


### CR53 Management of Cushing's disease when surgery is a tricky option – a case report

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**KEYWORDS:** adrenocorticotrophic hormone; cortisol; Cushing's disease

**INTRODUCTION/OBJECTIVES:** Cushing's disease, a form of Cushing's syndrome, is caused by excess ACTH production, a hormone that regulates cortisol production, by a benign tumor in the pituitary gland. As a result, cortisol levels are elevated, while ACTH levels are not suppressed due to autonomous secretion. The primary treatment option is surgery. Other therapeutic modalities include drug therapy, radiation, and bilateral adrenalectomy in selected cases.

**CASE PRESENTATION:** In 2019, in a 65-year-old female patient with bilateral cortical adenoma, hypercortisolemia was detected. Further testing revealed elevated late-night salivary cortisol and insufficient cortisol suppression in Liddle's test. ACTH was not suppressed. Initially, no pituitary tumor was detected on NMR scans. Fluconazole was used as her initial treatment to inhibit steroidogenesis, but without a proper response. Then the patient has been prescribed metyrapone. Three months after starting metyrapone, the patient began to experience side effects, including stomach pain and irregular blood pressure. After re-running pituitary tests, a microadenoma was found too small for surgery. Gamma knife radiosurgery was conducted, but one month postoperatively, cortisol levels remained high and cabergoline treatment was introduced without adequate response over time. In 2022, low-dose pasireotide eventually replaced cabergoline. The patient's cortisol level is currently normal with clinical improvement and is being regularly checked.

**CONCLUSION:** Cushing's disease is a serious condition with systematic deteriorating effects. Treatment is still challenging, and there is still enough space for new treatment options that may benefit the patient.


### CR54 Management of metabolic crisis in three-day-old newborn

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**KEYWORDS:** hyperammonemia; ornithine transcarbamylase deficiency disease; urea cycle disorder

**INTRODUCTION/OBJECTIVES:** Urea cycle disorders are a group of metabolic disorders, caused by deficiency of one of the enzymes in the urea cycle, presenting with hyperammonemia triggered by catabolism or protein overload. Newborns with severe mutations in the urea cycle, unless treated, rapidly develop cerebral edema, coma and death.

**CASE PRESENTATION:** We present a 3-day-old male newborn born from the first pregnancy of a 36-year-old mother, complicated with mother's convulsion during birth. During the second day of life neonate presented with hyperventilation and weak sucking, later sepsis was suspected and antibiotics were introduced, but the neonate developed agonal breathing, altered consciousness, generalized hypertonus and lack of pupil response. Ammonia concentration in the serum, at the age of 60 hours, was altered (270 μmol/L). The patient was transferred to our unit ten hours later. At the admission ammonia concentration was 2062 μmol/L. The administration of amino acids was terminated, catabolism stopped with glucose infusion, nitrogen scavengers (intravenous sodium benzoate and arginine hydrochloride) were introduced, and hemodialysis was started. Laboratory results showed high plasma alanine and glutamine and low citrulline concentrations and increased orotic acid excretion suggesting urea cycle disorder, more specifically ornithine transcarbamylase deficiency, which was proved by gene analysis. The patient was discharged at the age of 6 weeks. He is now two years old and has acceptable neurocognitive development but is still at high risk of hyperammonemic crisis.

**CONCLUSION:** Timely recognition of urea cycle disorders enables quick intervention and lowering of harmful ammonia concentrations. Additional education of neonatologists is necessary to accomplish that goal.