APPLIED RADIOLOGY
RADIOLOGICAL CASE

Congenital adrenal hyperplasia secondary to 3-beta hydroxysteroid dehydrogenase deficiency

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CASE SUMMARY

A 35-year-old Hispanic man presented to the emergency department with a two-day history of abdominal pain, non-bilious vomiting and acute anorexia.

The patient was previously diagnosed with hypospadias and congenital adrenal hyperplasia (CAH) but has been without treatment for 3 years, due to lack of insurance. Surgical, family and social history were non-contributory.

Review of systems was negative, including no weight loss and no fever. Physical exam was within normal limits.

IMAGING FINDINGS

Contrast-enhanced computed tomography (CECT) of the abdomen and pelvis demonstrates lobulated homogenously enhancing masses measuring up to 10 cm in the bilateral adrenal fossa (Figures 1 and 2).

DIAGNOSIS

Congenital adrenal hyperplasia secondary to 3-beta hydroxysteroid dehydrogenase deficiency

DISCUSSION

Various enzyme deficiencies are responsible for congenital adrenal hyperplasia (CAH) with 21-hydroxylase deficiency being the most frequent culprit.[1] Multiple subtypes of CAH can lead to bilateral adrenomegaly, evident on imaging which can aid in the diagnosis of unknown or untreated cases.[2] In asymptomatic or subclinical patients, the resulting adrenomegaly can predispose patients to adrenal hemorrhage or Waterhouse-Friderichsen Syndrome.[3]

Three-beta hydroxy-dehydrogenase (3-BHSD) deficiency is extremely rare with approximately 60 affected individuals reported.[1] The 3-beta hydroxyseteroid dehydrogenase (3-BHSD) enzyme is one of the most proximal enzymes essential for steroidogenesis in both the adrenal cortex and gonads. Complications in males often include hypospadias, ambiguous genitalia, and infertility.[1]

Most patients with 3-BHSD deficiency will require treatment with mineralocorticoid, glucocorticoid and androgen replacement. The level of 3-BHSD enzyme function can vary in patients with deficient enzymatic activity.[1] We postulate that our patient has residual enzyme function, as he was non-adherent to medication and asymptomatic for nearly three years. Furthermore, the imaging findings upon presentation likely represent a result of chronic adrenal stimulation and steroid precursor buildup of cholesterol, pregnenolone, 17-OH pregnenolone, and dehydroepiandrosterone (Figure 3).[4] The feared complication of CAH is adrenal crisis; a life threatening reaction to low cortisol characterized by

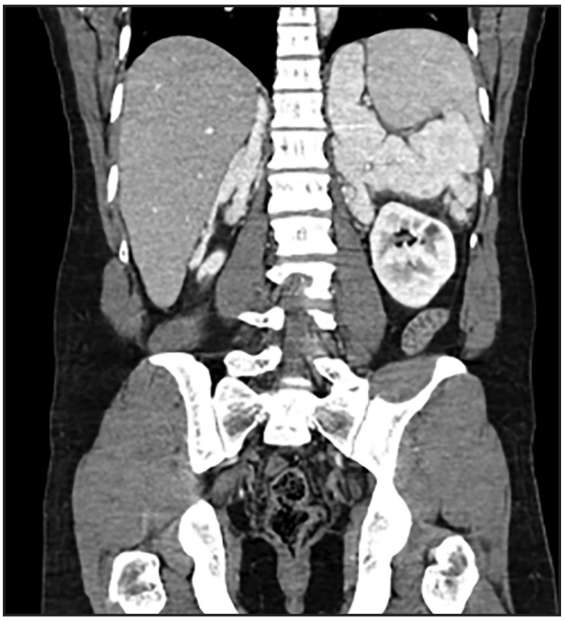

FIGURE 1. *Axial CECT. Lobulated homogenously enhancing masses within the adrenal fossa impressing on the spleen and stomach.*

