



the
Oncofertility[®]
Consortium



Center for Reproductive Health After Disease

Fertility Preservation Fact Sheet

Ataxia Telangiectasia (A-T)

5 Questions to Ask Your Child's Health Care Provider

1. How is A-T affecting my child's health right now?
2. How is my child's gonadal (testes/ovaries) function?
3. Will A-T or treatment for A-T related cancer harm my child's future fertility?
4. What fertility options are out there?
5. Can my child have children in the future?

What is Ataxia Telangiectasia?

Ataxia Telangiectasia (A-T) is a genetic disorder caused by mutations (misspellings) in a gene called *ATM*. Genes are the instructions that tell our body how to grow and function. When mutations (misspellings) are in the *ATM* gene, the body can't repair misspellings in other genes. The resulting damage affects many parts of the body, in particular the immune system and the nervous system.

People with A-T develop extreme susceptibility to infection. They also have an increased risk to develop cancer, typically leukemia and lymphoma. The disease is named after the characteristic *ataxia*, or difficulty coordinating movements. The ataxia begins in the legs and arms and eventually effects speech as well. The second part of the name comes from *telangiectasias*, which are dilated blood vessels in the eyes and occasionally the skin of the face and ears.

How Does Ataxia Telangiectasia Effect Fertility?

A-T poses a small risk for *gonadal dysfunction*: non-working ovaries or testes. This may result in *amenorrhea* (no menstrual cycle) and infertility in females. Males may have *azoospermia* (semen does not contain sperm) and infertility.

Correctly working ovaries and testes are needed for puberty. So, children with gonadal dysfunction may not produce enough hormones and not develop *secondary sex characteristics*.

Secondary sex characteristics in males include: deepened voice, facial and body hair growth, increase in muscle mass, increase in height, broadened shoulders and chest, enlargement of the penis, and increased sweat and oil production causing acne and body odor.

Secondary sex characteristics in females include: breast development, menstruation, body hair growth, widening hips, and fat distribution around the hips, thighs, and butt.

Ataxia Telangiectasia and Cancer Risk

In addition to the small risk of gonadal dysfunction, individuals with A-T have a ~33% lifetime risk of developing cancer. *Gonadotoxic* drugs, such as chemotherapy and total body irradiation (TBI) are typically used to treat cancer, however they can have harmful effects on fertility. In the unfortunate event that a child with A-T is diagnosed with cancer, fertility preservation options should be discussed.

Two gene mutations (miss spellings) are necessary to develop A-T. Carriers of A-T silently carry one gene mutation (miss spelling) associated with A-T have an increased risk of developing breast cancer. Research studies have found female A-T carriers have an increased risk of developing breast cancer at a young age and a greater survival rate following treatment.

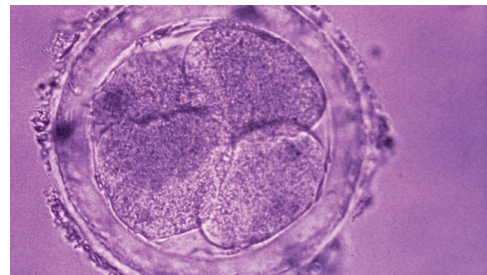
Carriers of A-T may be interested in preserving their fertility prior to cancer treatment or before prophylactic cancer risk-reducing surgery. Genetic counseling is encouraged to help explain the implications of A-T carrier status. Learn more about genetic counselors at: <http://oncofertility.northwestern.edu/your-genetic-counselor>



Female Fertility Preservation Options

Embryo Banking

Embryo banking is an option for females who have gone through puberty. First, a woman's ovaries are stimulated to mature multiple eggs, which are then removed and fertilized with sperm using *in vitro fertilization* (IVF) to create embryos. The embryos are frozen for future use. Embryo banking can take up to 1 month.



Egg Banking

Egg banking is an option for females who have gone through puberty. It is very similar to embryo banking, except the eggs are not fertilized before freezing. Egg banking is a good option for women who do not have a male partner and do not want to use a sperm donor at the time of the procedure. Egg banking can take up to 1 month.

Ovarian Tissue Banking

Ovarian tissue banking is an experimental option for females of any age. This is the only option for girls who have not started puberty. Part or all of an ovary is surgically removed and frozen for future use. The procedure can be done any time, as no stimulation of eggs is needed.

Information found here or elsewhere on the 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).

Online Resources for
More Information on
Fertility Preservation
Options

[MyOncofertility.org](http://www.myoncofertility.org)

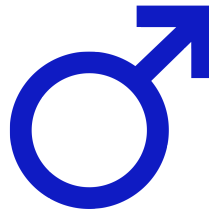
<http://www.myoncofertility.org>

SAVEMYFERTILITY

<http://www.savemyfertility.org>



<http://oncofertility.northwestern.edu>



Male Fertility Preservation Options

Sperm Banking

Sperm banking is an option for males who have gone through puberty. Most boys have sperm in their semen by age 13. Sperm cells are collected after ejaculation and frozen for future use.

Testicular Tissue Banking

Testicular tissue banking is a good option for males who have not gone through puberty, however it is an experimental procedure. For testicular tissue freezing, tissue from the testicles is collected during surgery and frozen in the hope that when thawed the *germ cells* (immature cells that normally mature into sperm) will develop into sperm and can be used in conjunction with *in vitro fertilization* (IVF) and *intra cytoplasmic sperm injection* (ICSI) to fertilize an egg.



Alternative Options

Individuals with A-T 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!



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