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Case Report

Sonography in the Diagnosis of Congenital Adrenal Hyperplasia in a Neonate

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Abstract

Congenital adrenal hyperplasia (CAH) is a relatively rare inherited disease that primarily affects the synthesis of steroid hormones by the adrenal glands. Patients usually present in the weeks following birth with salt wasting and virilization. In patients who are clinically suspected, a definitive diagnosis is established with the detection of increased levels of precursor hormones. An evaluation of the adrenal glands by ultrasonography may help make an accurate diagnosis of CAH prior to a biochemical confirmation. We report the case of a neonate with 21-hydroxylase deficiency who was diagnosed with the typical appearance of the adrenal glands for CAH revealed with ultrasonography.

Keywords: Congenital adrenal hyperplasia, 21-hydroxylase deficiency, ultrasonography

INTRODUCTION

Congenital adrenal hyperplasia (CAH) is a group of autosomal recessive disorders with an enzyme deficiency in one of the steps necessary to synthesize cortisol. The result is hyperplasia of the adrenal glands, high levels of adrenocorticotropic hormone, and increased production of the adrenal steroids that do not require the deficient enzyme. 21-Hydroxylase deficiency is the the most common, and its incidence is approximately 1 in 15,000 in the worldwide population. It results in impaired cortisol production and excess androgen production, particularly androstenedione. Rarer causes of CAH include 11-α-hydroxylase deficiency and 3-β-hydroxysteroid dehydrogenase deficiency (1).

Neonates can present with adrenal crisis having hyponatremia, hyperkalemia, and dehydration. Females with CAH demonstrate ambiguous genitalia. Symptoms of hypoglycemia, weakness, hypotension, and electrolyte abnormalities usually do not occur until 4 to 10 days of age and may not appear for as long as 4 weeks. An elevation in serum 17-OHP levels leads to making a diagnosis. We herein report the case of a newborn diagnosed as having CAH with the assistance of ultrasonography findings prior to a biochemical confirmation.

CASE REPORT

A 20-day-old male neonate presented with complaints of vomiting, poor feeding, and weight loss. A physical examination revealed dehydration, penile enlargement, and scrotal hyperpigmentation. Bilateral testes were detected in the scrotum. An urgent biochemical analysis of the blood showed hyponatremia (104 mmol/L, normal level: 135–146 mmol/L) and hyperpotassemia (8.6 mmol/L, normal level: 3.5-5 mmol/L). Bilateral enlargement of the adrenal glands was demonstrated by abdominal ultrasonography performed on having a suspicion of CAH; the length (30 mm and 36 mm for right and left adrenal glands, respectively) and the width (4.2 mm and 5 mm for right and left adrenal glands, respectively) of the wings of both adrenal glands were greater in size than that of a normal-sized neonatal adrenal gland (Figure 1). The adrenal glands had a cerebriform-wavy pattern specific for CAH (Figure 2). The diagnosis of CAH due to 21-hydroxylase deficiency was confirmed with the increased serum levels of the adrenocorticotropic hormone (212 pg/mL; normal level for prepubertal children: 7–28 pg/mL) and 17-OH progesterone (3000 ng/mL; normal levels for infants more than 24 days old: less than 1 ng/mL) (2). Subsequently, glucocorticoid and mineralocorticoid replacement therapy was initiated.

DISCUSSION

In patients with CAH, the insufficiency of serum cortisol levels with or without aldosterone deficiency and androgen excess leads to the emergence of symptoms and findings (salt wasting, ambiguous genitalia, and virilization). However, females diagnosed with CAH solely present with ambiguous genitalia and virilization. Males commonly present with adrenal crisis around 2-4 weeks following birth, and they are not diagnosed in the nursery. The definitive diagnosis of CAH is established by the identification of decreased serum levels of aldosterone and cortisol and increased serum levels of adrenocorticotropic hormone and 17-OH progesterone, which is a precursor hormone. However, achieving the aforementioned hormone levels may require up to 1 week, which may delay mak-

Figure I. a, b. Sagittal scans of the right adrenal gland acquired with a I0 MHz linear probe in neonates with (a) and without (b) CAH. Compare the enlarged adrenal gland that is coiling up the upper two-thirds of the kidney (arrows) (a) with the normal sized (arrows) (b)

