Globozoospermia

Subjects: Genetics & Heredity
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Globozoospermia is a condition that affects only males. It is characterized by abnormal sperm and leads to an inability to father biological children (infertility).

Keywords: genetic conditions

1. Introduction

Normal sperm cells have an oval-shaped head with a cap-like covering called the acrosome. The acrosome contains enzymes that break down the outer membrane of an egg cell, allowing the sperm to fertilize the egg. The sperm cells of males with globozoospermia, however, have a round head and no acrosome. The abnormal sperm are unable to fertilize an egg cell, leading to infertility.

2. Frequency

Globozoospermia is a rare condition that is estimated to affect 1 in 65,000 men. It is most common in North Africa, where it accounts for approximately 1 in 100 cases of male infertility.

3. Causes

Globozoospermia is most commonly caused by mutations in the *DPY19L2* gene, which are found in about 70 percent of men with this condition. Mutations in other genes likely also cause globozoospermia.

The *DPY19L2* gene provides instructions for making a protein that is found in developing sperm cells. The DPY19L2 protein is involved in the development of the acrosome and elongation of the sperm head, which are integral steps in sperm cell maturation. Mutations in the *DPY19L2* gene result in a loss of functional DPY19L2 protein. As a result, sperm cells have no acrosome and do not elongate properly. Without an acrosome, the abnormal sperm are unable to get through the outer membrane of an egg cell to fertilize it, leading to infertility in affected men. Researchers have described other characteristics of the abnormal sperm cells that make fertilization of an egg cell difficult, although it is not clear how changes in the *DPY19L2* gene are involved in development of these characteristics.

3.1. The gene associated with Globozoospermia

- DPY19L2

4. Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

5. Other Names for This Condition

- acrosome malformation of spermatozoa
- round-headed spermatozoa
- spermatogenic failure 9

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