

# Alpha Thalassemia X-Linked Intellectual Disability Syndrome

## What Your Results Mean

Test results indicate that you are a carrier of alpha thalassemia X-linked intellectual disability syndrome. Female carriers typically show no symptoms of alpha thalassemia X-linked intellectual disability syndrome. Female carriers are at an increased risk of having a child with alpha thalassemia X-linked intellectual disability syndrome. Risk for the current or future pregnancies is dependent on the sex of the baby.

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

There is a 50% risk to have a female child who is a carrier and a 50% risk to have a male child with alpha thalassemia X-linked intellectual disability syndrome. In the absence of clinical symptoms, reflexive testing for male partners of alpha thalassemia X-linked intellectual disability syndrome carriers is typically not indicated due to the X-linked inheritance pattern. Consultation with a genetic counselor for a more detailed risk assessment is recommended.

## Alpha Thalassemia X-Linked Intellectual Disability Syndrome Explained

### What is Alpha-Mannosidosis?

Alpha thalassemia X-linked intellectual disability syndrome is a condition that is characterized by intellectual disability and developmental delays in males. Most affected individuals have weak muscle tone which can delay motor skills such as sitting, standing, and walking. Many also have a significant speech delay and may never speak more than a couple of words in their lifetime. Additional signs and symptoms can include coarse facial features, anemia, small head size (microcephaly), short stature, skeletal abnormalities, and genital abnormalities or ambiguous genitalia.



### Prognosis

The prognosis for alpha thalassemia X-linked intellectual disability syndrome is not yet defined; however, affected males that have survived to their third decade of life have been reported.

### Treatment

Because there is currently no cure, treatment for alpha thalassemia X-linked intellectual disability syndrome is based on symptoms. Early intervention is beneficial to ensure affected individuals can reach their full potential. Physical, occupational, and speech therapy may be considered.



#### Resources

##### National Organization for Rare Disorders (NORD)

<https://rarediseases.org/rare-diseases/alpha-thalassemia-x-linked-intellectual-disability-syndrome/>

##### National Society of Genetic Counselors

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