Case Report

PHEOCHROMOCYTOMA WITH NEUROFIBROMATOSIS TYPE 1

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ABSTRACT:
Patients with early onset hypertension should be evaluated for secondary causes so that the underlying pathology can be ruled out effectively, we present a case of 25 years old female having pheochromocytoma with neurofibromatosis type 1. The patient presented in OPD with a complaint of episodic hypertension with palpitations and tremors for 7 months, the patient had this problem for almost 6 to 7 years. The patient was admitted to the hospital, and examination and initial workup were done leading to a diagnosis of pheochromocytoma. The patient's episodic complaint was treated with anti- hypertensive medications.

Keywords: pheochromocytoma, neurofibromatosis, antihypertensive medication.

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INTRODUCTION

Pheochromocytomas are rare Neuro-endocrine tumors occurring in about 2 to 8 out of every one million people that produce catecholamines. Approximately 80% of these tumors occur in the adrenal medulla (phaeochromocytomas), while 20% arise elsewhere in the body in the sympathetic ganglia (paragangliomas).[1] Mostly present as benign lesions others approximately 15% show malignant features. Around 40% are associated with inherited disorders, including neurofibromatosis, von Hippel–Lindau syndrome and sometimes as a part of MEN 2.1 Only 27 cases have been reported in Pakistan in the past two decades. Significantly 7 cases in 2021 and 4 in 2022. These tumors usually cause paroxysmal significantly 7 cases in 2021 and 4 in 2022. These tumors usually cause paroxysmal hypertension sometimes acute, and can also lead to serious health problems including stroke, and myocardial infarction.[2]

My patient started having paroxysmal episodes of headache, Palpitations, facial pallor and sweating off and on 5 years back. These were associated with severe abdominal pain, nausea and sometimes vomiting. She was found to have very high readings during these episodes. The episodes were relieved by oral antihypertensive and sometimes required IV antihypertensive treatment by local paramedics. She remained asymptomatic for a couple of years, only 2 episodes were experienced by her in the past 2 years after which she started having more frequent and severe episodes since the start of 2023. She also complains of a large raised firm tender nodule on the posterior aspect of the middle third of her left arm, measuring approximately 15cm x 12cm. It was initially small and gradually increased in size over the past 10 years.

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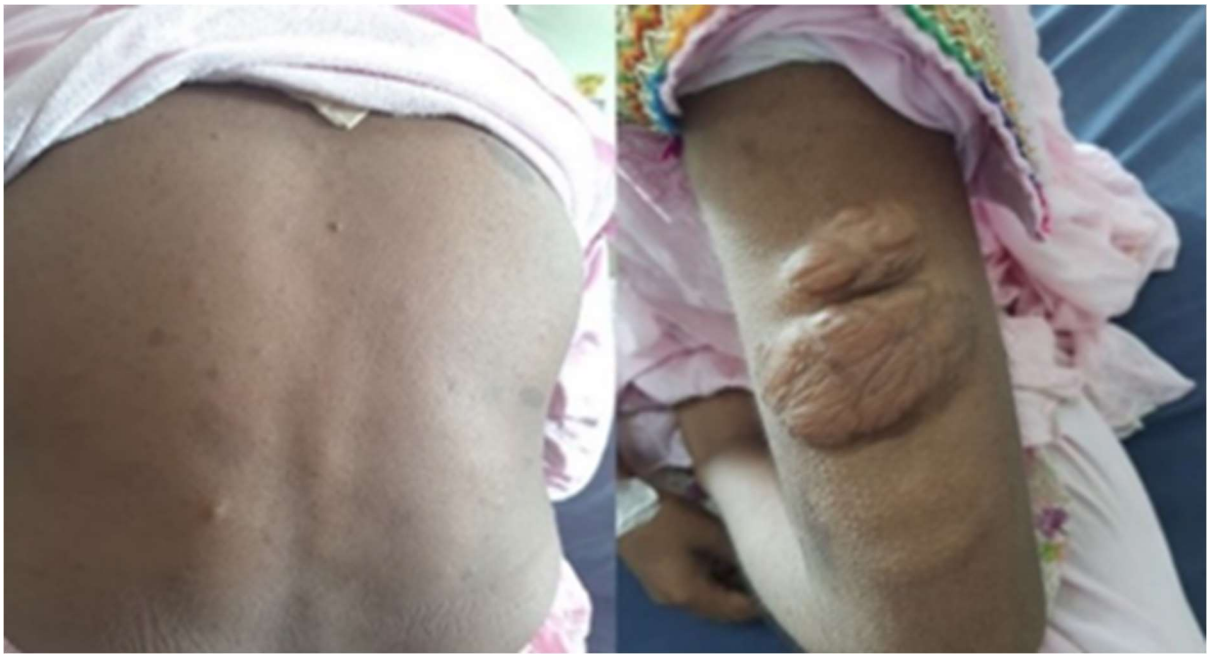

Fig 1: showing café au lait macules and small neurofibromas present on the back of the patient. A large neurofibroma is present in the middle of the left arm.

