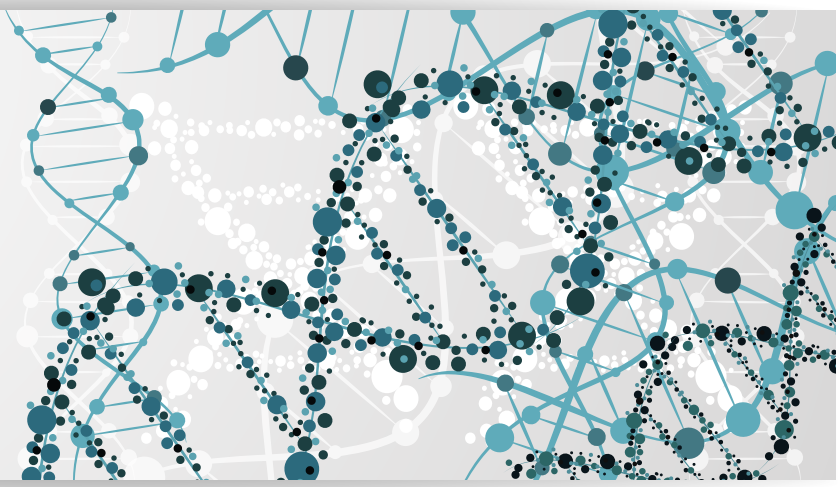


GENE Seeker

You know the risk of your child being a carrier of a genetic disease.



Congenital adrenal hyperplasia

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What is Congenital adrenal hyperplasia?

The salt wasting form of classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency (classical 21 OHD CAH; see this term) is characterized by virilization of the external genitalia in females, hypocortisolism, precocious pseudopuberty and renal salt loss due to aldosterone deficiency.

Patients suffer from glucocorticoid deficiency, aldosterone deficiency with salt wasting, failure to thrive, and potentially fatal hypovolemia and shock. Virilization of external genitalia in females is seen from birth. During the first weeks of life this salt wasting form can manifest in a salt-losing crisis (failure to thrive, vomiting, hyponatremia, hyperkalemia, acidosis, hypovolemia) which requires life saving treatment immediately. Precocious pseudopuberty, manifesting with various symptoms including accelerated growth velocity and bone maturation, is also present in both sexes.

What is the next step if I'm a carrier of Congenital adrenal hyperplasia?

If you are found to be a carrier of Congenital adrenal hyperplasia, it is important that your partner be tested for the same genetic disorder.

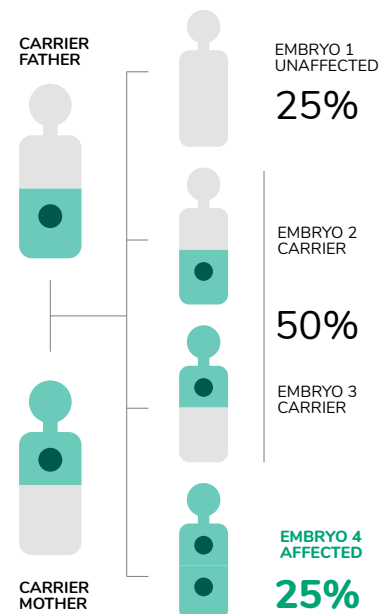
What if my partner is not a carrier?

If your partner's test for Congenital adrenal hyperplasia is negative, the chance to have an affected child is low. However there is currently no test able to detect all existing mutations, so there is always a residual risk that the person who has done the test is a carrier of other less frequent mutations.

What if both me and my partner are carriers of Congenital adrenal hyperplasia?

When both parents are carriers of Congenital adrenal hyperplasia, the probability of having a child with Congenital adrenal hyperplasia is 25%.

We recommend that you discuss your results with your doctor or genetic counselor in order to know more about reproductive options.



If both you and your partner are carriers, speak with your doctor or genetic counselor about reproductive options.

