

Rare Case of Spontaneous Conception in Female with Classic CAH



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Introduction: Congenital Adrenal Hyperplasia(CAH) is associated with the defective conversion of 17 hydroxyprogesterone to 11 deoxy cortisol mediated by 21-hydroxylase. Patients with classic and more severe forms of CAH (salt wasting) due to 21-hydroxylase deficiency present during neonatal or early infancy. Fertility is markedly reduced in females with classic 21-hydroxylase deficiency and its mechanism is multifactorial. The majority never attempt to conceive while only 10% of severely affected (salt wasting) would ever attempt to conceive. There are only a few case reports of pregnancy in patients with CAH.

Case: Our case is Mrs MS, a 37 years old female who was diagnosed with classical CAH at birth, was born with ambiguous genitalia and underwent 2 vaginal reconstruction surgeries at the age of 1 month and 10 years. She was started on lifelong Mineralocorticoids/Glucocorticoids replacement and had spontaneous conception at the age of 30 years. Genetic counselling was offered to the couple and on further testing, the partner was tested normal. The couple underwent thorough counselling about the implications of the condition and carrier status of the offspring. Her pregnancy was managed in a joint high-risk Obstetric and Endocrinology clinic. She was delivered via Caesarean section at 36 weeks due to previous vaginal reconstruction. Mrs MS gave birth to a healthy unaffected baby girl. Her second pregnancy was spontaneous conception as well which ended in the first trimester miscarriage requiring dilatation and curettage. The couple conceived third time and was managed again in a joint high-risk Obstetrics and Endocrinology clinic. She was diagnosed with Gestational diabetes (insulin dependent) and complicated with Placental abruption requiring Caesarean section at 32 weeks of gestation.

Discussion: Conception in females with CAH is a rare situation which should be managed by a multidisciplinary team including Obstetrician, Endocrinologist, Genetic Counsellor, Paediatrician and Anaesthetist. Ideal glucocorticoid replacement in pregnancy is hydrocortisone or prednisolone and they are continued till term. During the third trimester, about 50% of patients will need an increase in dose to adequately achieve adrenal hormone replacement. Dose adequacy is guided clinically. Mode of delivery is usually via LSCS due to vaginal inadequacy with an extra dose of hydrocortisone pre-operatively. With careful surveillance and multidisciplinary team involvement, the goal of term pregnancy and healthy infants can be achieved.

Conclusion: Counselling of females with CAH in regard to fertility during adolescence and later at the time of transition to adult life should be considered. Adequate glucocorticoid replacement will help achieve a successful pregnancy. There is no established guideline or consensus regarding the management of Mineralocorticoids/Glucocorticoids.