Small nodular lesions were also noticed on her face, arm and back, and hyper pigmented macules (café au lait) were present in the upper and middle back as well as her left arm as shown above. Freckles were present in axillary areas bilaterally. Her mother had the same nodular lesions on her body, she died a few months back due to some unexplained cause at the age of 55 years according to the patient. Her brother also has the same nodular lesions and hyper pigmented macules on his entire body as shown below. But he never**.**

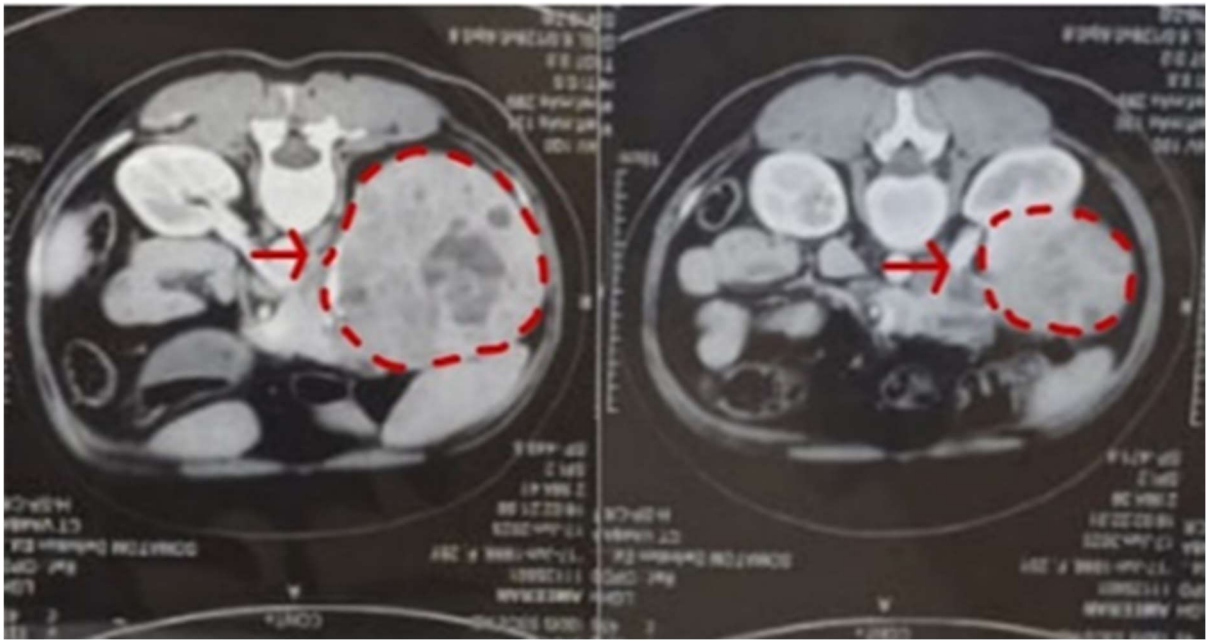

Fig 2: showing neurofibromas on different body parts of the brother of the patient.

Experienced symptoms like palpitations, and episodic hypertension, signifying tumors can be asymptomatic.[3] On abdominal ultrasound a large complex retroperitoneal mass was seen in the right lumbar region with echogenic foci and thin septations, Doppler showed minimal vascularity. It appeared to be originating in the anterior pole of the right kidney, a differential of pheochromocytoma was suggested by the radiologist. On computed tomography scan of the abdomen and pelvis(shown below), there was a 10x11x11 cm well-defined solid lesion in the right supra renal retroperitoneal location, the contralateral adrenal gland appeared to be unremarkable it was demonstrating internal foci of necrosis, compressing upon the right lobe of the liver. The lesion was also compressing upon and displacing the right kidney in the caudal direction, mass effect was also noted on IVC, The Liver was unremarkable for any focal lesions abdominopelvic vasculature was patent. There was no significant lymphadenopathy. The visualized skeleton was unremarkable for any osseous lesions.

