

CONGENITAL ADRENAL HYPERPLASIA AND PREGNANCY

Congenital adrenal hyperplasia (CAH) is an inherited endocrine disorder which affects the adrenal gland. Clinical symptoms range from hirsutism, irregular menses, and infertility in the mild form (which is also known as late onset or nonclassical CAH). Clinical features of the classical forms of CAH include effects on the development of female external genital structures as well as adrenal hormone (cortisol and aldosterone) deficiencies.

The management of women with CAH or at risk for having a child with CAH involves an interdisciplinary team approach focusing on preconception counseling and optimal management during pre-conception, pregnancy, labor, and delivery.

Will I be sterile? Women with CAH can have irregular menses and fail to ovulate (release an egg) regularly. Women with CAH can get pregnant and have healthy infants. Most importantly, CAH is not an effective form of birth control.

What preparation should I complete before trying to get pregnant? You should meet with your endocrinologist, reproductive endocrinologist, and obstetrician/gynecologist to discuss your hormone replacement treatments, how your medications may change when you are trying to conceive, and how they will monitor your pregnancy. Additional topics include possible need for medications to help you ovulate and achieve pregnancy. It is also important to meet with a genetic counselor so that you and your partner can determine the likelihood for you to have a child with CAH.

What tests are necessary during my pregnancy? You will need to have bloodwork to check your hormone replacement therapies (hydrocortisone and fludrocortisone) every 6-8 weeks during your pregnancy.

If my baby is a girl, will she have problems with external genital development observed in girls with CAH? A baby girl will have abnormally appearing external

genitalia only if she has classical CAH (severe form). If your baby girl does not have classical CAH, she will have normal female external genitalia.

What happens for labor and delivery? At the onset of active labor, you should be given hydrocortisone by vein. This medicine should be given every 6-8 hours until after the baby is born. Most women with CAH who have undergone vaginal surgery will deliver their babies by an elective Caesarian section.

What about treatment with dexamethasone during my pregnancy?

Dexamethasone is a very strong steroid medicine that crosses the placenta. This treatment should only be considered when there is a high chance of having a baby girl who has classical CAH. The goal of this treatment is to minimize the changes of the external genitalia of infant girls with CAH. However, there are potential major side effects for the baby. You should discuss the use of this medication with your partner and your physicians.

Can we decrease the risk of transmitting the genes associated with CAH to the baby?

Newer techniques such as preimplantation genetic diagnosis allow the selection of embryos (created by in vitro fertilization) that do not carry the abnormal genes for CAH. These specific embryos can be transferred back to the uterus to establish a pregnancy. You can discuss this option with your reproductive endocrinologist and genetic counselor.

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