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Case Report
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# Hereditary Paraganglioma as a Cause of Secondary Hypertension: A Case Study

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***Abstract***

***Introduction:*** *Paragangliomas are rare neuroendocrine tumors originating from extra-adrenal paraganglia that secrete catecholamines. Clinically, patients present with headaches, sweating, tachycardia, and can be a cause of secondary hypertension. While most cases are sporadic, approximately one-third are associated with hereditary syndromes, involving mutations in genes encoding the succinate dehydrogenase (SDH) enzyme.*

***Case Description:*** *A 29-year-old male, former smoker (10 pack-years), with obesity and childhood-onset hypertension, without relevant family history, presented to a primary care setting with a severe headache, dizziness, vomiting, diaphoresis, and an average blood pressure (BP) of 180/100 mmHg, despite being on lisinopril 20 mg + amlodipine 5 mg once daily. On physical examination, he had a BMI of 31, central obesity, tachycardia, and a BP of 160/95 mmHg. Further testing revealed significantly elevated levels of renin, normetanephrine, and norepinephrine. Imaging studies identified two solid masses, one anterior to the abdominal aorta and one sacral, suggestive of paraganglioma, with a confirmed SDH mutation on genetic testing. The patient underwent laparotomy for paraganglioma excision and has since maintained a BP of 120/80 mmHg without the need for antihypertensive medication.*

***Conclusion:*** *This case reinforces the fundamental role of family physicians in recognizing atypical clinical presentations and initiating timely, targeted investigations. Their central position in coordinating care allows for early referral, diagnostic accuracy, and multidisciplinary management—particularly in rare but treatable conditions. In this case, a prolonged diagnostic delay contributed to avoidable morbidity and impacted both the patient's quality of life and family well-being, highlighting the need for increased clinical awareness of secondary causes of hypertension in young adults.*

**Keywords:** Paraganglioma, Secondary Hypertension, SDHB Mutation, Hereditary Tumors, Case Report, Family Medicine.

## Introduction

Paragangliomas are rare neuroendocrine tumors that arise from extra-adrenal paraganglionic tissue. When functional, paragangliomas can produce excessive catecholamines—especially norepinephrine and epinephrine. Clinically, they may manifest with secondary hypertension (HTN), headaches, diaphoresis, and tachycardia. [1, 2].

Although many paragangliomas are sporadic, approximately one-third are hereditary, most frequently involving mutations in the succinate dehydrogenase (SDH) gene complex. (Fishbein, 2016; Amar et al., 2005) [3, 4]. Among these, SDHB mutations are particularly significant due to their strong association with metastatic potential [5]. Early genetic diagnosis is crucial for both therapeutic planning and familial risk assessment. SDHB-related paragangliomas often arise in the abdomen, including the para-aortic region, particularly near the organ of Zuckerkandl [3]. Due to their rarity and the non-specific nature of clinical presentation, paragangliomas are often diagnosed late. Many patients undergo years of empirical treatment for hypertension before a

definitive cause is identified [2]. This case reinforces the critical role of primary care physicians in recognizing atypical features in young patients with treatment-resistant hypertension, initiating appropriate investigations, and coordinating timely referrals.

Here, we describe the case of a 29-year-old man with longstanding hypertension ultimately diagnosed with hereditary paraganglioma due to an SDHB gene mutation. The case underscores the importance of high clinical suspicion and multidisciplinary management in rare but treatable conditions.

## Case Description

### Identification

A 29-year-old male, former smoker (10 pack-years), presented with a long-standing history of arterial hypertension (HTN). He had no relevant family history, denied alcohol or drug use, and had no history of physical activity. His medical history included grade I obesity since childhood (BMI 31 kg/m²).

### Chronological Timeline of Events

At age 14, he presented with fatigue on minimal exertion and resting precordial discomfort, though he denied headaches, vision changes, diaphoresis, or palpitations. The diagnosis of primary HTN confirmed through 24-hour ambulatory blood pressure monitoring (ABPM). Lifestyle modification was recommended.

At age 18, he remained hypertensive and began pharmacological treatment with lisinopril 20 mg and amlodipine 5 mg, achieving partial blood pressure control (120–130/80–90 mmHg). Over the following years, he continued therapy without major complications.

