What is Adrenoleukodystrophy (ALD)?

Adrenoleukodystrophy (ALD) is a rare genetic condition that causes neurological deficits in males. ALD is caused by a gene mutation, or a misspelling in the body’s instructions. The mutation stops the body from breaking down very-long-chain fatty acids (VLCFAs). VLCFAs come from our food and our body breaks them down for energy. Males with ALD can’t break down the VLCFAs and they build-up in the nervous system, adrenal gland, and testis.

The cerebral form of ALD becomes symptomatic in childhood and the first features of ALD are often misdiagnosed as Attention Deficit Hyperactivity Disorder (ADHD). ALD causes cognitive deficits, including: poor handwriting, impaired memory, poor attention and reasoning, and difficulties with hearing and vision. As the disease progresses, children begin to have trouble moving muscles, trouble identifying objects by touch, “word deafness”, weakness on one side of the body, seizures, and adrenal insufficiency.

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<th>5 Questions to Ask Your Child’s Health Care Provider</th>
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Why is Fertility at risk in ALD?

Boys diagnosed with ALD are strongly recommended to have hematopoietic stem cell transplantation (HSCT) as early as possible. HSCT slowly provides the body with a new set of cells that can break down VLCFAs—leading to possible disease stabilization and reversal of the neurological changes seen in childhood ALD. 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, like chemotherapy, are considered gonadotoxic. Gonadotoxic medications cause azoospermia.

What is Azoospermia?

Azoospermia is the medical term used to describe a defect in how the body creates sperm. It causes ejaculation of semen that does not contain sperm and results in infertility.

Fertility Preservation Options

HSCT has a high risk of azoospermia, therefore fertility preservation options should be discussed with all young men of reproductive age and parents of children needing HSCT.

Boys with ALD 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 in microinjected directly into an egg.

Future Directions in Fertility Preservation

Current research may make it possible in the future to thaw testicular tissue that has been cryopreserved and transplant it back to the testes to result in possible natural creation of sperm within the body.

Alternative Options

Males with ALD who survive into adulthood may choose to use donor sperm 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