Macrozoospermia

Description

Macrozoospermia is a condition that affects only males. It is characterized by abnormal sperm and leads to an inability to father biological children (infertility).

In affected males, almost all sperm cells have abnormally large and misshapen heads. The head of the sperm cell contains the male's genetic information that is to be passed on to the next generation. Normally, the head of a sperm cell contains one copy of each chromosome. In men with macrozoospermia, the sperm cell head contains extra chromosomes, usually four copies of each instead of the usual one. This additional genetic material accounts for the larger head size of the sperm cell. Additionally, instead of having one tail (flagellum) per sperm cell, affected sperm have multiple flagella, most often four.

Because of the additional genetic material, if one of these abnormal sperm cells combines with an egg cell, the embryo will not develop or the pregnancy will result in miscarriage.

Frequency

Macrozoospermia is estimated to affect 1 in 10,000 males in North Africa. The prevalence of the condition outside this region is unknown.

Causes

Mutations in the *AURKC* gene cause macrozoospermia. The *AURKC* gene provides instructions for making a protein called aurora kinase C. This protein is abundant in male testes, which are the male reproductive organs in which sperm are produced and stored. In the testes, this protein regulates the division of sperm cells. Aurora kinase C ensures that the mechanisms for cell division are in place and helps chromosomes properly align with each other so that every new sperm cell contains one copy of each chromosome after cell division.

AURKC gene mutations that cause macrozoospermia lead to the production of a nonfunctional protein or a protein that is quickly broken down. This lack of aurora kinase C blocks cell division in sperm cells. Without cell division, the chromosomes are not split among multiple new sperm cells. As a result, affected sperm cells contain extra chromosomes, usually four copies of each instead of the usual one.

Learn more about the gene associated with Macrozoospermia

- AURKC

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.

Other Names for This Condition

- Infertility associated with multi-tailed spermatozoa and excessive DNA
- Large-headed multiflagellar polyploid spermatozoa
- Spermatogenic failure 5

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Infertility associated with multi-tailed spermatozoa and excessive DNA (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0403812/)

Genetic and Rare Diseases Information Center

- Male infertility due to large-headed multiflagellar polyploid spermatozoa (https://rarediseases.info.nih.gov/diseases/12385/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Catalog of Genes and Diseases from OMIM

- SPERMATOGENIC FAILURE 5; SPGF5 (https://omim.org/entry/243060)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28macrozoospermia%29+OR+%28%28AURKC%5BTIAB%5D%29+AND+%28sperm%5BALL%5D%29%29+AND+english%5Bla%5D)

References

- Ben Khelifa M, Coutton C, Blum MG, Abada F, Harbuz R, Zouari R, Guichet A,May-Panloup P, Mitchell V, Rollet J, Triki C, Merdassi G, Vialard F, Koscinski I,Viville S, Keskes L, Soulie JP, Rives N, Dorphin B, Lestrade F, Hesters L, PoirotC, Benzacken B, Jouk PS, Satre V, Hennebicq S, Arnoult C, Lunardi J, Ray PF. Identification of a new recurrent aurora kinase C mutation in both European andAfrican men with macrozoospermia. Hum Reprod. 2012 Nov;27(11):3337-46. doi: 10.1093/humrep/des296. Epub 2012 Aug 11. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/22888167)
- Ben Khelifa M, Zouari R, Harbuz R, Halouani L, Arnoult C, Lunardi J, Ray PF. Anew AURKC mutation causing macrozoospermia: implications for humanspermatogenesis and clinical diagnosis. Mol Hum Reprod. 2011 Dec;17(12): 762-8.doi: 10.1093/molehr/gar050. Epub 2011 Jul 6. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/21733974) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3639514/)
- Dieterich K, Soto Rifo R, Faure AK, Hennebicq S, Ben Amar B, Zahi M, Perrin J, Martinez D, Sele B, Jouk PS, Ohlmann T, Rousseaux S, Lunardi J, Ray PF. Homozygous mutation of AURKC yields large-headed polyploid spermatozoa and causesmale infertility. Nat Genet. 2007 May;39(5):661-5. doi: 10.1038/ng2027. Epub 2007Apr 15. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17435757)
- Dieterich K, Zouari R, Harbuz R, Vialard F, Martinez D, Bellayou H, Prisant N, Zoghmar A, Guichaoua MR, Koscinski I, Kharouf M, Noruzinia M, Nadifi S, SefianiA, Lornage J, Zahi M, Viville S, Sele B, Jouk PS, Jacob MC, Escalier D, Nikas Y, Hennebicq S, Lunardi J, Ray PF. The Aurora Kinase C c.144delC mutation causesmeiosis I arrest in men and is frequent in the North African population. Hum MolGenet. 2009 Apr 1;18(7):1301-9. doi: 10.1093/hmg/ddp029. Epub 2009 Jan 15. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/19147683)
- Eloualid A, Rouba H, Rhaissi H, Barakat A, Louanjli N, Bashamboo A, McElreaveyK. Prevalence of the Aurora kinase C c.144delC mutation in infertile Moroccanmen. Fertil Steril. 2014 Apr;101(4):1086-90. doi:10.1016/j.fertnstert.2013.12.040. Epub 2014 Jan 30. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/24484996)
- Ounis L, Zoghmar A, Coutton C, Rouabah L, Hachemi M, Martinez D, Martinez G, Bellil I, Khelifi D, Arnoult C, Faure J, Benbouhedja S, Rouabah A, Ray PF.Mutations of the aurora kinase C gene causing macrozoospermia are the mostfrequent genetic cause of male infertility in Algerian men. Asian J Androl. 2015Jan-Feb;17(1):68-73. doi: 10.4103/1008-682X.136441. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/25219909) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4291881/)

Last updated January 1, 2015