

# Anaesthetic Concerns In A Child With 21-Hydroxylase Deficiency For A Day Care Surgery.

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## Abstract

Congenital adrenal hyperplasia due to 21-hydroxylase deficiency is one of the most common autosomal recessive hereditary diseases. The impairment of cortisol synthesis leads to excessive stimulation of adrenal gland by adrenocorticotrophic hormone (ACTH), adrenal hyperplasia and excessive androgen synthesis. More than 90% of congenital adrenal hyperplasia is caused by 21-hydroxylase deficiency which is found in 1:10,000 to 1:15,000 live births. Children with congenital adrenal hyperplasia due to 21-hydroxylase deficiency presenting for surgeries is very unusual. Here we are presenting a fifteen year old male child with classical 21-hydroxylase deficiency, who was successfully managed as a day case for elective orchidopexy under general anaesthesia. The consent for publication for the case was obtained by mother of child after surgery.

## INTRODUCTION

Congenital adrenal hyperplasia (CAH) is an autosomal recessive disorder that is associated with deficiencies in 21-, 11-, or 17-hydroxylase in children[1-2]. It can be partial (simple virilising form) or more complete (salt-losing form). At birth, children demonstrate masculinisation of external genitalia and those with complete deficiency show a progressive salt losing state. This condition usually manifests as difficulty in feeding and failure to thrive in early weeks of life. In patients with classic CAH obesity, hyperinsulinemia, insulin resistance and hyperleptinaemia is seen more often than the general population[2]. The clinical manifestations are mainly due to adrenocortical insufficiency, hyperandrogenism and adverse effects of glucocorticoids used in the treatment of this condition[2]. Patients with CAH require frequent monitoring of biochemical parameters (serum 17-hydroxy progesterone and androstenedione levels)[2-3]. The current literature examining anesthetic management of children with CAH due to 21-hydroxylase is limited to a few case reports. We describe a child with classical 21-hydroxylase deficiency on steroid treatment posted for elective orchidopexy with a review of literature.

## CASE REPORT

A 15-year-old male child weighing 43 kg with left sided undescended testis presented for elective orchidopexy. The patient was a diagnosed case of congenital adrenal

hyperplasia (CAH) due to complete 21-hydroxylase deficiency at the age of 4 years. His mother gave the history of failure to thrive since birth and hypertension from the age of four years. His medications included Tab. Fludrocortisone 0.05 mg once daily, Tab. Hydrocortisone 8 mg in the morning and 4 mg at night, and Tab. Nifedipine 5 mg twice daily, from the age of 4 years. He was withdrawn from Tab. Nifedipine three months back by his physician. The child was having regular checkups with monitoring of serum 17-hydroxy progesterone level once in every two months. The last value obtained two weeks prior to surgery was 60 ng/dl and it was within normal limits. On examination, vitals were normal and fasting status was confirmed. A systemic examination was unremarkable. The laboratory values showed haemoglobin of 11 g/dl and random blood sugar, urea and electrolytes were within normal limits. An ultrasound examination of abdomen showed a hypoplastic left kidney and a normal right kidney. It did not reveal the location of testes.

Since the patient was well controlled he was scheduled as a day care procedure after thorough discussion with his parents and the surgeon. The operating room, standard anesthesia monitoring was initiated with Electrocardiogram (ECG), Noninvasive blood pressure monitoring (NIBP) and Pulse oximetry. A 20G intravenous cannula was secured and infusion of normal saline started. Intravenous hydrocortisone succinate 75 mg was given before the induction of anaesthesia. Anaesthesia was induced

with fentanyl 75 µg, propofol 100 mg and mivacurium 4 mg. A size 3 Laryngeal mask airway was introduced and anaesthesia was maintained with sevoflurane, oxygen and air using circle system. Controlled ventilation was used to maintain  $\text{ETCO}_2$  around 35mmHg. Throughout surgery the vital parameters were stable and the surgery lasted for 45 minutes. The child received 350 ml of normal saline during surgery. The postoperative recovery was uneventful. The patient was discharged after obtaining a satisfactory Aldrete score following surgery with instructions to contact if any need arises.