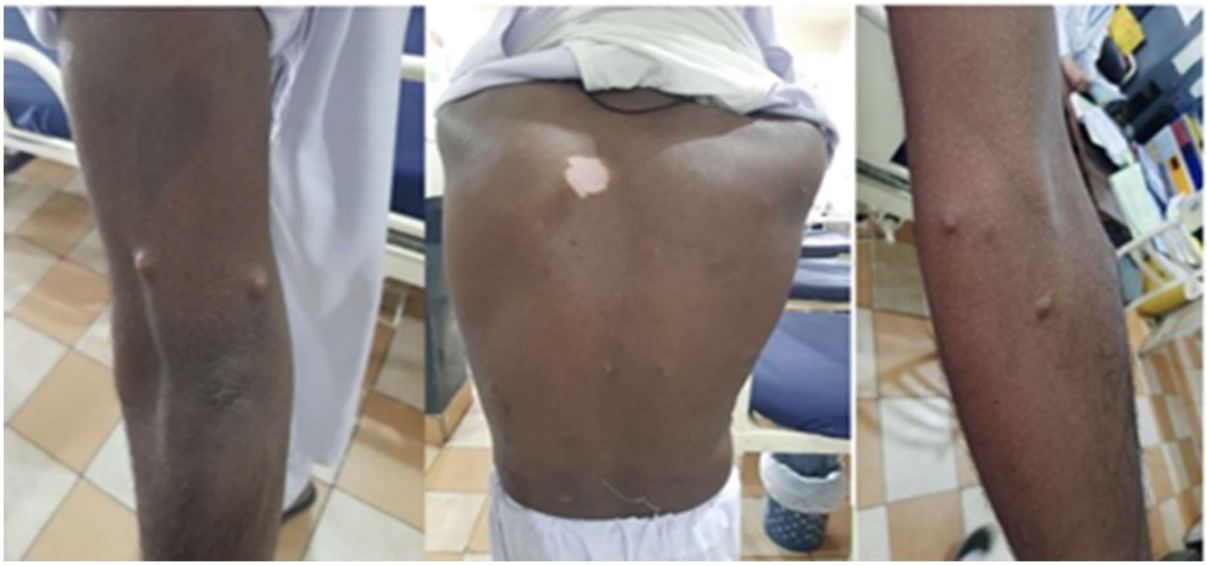

Fig 3: CT scan of the abdomen showing mas in retroperitonal location. Compressing surrounding visceras.

24 hours urinary metanephrine levels were raised significantly to 1859.78 micrograms/day (N=350 microgram/ day). The patient required stat doses of inj. labetalol 10mg to control episodes of hypertensive urgency. She was managed by controlling the Blood pressure with alpha and beta blockers. She was initially started on the tab. Doxazosin 4mg 1 x PO x OD. After 2 days tab. Carvedilol 6.25mg 1x Po x BD was added to her treatment regimen. Her Blood pressure was monitored 6 hours daily. On surgical evaluation by a consultant surgical Oncologist Laparoscopic Adrenalectomy was advised but unfortunately despite extensive counselling the patient was reluctant to get an invasive procedure and opted to be managed medically for symptomatic relief of her paroxysmal episodes of headache and palpitations only, hence she was discharged on alpha blockers and beta blockers therapy and was advised weekly follow up. The patient was followed every week henceforth in opd for 2 months where she reported no further symptoms since discharge indicating that her symptoms were adequately controlled with medical intervention, the patient remains in close follow up regarding her condition.

DISCUSSION

This case presents a unique clinical scenario of a young female patient with a rare combination of pheochromocytoma and neurofibromatosis type 1.[4] The coexistence of these two conditions poses diagnostic challenges and underscores the importance of considering secondary causes in cases of early-onset hypertension. The patient's initial presentation of episodic hypertension, palpitations, and tremors, lasting for 7 months and recurring over the past 6 to 7 years, prompted further investigation.[5] The identification of a large retroperitoneal mass on imaging, coupled with significantly elevated urinary metanephrine levels, led to the diagnosis of pheochromocytoma. This neuroendocrine tumor, originating from the adrenal medulla, is known for its potential to produce catecholamines, resulting in episodic symptoms of hypertension. Of particular interest in this case is the concomitant presence of neurofibromatosis type 1, a genetic disorder characterized by nodular lesions, hyper pigmented macules, and café au lait spots. The patient's family history revealed a similar condition in her mother and brother, adding a familial aspect to the case.[6] The mother's unexplained death at the age of 55 raises questions about the potential impact of these conditions on overall health and lifespan. The decision-making process regarding the management of this patient's pheochromocytoma is noteworthy. While laparoscopic adrenalectomy was recommended, the patient opted for medical management due to concerns about undergoing an invasive procedure. This decision emphasizes the importance of patient preferences and the need for individualized approaches in the management of complex cases. The successful control of the patient's symptoms with alpha and beta blockers highlights the efficacy of medical intervention in certain cases of pheochromocytoma. The rarity of this case, especially in the context of neurofibromatosis type 1, contributes to the existing body of knowledge on the manifestations of genetic disorders and their intersection with rare endocrine tumors. In conclusion, the interplay between pheochromocytoma and neurofibromatosis type 1, along with the challenges in decision-making and management, adds a layer of complexity to this clinical scenario, emphasizing the need for a multidisciplinary and patient-centred approach.

CONCLUSION

This case highlights the rare co-occurrence of pheochromocytoma and neurofibromatosis type 1, presenting with episodic hypertension. The patient's familial history adds complexity, emphasizing the importance of genetic factors. Medical management effectively controlled symptoms, showcasing the role of tailored interventions. The patient's preference for non invasive treatment underscores the need for individualized care. This case contributes valuable insights into the intricate interplay of genetic disorders and rare endocr intumors, emphasizing the significance of a multidisciplinary approach.

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CONFLICTS OF INTEREST

The authors declare that they have no competing interests.

AUTHOR'S CONTRIBUTION

ZI: Introduction & review
SI: Abstract, manuscript writing
HA: Case history, bibliography & review
AYY: Discussion writing & conclusion

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