



the  
**Oncofertility**<sup>®</sup>  
Consortium



## Center for Reproductive Health After Disease

Fertility  
Preservation Fact  
Sheet

## Aspartylglucosaminuria (AGU)

### 5 Questions to Ask Your Child's Health Care Provider

1. How is AGU affecting my child's health right now?
2. How quickly does my child need to start treatment?
3. Will treatment harm my child's future fertility?
4. What fertility options are out there?
5. Can my child have children in the future?

#### What is Aspartylglucosaminuria?

Aspartylglucosaminuria (AGU) is a rare, inherited lysosomal storage disorder. People with AGU have gene mutations, or misspellings in the body's instructions, which stop the body from breaking down an important sugar molecules. This causes the sugar molecules to build up in the body and prevents cells from doing their jobs, often resulting in cell destruction. Destroyed nerve cells cause many of the AGU symptoms, including mental retardation.

The first sign of AGU is delayed speech- children with AGU begin to speak between ages 2 and 3 years. As they age, a mild intellectual disability becomes more noticeable and learning occurs at a slower pace. The intellectual level of a 13-to-16 year old adolescent with AGU reaches the level of a 5-to-6 year old child without AGU before a decline in mental ability occurs. Adults with AGU have mental retardation and a limited vocabulary; they tend to survive into their third or fourth decade.

#### How Does Aspartylglucosaminuria Treatment Harm Fertility?

The standard treatment for AGU is hematopoietic stem cell transplant (HSCT). However, AGU is rare, so there is limited data regarding how safe and effective HSCT is for people with AGU. The outcome of HSCT usually depends on how quickly the disease progression occurs and the age of the transplant.

# The Center for Reproductive Health After Disease: Aspartylglucosaminuria (AGU)

It is recommended that HSCT occur as early as possible during infancy in individuals with AGU to have the best chance at stopping the disease from progressing. Before a child can have HSCT, they may have to have total body irradiation (TBI) and conditioning medication to prepare their body for the transplant. Both TBI and the necessary conditioning medications are considered *gonadotoxic*, meaning they harm the reproductive organs. A well-established risk of HSCT conditioned with TBI and high doses of gonadotoxic drugs is infertility.

## Female Fertility Preservation Options

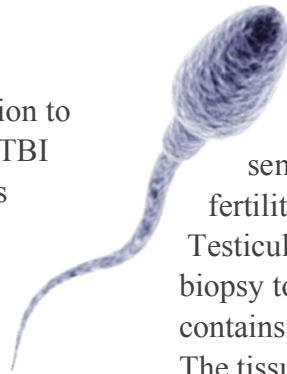
HSCT has a high risk of ovarian failure in females, which leads to premature menopause, subfertility, and infertility. Fertility preservation options should be discussed with parents of children needing HSCT for AGU.

Girls with AGU need HSCT as early as possible and usually have not gone through puberty at this time. Young girls' ovaries can't be stimulated, so the only option available to preserve fertility is *ovarian tissue cryopreservation*. Ovarian tissue cryopreservation involves surgically removing part or all of an ovary and freezing it until the female is ready to start a family. At that time the tissue is thawed and either transplanted back to the female to begin menstrual cycles and achieve a natural pregnancy, or the follicles are grown into eggs which are then fertilized with sperm and the embryo (fertilized egg) is implanted into the female.

## Male Fertility Preservation Options

HSCT has a high risk of *azoospermia*- a medical condition where there are no sperm present in semen, therefore fertility preservation options

should be discussed with parents of children needing HSCT for AGU.



Boys with AGU need HSCT as early as possible and have usually not gone through puberty at this time. Because pre-pubertal boys cannot produce semen, the only option available to preserve fertility is *testicular tissue cryopreservation*. Testicular tissue cryopreservation involves a biopsy to remove testicular tissue, which contains cells that create sperm and sperm itself. The tissue is frozen until the male wishes to start a family. At that time *intracytoplasmic sperm injection* (ICSI) can be used to achieve fertilization. ICSI is when a single sperm is microinjected directly into an egg.

## Alternative Options

Those with AGU who survive into adulthood may choose to use donor sperm or eggs, or consider adoption when family planning.

## Questions?

Call the 24-hour FERTLINE to ask your fertility preservation questions, get connected with a fertility preservation program near you, and access resources, tools, and support!



## Online Resources

<http://oncofertility.northwestern.edu>

<http://www.myoncofertility.org>

<http://www.savemyfertility.org>

Information found here or elsewhere on the [oncofertility.northwestern.edu](http://oncofertility.northwestern.edu) website should not be considered medical advice, diagnosis, or treatment. Any information on this document or website should not be used in lieu of consultation with your healthcare provider or physician. Before starting any course of treatment, always consult a qualified health care provider. Do not delay seeking or disregard medical advice because of anything you have read or seen here. For information regarding fertility options contact the FERT line at 866-708-FERT (3378).