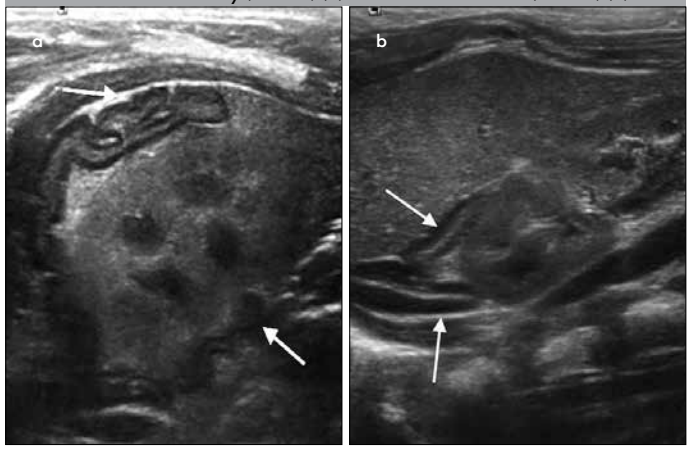
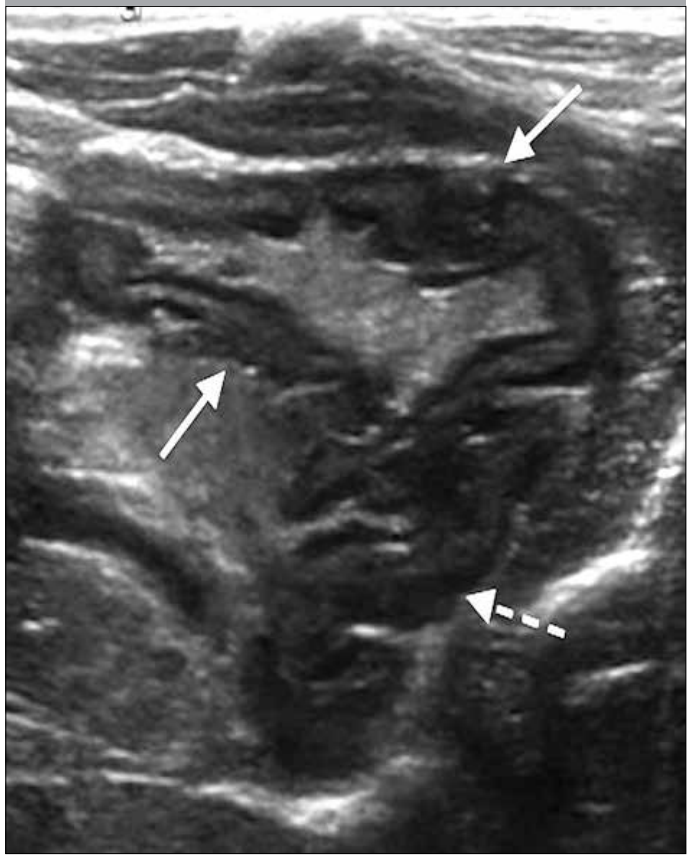


Figure 2. Axial sonographic image (I0 MHz linear probe) showing the cerebriform-wavy pattern of the left adrenal gland resembling cerebral gyri. The body (dashed arrow) and wings (arrows) of the adrenal gland are shown



ing an accurate diagnosis of CAH (3). In these circumstances, ultrasonography may play a role in diagnosing CAH prior to a biochemical confirmation (4, 5).

On imaging, adrenal glands have a "V" shape with a body and two wings. The measurement of the adrenal glands acquired by ultrasonography enables the sonographic diagnosis of CAH: a mean length of the wings of greater than 20 mm and a mean width of the wings of greater than 4 mm (4). However, normal-sized adrenal glands on ultrasonography are not sufficient for ruling out CAH. In these cases, the cerebriform pattern of the adrenal glands is reported to be specific for CAH even in the absence of enlarged adrenal glands and help establish the diagnosis (5). The wavy contours of both adrenal glands resemble cerebral gyri.

Hydrocortisone (glucocorticoid) and fludrocortisone (mineralocorticoid) replacement is the mainstay of therapy. Sick patients may require aggressive fluid and electrolyte management in addition to stress-dose hydrocortisone. Adequacy of glucocorticoid dosing can be assessed via measuring I7-OHP levels. Stress-dose steroids are needed during illness or other times of stress. Surgery may be required on female genitalia depending on the degree of virilization.

In conclusion, ultrasonography, as an easily applicable imaging modality to evaluate the adrenal glands in newborns, may have a crucial role in diagnosing CAH prior to a biochemical confirmation.

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