At age 26, the patient presented with new-onset holocranial pressure-like headaches, dizziness, visual disturbances, and hypertensive episodes, including post-prandial and post-coital nausea and vomiting. Ambulatory readings showed persistent BP >180/100 mmHg despite regular therapy. He sought care at the emergency department with a hypertensive crisis of 300/100 mmHg, intense headache, diaphoresis, tremors, and palpitations. He was treated with intravenous antihypertensives and underwent a Computed Tomography (CT) angiography and cervical vessel CT, which revealed no significant findings, and was subsequently discharged.

At age 28, the patient initiated regular follow-up in Primary Health Care. Home blood pressure monitoring (HBPM) showed consistent readings of approximately 180/100 mmHg, despite appropriate medication adherence. The clinical presentation, long-standing history, and poor therapeutic response raised suspicion for secondary hypertension. A full diagnostic investigation was initiated, including hormonal assays and abdominal imaging, detailed in the following section.

### Investigation and Diagnosis

In 2022, given the patient's persistent and treatment-resistant hypertension, a comprehensive diagnostic workup was initiated in Primary Health Care. Routine blood tests revealed normal renal function, liver enzymes, and electrolytes. However, hormonal assessment showed significantly elevated plasma renin (428.8 mU/mL) and epinephrine (>2400 pg/mL), along with abnormal levels of norepinephrine and normetanephrine. Aldosterone levels were within normal range. (Table 1)

**Table 1:** Table of diagnostic test results in 2022

| Test | Findings |
|---|---|
| Laboratory tests | Renin: 428.80 mU/mL • Epinephrine: >2400 pg/mL |
| ECG and Echocardiogram | Normal |
| Thyroid and Abdominal ultrasound | Normal |
| Renal and adrenal ultrasound | Heterogeneous vascularized pre-aortic solid mass (72×83×38 mm), highly suggestive of paraganglioma |
| Renal artery Doppler | Normal |

Renal and adrenal ultrasound showed a highly vascularized retroperitoneal mass anterior to the abdominal aorta, measuring approximately 72×83×38 mm, with well-defined borders and heterogeneous texture. No abnormalities were found in the kidneys or adrenal glands. A second solid mass was later detected in the presacral region.

Doppler ultrasound of the renal arteries showing no evidence of stenosis or altered flow indices. A thyroid ultrasound showed a small bilateral cystic nodule without suspicious features. Abdominal ultrasound revealed moderate hepatic steatosis but no focal lesions or biliary abnormalities. Given the clinical presentation and the initial biochemical and imaging findings suggestive of a catecholamine-secreting tumor, the patient was referred to the Endocrinology Department for further evaluation and management. Upon reassessment, all relevant biochemical parameters were repeated, and the results confirmed persistent abnormalities: plasma renin, epinephrine, norepinephrine, and normetanephrine levels remained markedly elevated, consistently above the reference ranges, supporting the suspected diagnosis. (Table 2)

**Table 2:** Table of diagnostic test results in 2023: laboratory findings, CT scan and MRI

| Test | Findings |
|---|---|
| Laboratory tests | Renin: 425 mU/mL; Epinephrine: >2400 pg/mL; Norepinephrine: 1561 pg/mL; Normetanephrine: 30× above reference value; |
| Abdominopelvic CT with contrast | Hypervascular pre-aortic solid mass (73×75×61 mm) suggestive of paraganglioma; second presacral solid similar mass (37×45×46 mm); |
| Abdominopelvic MRI with contrast | indicated in the image caption |

Further characterization by contrast-enhanced Computed Tomography (CT) confirmed two distinct retroperitoneal masses: one pre-aortic (73×75×61 mm) and one presacral (37×45×46 mm) (Table 2). Magnetic Resonance Imaging (MRI) showed both lesions to be hyperintense on T2-weighted images, with heterogeneous enhancement, central cystic areas, and no signs of invasion. Their location was consistent with paragangliomas, particularly in the region of the organ of Zuckerkandl. (Image 1)

