DISORDER: Congenital adrenal hyperplasia (CAH)

INCIDENCE: 1:16000 - 1:100000

MUTATION: Mutation in CYP21A2 gene transcript of 21 hydroxylase → enzyme that produces cortisol

Rare form: mutation in the CYP11B1 **Even rarer:** 3β-hydroxysteroid dehydrogenase deficiency

INFERTILITY SYMPTOMS:

Females: Virilization of female genitalia in utero, elevated androgen and progesterone production, impaired gonadotropins secretion, anovulation

Males: gonadal dysfunction, low sperm concentration and precocious puberty

In addition: hypertension and excessive production of deoxycorticosterone and its metabolites & deranged gonadal steroidogenesis in addition to adrenal defects

FERTILITY TREATMENT & CONCEPTION RATE: Glucocorticoids treatment or androgen blocking agents

CONCEPTION RATE (%): 0 – 10

DISORDER: Adult CAH

INCIDENCE: 1:600

MUTATION: characterized by enzyme deficiency which is phenotypically expressed after the onset of puberty

INFERTILITY SYMPTOMS:

Symptoms like polycystic ovarian syndrome, elevated androgen production, impaired fertility

DISORDER: Congenital adrenal hyperplasia (CAH)

INCIDENCE: Less than 300 cases worldwide

MUTATION: Deficiency in 17α-hydroxylase

INFERTILITY SYMPTOMS:

Females: Glucocorticoid, androgen, and estrogen synthesis is reduced, high FSH levels, multiple ovarian cysts, sexual infantilism hypergonadotropic hypogonadism, hypertension, and hypokalemia

Males: female phenotype with no breast development, a blind-ending vagina, and abdominal testes

FERTILITY TREATMENT & CONCEPTION RATE: pituitary down-regulation with GnRH agonists in conjunction with glucocorticoid treatment and exogenous estrogen treatment (cautious, however, to avoid ovarian hyperstimulation syndrome (OHSS))

CONCEPTION RATE (%): only one case of successful IVF treatment using this strategy with autologous oocytes has been reported