### **DISCUSSION**

Congenital adrenal hyperplasia (CAH) is an autosomal recessive disorder that is associated with deficiencies in 21-, 11-, or 17-hydroxylase in children [2]. More than 90% of congenital adrenal hyperplasia is caused by 21-hydroxylase deficiency which is found in 1:10,000 to 1:15,000 live births [4]. The syndrome is characterized by a considerable correlation between genotype and phenotype with the type of CYP21A2 gene mutation affecting the severity of 21-hydroxylase deficiency [2].

It can be partial (simple virilising form) or more complete (salt losing form). Prolonged gestation is more likely in pregnancies where the fetus has the salt-wasting form of CAH. This may be due to impaired cortisol production, although other changes in steroidogenesis may also be contributory.

At birth, children demonstrate masculinization of external genitalia and those with complete deficiency show a progressive salt losing state. This condition usually manifests as difficulty in feeding and failure to thrive in early weeks of life [1]. In patients with classic CAH, obesity, hyperinsulinemia, insulin resistance and hyperleptinemia is seen more often than general population [2]. These abnormalities promote the development of metabolic syndrome and its sequelae, including endothelial dysfunction and cardiovascular disease [2]. The clinical manifestations are mainly due to adrenocortical insufficiency, hyperandrogenism and adverse effects of glucocorticoids used in the treatment of condition [2]. Non classic CAH may be sometimes asymptomatic with quite variable ranging from premature pubarche, hirsutism, acne, menstrual irregularities, infertility and spontaneous abortion [10]. Patients with CAH require frequent monitoring of biochemical parameters such as serum 17-hydroxy progesterone and androstenedione levels, to assess the

therapeutic control [11].

The diagnosis in early period of life has better prognosis. The usual presentation is failure to thrive, hyperkalemia, hyponatremia and features of hyperandrogenism [1]. An infant presenting with ventricular tachycardia and cardiac arrest due to hyperkalemia has been reported. Sudden death in such patients suggests underlying electrolyte abnormalities [8-9].

Once the diagnosis is confirmed, patient should be managed with steroids with biochemical monitoring of 17-hydroxy progesterone [5]. The clinical parameters such as growth velocity and bone age remain the gold standard for monitoring the adequacy of therapy in congenital adrenal hyperplasia [6].

The present child was diagnosed at the age of four years with complete 21-hydroxylase deficiency. Since then he was on oral steroids with proper monitoring. The patient was managed with glucocorticoids and mineralocorticoids. The only cardiovascular in this patient was hypertension which was treated with Nifedipine, which was withdrawn as the blood pressure had come down to normal range.

The anaesthetic management surrounds the complications of steroid therapy and suppression of ACTH due to long term steroid therapy. The adverse effects of long term steroids should be kept in mind while anaesthetising these patients. The most important among them are hypertension, hyperglycemia, ischaemic bone necrosis, adrenal and pituitary suppression, immune suppression and osteoporosis. Long term steroid therapy can even prolong the effect of the muscle relaxants. It is prudent to monitor blood glucose level during prolonged surgery. The patient should receive a dose of hydrocortisone succinate 2 mg/kg and repeated after four hours if duration of surgery is long [1]. In our case it was a short case and we administered a single dose of hydrocortisone succinate. Another case of clitoroplasty has been successfully managed with perioperative steroid supplementation [7].

The electrolyte abnormalities such as hyperkalemia should be addressed prior to surgery. The proper evaluation of child prior to surgery avoids most of the life threatening complications during anaesthesia. It has been proven that there is association between HLA and congenital adrenal hyperplasia [12], so while anaesthetising patients we should keep in mind conditions like ankylosing spondylitis, which may cause difficulty in airway management apart from the

obesity associated with CAH.

## SUMMARY

This case report describes successful anesthetic management of a child with classical CAH for day care surgery. Most of the case reports in the literature suggest uneventful perioperative period, giving importance to proper history and physical examination, and children with CAH can be managed successfully during anaesthesia after understanding the pathophysiology of disease with proper history and perioperative steroid supplementation.

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