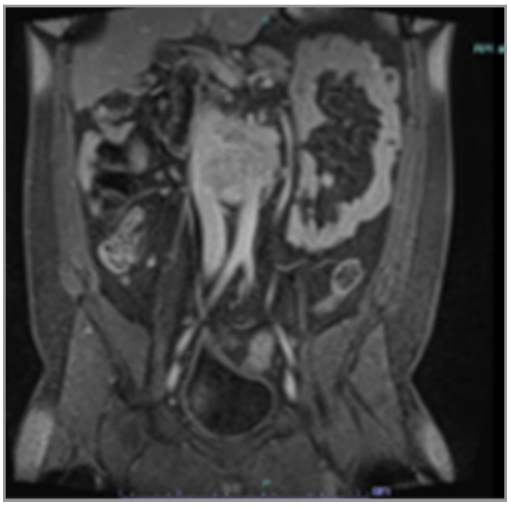

**Image 1:** Abdominopelvic MRI with contrast in 2023: "Two nodular lesions previously described on CT, with marked heterogeneous T2 signal; located at the organ of Zuckerkandl, likely paragangliomas" (image included with informed consent from the patient)

To confirm neuroendocrine activity, a Positron Emission Tomography (PET) scan using [68Ga] Ga-DOTANOC was performed, showing intense somatostatin receptor expression in both masses and in a para-aortic lymph node. No distant metastases were identified. (Table 3)

**Table 3:** Table of diagnostic test results in 2023: PET scan and genetic testing

| Test | Findings |
|---|---|
| PET Scan | Somatostatin receptor overexpression in two large masses (pre-aortic and presacral) and in a left celiac lymph node, confirming their neuroendocrine nature |
| Genetic Testing | SDHB mutation confirmed – heterozygous deletion of exon 1 of the SDHB gene |

Given the patient's age and imaging pattern, genetic testing was conducted, revealing a heterozygous deletion of exon 1 of the SDHB gene, consistent with Hereditary Paraganglioma Syndrome. The diagnosis of functional, hereditary, multiple paragangliomas was thus established.

**Treatment and Follow-up**

In 2024, following the biochemical confirmation of a catecholamine-secreting tumor and genetic diagnosis of SDHB-related Hereditary Paraganglioma Syndrome, the patient was referred to a Specialized Oncology Center (Instituto Português de Oncologia – IPO). Preoperative optimization included the initiation of alpha-adrenergic blockade with phenoxybenzamine, progressively titrated to therapeutic effect to reduce perioperative cardiovascular risk.

The patient underwent surgical excision of both retroperitoneal masses. At the time of discharge, the patient's blood pressure was stable (120/80 mmHg) without the need for any antihypertensive medication — a significant improvement after over a decade of pharmacological treatment. Long-term surveillance is maintained both at the Portuguese Oncology Institute (IPO) and

in Primary Health Care, with a focus on hereditary paraganglioma syndrome and associated comorbidities.

## Conclusion

This case illustrates the crucial role of Family Physicians in recognizing and managing rare causes of secondary hypertension in young patients with early-onset and treatment-resistant blood pressure elevation. Despite more than a decade of empirical antihypertensive therapy, the underlying etiology—a functional hereditary paraganglioma—remained undiagnosed until a systematic evaluation was performed in Primary Health Care.

The diagnostic process required a multidisciplinary approach, integrating biochemical testing, advanced imaging, and genetic analysis. The identification of an SDHB gene mutation not only confirmed the diagnosis but also prompted appropriate surgical management and long-term oncological surveillance.

Primary care physicians play a pivotal role in the early recognition and referral of rare but clinically significant conditions. This case reinforces the value of continuity of care and comprehensive evaluation in uncovering potentially curable causes of hypertension with systemic implications for both the patient and their family.

## Ethical Considerations

This case report was prepared in accordance with international ethical standards for medical publishing. The patient's data were fully anonymized to protect confidentiality, and all identifying details were removed. Informed consent for publication was obtained from the patient. No experimental procedures were performed, and no institutional ethics committee approval was required under national guidelines for single anonymized case reports. The authors declare no conflicts of interest and received no external funding for this work.

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