Prenatal Genetics, Reproductive Genetics

# AB111. Prenatal treatment and fertility of female patients with congenital adrenal hyperplasia

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**Background:** Congenital adrenal hyperplasia (CAH) comprises a group of autosomal recessive disorders caused by defects in one of several steroidogenic enzymes involved in the synthesis of cortisol from cholesterol in the adrenal glands. The biochemical and clinical phenotype depends on the specific enzymatic defect and the impairment of specific enzyme activity. More than 90% of all cases of CAH are caused by 21-hydroxylase deficiency (21-OHD), of which in addition to cortisol, he synthesis of aldosterone is also impaired. The world-wide incidence of classic CAH ranges from 1:10,000 to 1:15,000 live births. 21-OHD is most common cause of ambiguous genitalia. Girls with classic 21-OHD of all severities are often born with varying degrees of genital ambiguity. Prenatal exposure to adrenal androgens activates the androgen receptors in genital skin, favors clitoral enlargement and labial fusion, and interferes with the urogenital sinus septation, which normally occurs at 7 weeks of gestation in girls. Women with 21-OHD have lower fertility rates, which correlate inversely with disease severity. The live-birth rates in salt-wasting classic 21-OHD have been reported to be only up to 10%, 33% to 50% in simple virilizing forms, and 63% to 90% in nonclassic 21-OHD, the latter rate being similar to that observed in the general population.

**Methods:** A case series of 21-OHD and its reproductive effect are reviewed and reported.

**Results:** We described prenatal treatment by maternal oral dexamethasone from 9 weeks of gestation in one female fetus with classic phenotype. She appeared to have normal external genitalia at birth. In addition, 3 CAH cases of late diagnosis, including 1 case with 11-beta-hydroxylase deficiency and 2 cases with 21-OHD, were reported. The pregnancies of these cases went well and normal infants were delivered.

**Conclusions:** Our data support favorable outcome of prenatal treatment for CAH.

**Keywords:** Congenital adrenal hyperplasia (CAH); prenatal treatment; reproductive in CAH

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