Adrenal

ADRENAL CASE REPORTS

Rare Case of Huge Adrenocortical Carcinoma in Surprisingly Asymptomatic Patient

Samia Mohammad AlJedaani, MD[1], Sarah Al-Obaydi, MD[1], Aesha Shah, MD[1], Chris fan, MD[1], Andrea Manni, MD[2].
[1]PENN STATE UNIVERSITY MILTON S HERSHEY MEDICAL CENTER, Hershey, PA, USA, [2]Milton S Hershey Medical Center, Hershey, PA, USA.

Introduction: Adrenal Carcinoma (AC) is a rare endocrine malignancy of the adrenal glands. It can affect up to 3–10% of the human population. It is common in children as secondary malignancies or as a manifestation of a tumor syndrome. Typically patients with AC start to become symptomatic after extensive tumor development- 10–13 cm. The presentation of AC includes signs of hormone excess. The most common hormone in excess is cortisol. Thus individuals with AC typically present with symptoms of hypercortisolism or Cushing's Syndrome. In chronic cases the high concentration of glucocorticoids can start stimulating aldosterone receptors causing hypertension and hypokalemia. Furthermore, patients with AC also have symptoms of tumor growth, such as abdominal pain, flank pain, and early satiety. Currently prognostic factors for AC are undefined. AC generally has a poor prognosis as a majority of patients are diagnosed during advanced stage of disease (stage IV). Furthermore, it was found that patients with stage IV disease had a survival of less than one year. Currently the only curative treatment is complete tumor resection. Medical management with chemotherapy can be utilized in cases of extensive metastasis and poor prognosis. A common chemotherapeutic agent is mitotane, which is a steroidogenesis inhibitor. Case Presentation:Patient is a 19 year old female, who initially presented to her primary care provider's (PCP) office for an annual wellness visit during August 2020. She was subsequently discovered to have elevated blood pressure. At this time she was not symptomatic. She was advised on lifestyle modifications. She was also stopped on her birth control medication. Furthermore, she later presented to her PCP with continued elevated blood pressure, still not symptomatic. In an outpatient cardiology office, she presented with blood pressures in the 200s/100s range and a new murmur. She was sent to the ED for evaluation (11/2020). On her presentation to the ED, she still did not have any symptoms. She also denied any fatigue or unintentional weight changes. Her only concern was mild edema in the lower legs after standing for extended periods of time. Her medical history was insignificant. Her family history was significant for her paternal essential HTN diagnosed in the early 20s. Laboratory evaluation revealed unremarkable electrolyte levels and CBC. Furthermore, her TSH level was also normal. A chest ray revealed multiple pulmonary nodules ranging from 3–5 mm. A renal ultrasound revealed a large mass in the RUQ (16.9x9.6x12 cm). She was also found to have mildly elevated cortisol levels at 22.9, suppressed ACTH levels, and normal levels of aldosterone and metanephrines. Subsequent CT imaging and biopsy revealed adrenal carcinoma with extensive metastasis to the lungs and liver. Patient was referred to Oncology service to start Chemotherapy.

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Resistant Hypertension Due to Familial Hyperaldosteronism Type III: First Report From Indian Sub-Continent

Sarah Alam, MD DM, Alpesh Goyal, MD DM, Devasenathipathy Kandasamy, MD, Virinder Kumar Bansal, MS, Krishna Asuri, MS, Shipra Agarwal, MD, Rajesh Khadgawat, MD DM
All India Institute of Medical Sciences, New Delhi, India.

Background: Familial hyperaldosteronism type III (FH-III) is caused by germline mutations in KCNJ5 gene. FH-III presents with phenotypic variability from spironolactone-responsive hypertension to massive adrenal hyperplasia requiring bilateral adrenalectomy. Till date, seven different pathogenic mutations in KCNJ5 gene have been identified. Here we describe a sporadic FH-III case, due to a Gly151Arg mutation, first from Indian subcontinent, presenting with extremely high plasma aldosterone concentration (PAC) values, further expanding our knowledge of this rare condition. **Clinical Case:** A 24-year-old female, symptomatic since age of 5 years with periodic limb weakness and gradual increase in frequency of episodes over the years. Her blood pressure (BP) was recorded for first time at 9 years of age, and it was 170/110 mm Hg. On evaluation at this time, PAC was highly elevated at 1007 ng/dL and plasma renin activity (PRA) was suppressed at 0.04 ng/ml/h with aldosterone renin ratio (ARR) of 25,175 ng/dL per ng/mL/h (>20 suggestive of primary aldosteronism). CT scan demonstrated mild enlargement of bilateral adrenal glands. A presumptive diagnosis of Glucocorticoid-remediable aldosteronism was made. She was started on dexamethasone, spironolactone and nifedipine but was not improved. Dexamethasone was stopped after 1 year of initiation. Before presenting to our referral center in 2018, she was having uncontrolled hypertension with recurrent episodes of hypokalemic paralysis. She was on maximal doses on four antihypertensive agents, further increased to six agents (including spironolactone 100 mg BD), and potassium chloride supplementation (120 mEq/day). Despite this, she had a serum potassium of 2.6 mEq/L. Her biochemical investigations demonstrated that PAC was 2070 ng/dL, direct renin concentration (DRC) was 2.35 μIU/mL (PRA 0.2 ng/ml/h) and ARR was 880.9 ng/dL per μIU/mL. CT scan revealed massive bilateral adrenal hyperplasia. Genetic analysis by whole exome sequencing detected KCNJ5 (p.Gly151Arg) mutation, confirming the diagnosis of FH-III. She was subjected to bilateral adrenalectomy and she became normokalemic. Dramatic reduction in antihypertensives with BP control achieved only on amlodipine post-operatively. Genetic testing of family members was not done but they were normotensive and normokalemic. Histopathological examination revealed bilateral adrenal hyperplasia. PAC levels up to 297 ng/dL have been described previously in FH-III but our patient had exceedingly high-level of 2070 ng/dL. **Conclusion:** This case demonstrates florid clinical and biochemical manifestations of FH-III and gradual worsening of symptoms, consistent with progression of disease with age. It illustrates that proper investigations and treatment can lead to remission of symptoms. Further studies are warranted to elucidate the full clinical and biochemical spectrum of FH-III.