Answer: Hyponatremia

MCQ Explanation:

Hyponatremia, hyperkalemia, and hypoglycemia help with the diagnosis of CAH in infants. Genetic testing is available in the newborn screening. Insufficient therapy in adults may lead to hyponatremia and hypovolemia as in this patient.

There are other case reports of congenital adrenal hyperplasia leading to adrenal tumors of various types (1). Published articles propose that the mechanism for the disorganized adrenal growth in undertreated CAH is associated with ACTH overproduction as a response to inadequate cortisol levels (1). This patient had hyponatremia that may have been indicative of inadequate treatment of her salt-wasting CAH.

Following surgical removal of the tubo-ovarian abscess, bilateral adrenalectomy was planned, given the possible malignant nature of the masses. The adrenal glands were removed and analyzed by pathology. The pathology was concerning for malignancy with zones of necrosis, extension beyond the capsule, and lack of clear cells. Given the rarity of primary bilateral adrenal malignancies, the samples were sent to an external pathology laboratory for further analysis. The final pathology report indicated bilateral adrenocortical hyperplasia with atypia, with the need for close monitoring for metastasis, suggestive of possible adrenocortical carcinoma.

CAH is often diagnosed in infancy. However, children with CAH will eventually become patients of internal medicine physicians. This case and others suggest that undertreating this disorder in adults may be associated with adrenal tumors, such as adrenal adenomas, and even malignancies. Therefore, internal medicine physicians should be mindful of the congenital disorders of their patients, as undertreating these disorders may have profound health implications.

Reference:

1. Wang J, et al. Adrenal tumors associated with inadequately treated congenital adrenal hyperplasia. Can J Urol. 2002 June; 9(3):1563-4.