compatible with neurofibromas.

Given the clinical suspicion of a catecholamine-secreting tumor, abdominal ultrasonography was performed.

Ultrasound Findings

Soft-Tissue Ultrasound

Ultrasound evaluation of a palpable cutaneous lesion demonstrated:

- A well-defined hypoechoic soft-tissue mass.
- Homogeneous echotexture.
- Fusiform morphology following the course of a peripheral nerve.
- Findings compatible with a neurofibroma.

Abdominal Ultrasound

Abdominal sonography revealed:

- A well-circumscribed heterogeneous right adrenal mass.
- Mixed echogenicity with predominantly hypoechoic components.
- Mild internal heterogeneity suggestive of cystic or hemorrhagic degeneration.
- No evidence of local invasion on ultrasound examination.

The lesion was highly suspicious for pheochromocytoma in the context of NF1.

Subsequent biochemical investigations confirmed elevated catecholamine levels.

DISCUSSION

Neurofibromatosis Type 1 is associated with an increased risk of several neoplasms, including pheochromocytoma [1,2]. The prevalence of pheochromocytoma among patients with NF1 is estimated to be higher than in the general population, particularly in symptomatic individuals presenting with hypertension or adrenergic symptoms [3,4].

Ultrasound is often the first imaging modality performed in patients with abdominal symptoms or suspected adrenal pathology. Although CT and MRI are generally used for definitive characterization and staging, ultrasound may provide the initial clue leading to diagnosis [4].

In the present case, the coexistence of a sonographically proven neurofibroma and a suprarenal mass strongly suggested an underlying syndromic association. Recognition of this relationship was essential in guiding further biochemical and radiological investigations.

The differential diagnosis of an adrenal mass includes:

- Adrenal adenoma
- Adrenal cortical carcinoma
- Ganglioneuroma
- Metastatic disease
- Pheochromocytoma

The presence of adrenergic symptoms in a patient with NF1 strongly favored pheochromocytoma [3,5].

Management generally involves preoperative alpha-adrenergic blockade followed by surgical excision of the adrenal tumor.

CONCLUSION

This case illustrates the important association between neurofibromatosis type 1 and pheochromocytoma. Ultrasound played a pivotal role in identifying both a peripheral neurofibroma and a suspicious adrenal mass, leading to the diagnosis of a syndromic tumor association. Awareness of this relationship is essential for early diagnosis and prevention of potentially severe cardiovascular complications.

FIGURES:

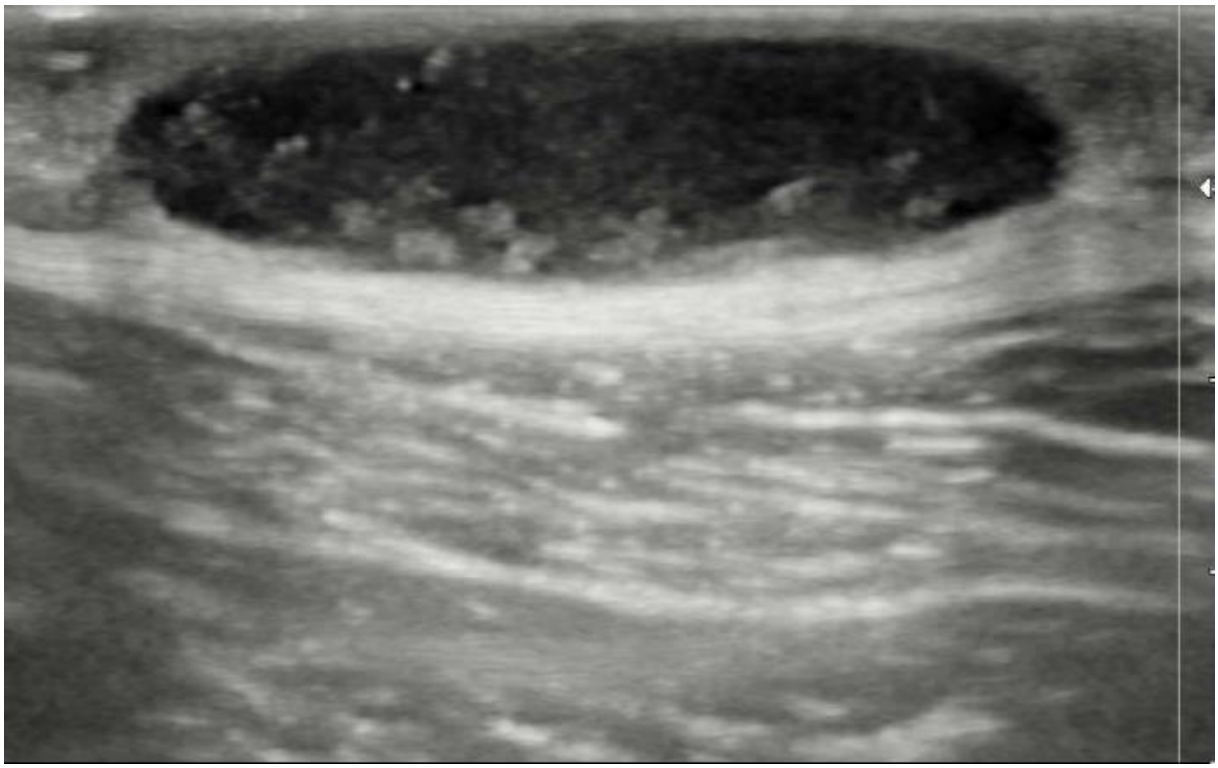

Figure 1: Ultrasound image demonstrating a well-defined hypoechoic soft-tissue lesion compatible with a neurofibroma.

Figure 2: Abdominal ultrasound showing a heterogeneous right adrenal mass suggestive of pheochromocytoma.

Figure 3: Ultrasound image illustrating the heterogeneous internal architecture of the adrenal lesion.

ACKNOWLEDGEMENTS

The authors would like to acknowledge the radiology and endocrinology departments involved in the diagnostic evaluation and management of this patient.

REFERENCES

1] Legius E, Messiaen L, Wolkenstein P, et al. Revised diagnostic criteria for Neurofibromatosis Type 1 and Legius syndrome. Genet Med. 2021;23(8):1506-1513. https://doi.org/10.1038/s41436-021-01170-5

[2] Miller DT, Freedenberg D, Schorry E, et al. Health supervision for children and adults with Neurofibromatosis Type 1. Genet Med. 2023;25(3):100338.

[3] Fishbein L, Nathanson KL. Pheochromocytoma and paraganglioma: genetics, diagnosis, and management. Hematol Oncol Clin North Am. 2022;36(4):739-754.

[4] Taïeb D, Timmers HJLM, Hindié E, et al. Current approaches and recent developments in the management of pheochromocytoma and paraganglioma. Lancet Diabetes Endocrinol. 2023;11(8):582-596.

[5] Lenders JWM, Kerstens MN, Amar L, et al. Genetics, diagnosis, and management of pheochromocytoma and paraganglioma in hereditary syndromes. Endocr Rev. 2024;45(1):1-38.