Anesthetic Management for Genitoplasty in a 21-hydroxylase deficient patient a case report

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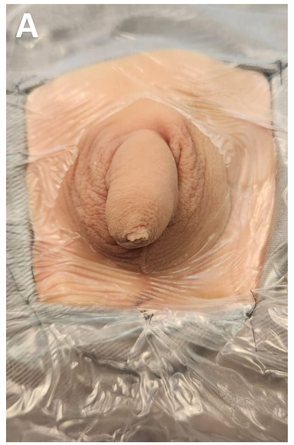
BACKGROUND

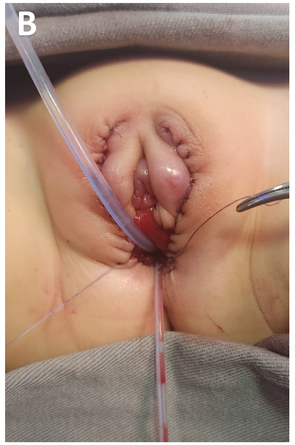
The treatment for newborns with adrenal hyperplasia, one of the most common autosomal recessive disorders, has advanced to the point of turning this patient's survival high. Mutations in the gene encoding 21-hydroxylase (*CYP21A2*) result in a lack of 21-hydroxylase, which is required for the production of cortisol and aldosterone in the adrenal cortex. This results in the overproduction of corticotropin and cortisol precursors and their diversion through the steroid pathway that produces adrenal androgens and atypical genitalia in 46XX and primal adrenal insufficiency during childhood.

CASE REPORT

A 16-month-old patient (46XX) with the classic virilizing form of congenital adrenal hyperplasia (CAH) was admitted for perineal vaginoplasty. The patient kept using fludrocortisone (150 mcg) and hydrocortisone (14.4 mg/m²) until the day of surgery as prescribed by the endocrinologist. The anesthetic technique was general anesthesia combined with a caudal epidural block with bupivacaine and morphine (30 mcg/kg) to optimize intra and postoperative analgesia. Hydrocortisone 50 mg was administered during intravenous induction and after 4 hours. Besides, strict glycemic and electrolyte control was carried out during the procedure. The post-anesthetic recovery was uneventful, and the patient was discharged from the hospital after six days.

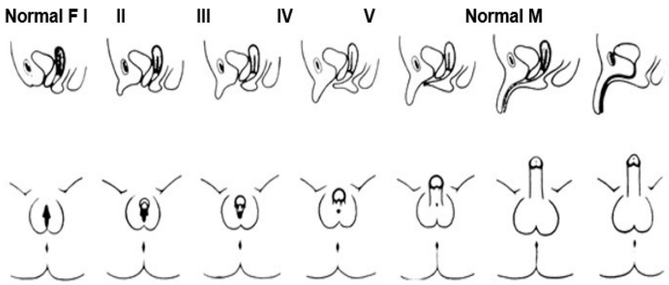


A - Before surgery
Complete posterior fusion of the labia and clitoromegalia



B - After surgery

LAB TESTS	30/03/22	06/04/22	28/09/22	25/11/22	08/03/23	14/06/23	26/07/23
Hydrocortisone	1-1-1 (15mg/m²)	1-1-1 (14,2mg/m²)	1-1-1 (9,4mg/m²)	1-1-2	2-1-2 (13mg/m²)	2-2-2 (14,4mg/m²)	2-2-2 (14,4mg/m²)
Fludrocortisone (mcg)	50	50	100	150	150	150	150
17OHP (<1,73 ng/ml)	126	55,5	163	8,9	107	8,3	198
Cortisol/ ACTH	9,3 /		/ 157	10,5 /	1,2 / 87	/ 36	<0,5
Total Testosterone (ng/dL)	1011	278	13	<15	17	<15	26
Androstenedione (<0,5 ng/mL)	13,9	3,9	0,9	<0,5		<0,5	0,8
Aldosterone (<23 ng/dL) / Renine (2,8- 39 uUI/ml)	- / 44		- / 3641	- / 76	3,5 / 76		
Sodium/ Potassium (mmol/L)	140 / 5,3	138 / 6,2	133 / 6,4	135 / 5,0		136 / 4,4	141 / 4,9



Prader scale I-V

DISCUSSION

Patients with CAH 21-hydroxylase deficiency present with elevated 17-hydroxyprogesterone due to defective conversion to 11-deoxycortisol, resulting in decreased cortisol synthesis and loss of negative feedback leading to increased production of ACTH. The adrenal stimulation increases the production of adrenal androgens, resulting in atypical genitalia characterized by clitoral enlargement, labial fusion, and the formation of a urogenital sinus in 46XX females. Anesthetic management is complex and includes glucocorticoid replacement and management of adrenal crisis, hypotension or shock, hypoglycemia, metabolic acidosis, hyponatremia, and hyperkalemia.

CONCLUSION

A multidisciplinary team should include endocrinologists. The surgery must be performed at an experienced medical center, and the anesthetic plan must include early recognition and management of adrenal crisis and glucocorticoid replacement therapy. Caudal epidural may contribute to reasonable pain control with hemodynamic stability. The primary goal is to ensure the replacement of the adrenal insufficiency of CAH in order to maintain normal plasma volume and physiological balance in the preoperative period.

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