

# X-Linked Congenital Adrenal Hypoplasia

## What Your Results Mean

Test results indicate that you are a carrier of X-linked congenital adrenal hypoplasia. Some female carriers can show symptoms, while others show no symptoms at all. Female carriers also have an increased chance to have a child with X-linked congenital adrenal hypoplasia. The chance to have a child with X-linked congenital adrenal hypoplasia depends on the sex of the child. Consultation with a genetic counselor for a more detailed risk assessment is recommended.

Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning.



## Recommended Next Steps

Male children have a 50% chance to have X-linked congenital adrenal hypoplasia, and female children have a 50% chance to be carriers. Some female carriers can show symptoms of X-linked congenital adrenal hypoplasia, while others show no symptoms at all. Prenatal testing for X-linked congenital adrenal hypoplasia is available, as well as testing shortly after birth to rule out a diagnosis in a child, particularly if the child is a male. In the absence of symptoms, reflexive testing of your male partner or donor for X-linked congenital adrenal hypoplasia is not recommended due to the X-linked inheritance pattern of the condition.

## X-Linked Congenital Adrenal Hypoplasia Explained

### What is X-Linked Congenital Adrenal Hypoplasia?

X-linked adrenal hypoplasia congenita is an inherited disorder that disrupts the body's production of hormones and is usually present in boys. One of the main signs is adrenal insufficiency, which begins in infancy or childhood with symptoms such as vomiting, difficulty with feeding, dehydration, low blood sugar, and shock. Boys with this disorder may also have a shortage of male sex hormones, causing underdeveloped reproductive tissues, undescended testicles, delayed puberty, and infertility.



### Prognosis

If untreated, X-linked adrenal hypoplasia can be life-threatening. With early diagnosis and proper medication management, most individuals have a normal life expectancy.

### Treatment

There is no cure for X-linked adrenal hypoplasia. Acute salt-wasting is usually treated with close monitoring in a hospital setting. Hormone replacement medications can help reduce some symptoms.



#### Resources

##### Genetics Home Reference

<https://ghr.nlm.nih.gov/condition/x-linked-adrenal-hypoplasia-congenita>

##### National Adrenal Diseases Foundation

<https://www.nadf.us/>

##### National Society of Genetic Counselors

<https://www.nsgc.org/>