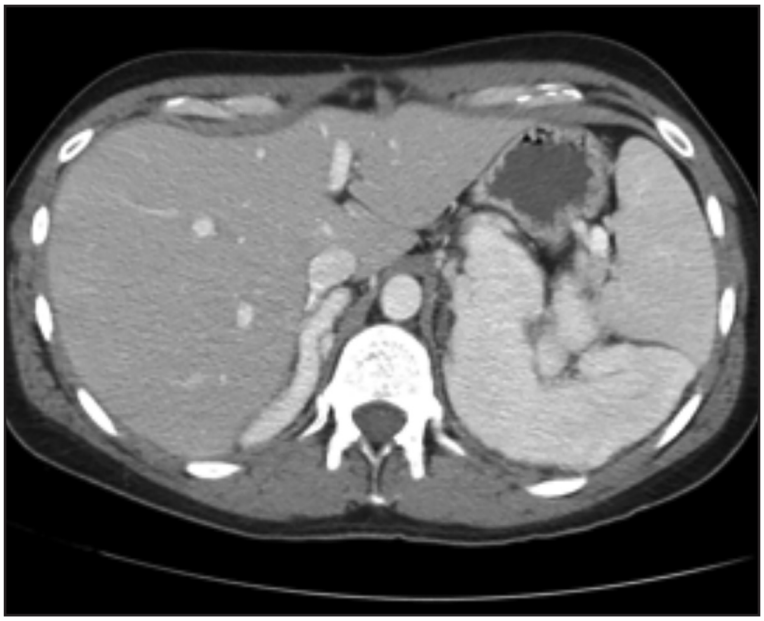

FIGURE 2. *Coronal CECT Lobulated homogenously enhancing masses within the adrenal fossa. Note the adrenal morphology.*

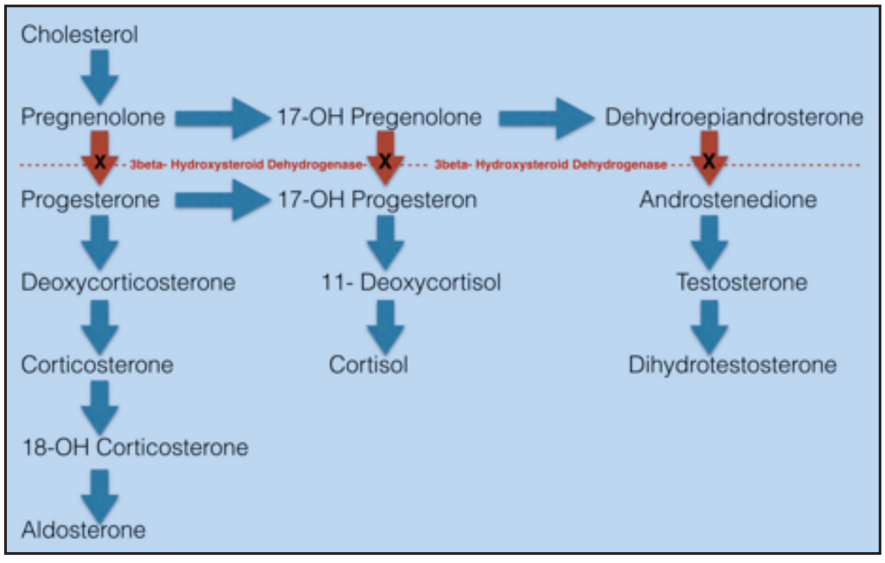

FIGURE 3. *Adrenal cortical hormone synthesis demonstrating the absent enzyme function of 3beta-Hydroxysteroid Dehydrogenase (red) and subsequent loss of downstream hormone products.*

hypoglycemia, hyponatremia and hypovolemic shock. Patients with CAH may also struggle with infertility.

CONCLUSION

Imaging findings of adrenomegaly can be seen with 3-BHSD deficiency, a subtype of CAH that leads to deficiencies in mineralocorticoid, glucocorticoid, and androgen synthesis. Although some enzyme function may persist, treatment with fludrocortisone, prednisone, and testosterone is the standard of care. Patients who are withheld treatment risk an adrenal crisis, especially when stressful life events occur.

REFERENCES

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