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A novel mutation causes non obstructive azoospermia in infertile men

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Introduction: Infertility is defined as a failure of conception after 12 months of having unprotected intercourse, and male infertility accounts for 30-55% of infertile couples. Azoospermia, is diagnosed when sperm is completely absent in the ejaculate even after centrifugation.

Materials & Methods: Genotyping was done on four azoospermic individuals of a consanguineous Bedouin family and their parents. Exome sequencing was performed on the DNA of one patient.

Results: Assuming homozygosity of a recessive founder mutation as the likely cause of the disorder, we have genotyped 4 patients and their parents. We identified 5 shared homozygous regions larger than 2 cM, encompassing a total of 13.8Mbp on the autosomal chromosomes. In these regions only one homozygous variant with allele frequencies of less than 1% in the public databases (ExAc browser, 1000 Genomes and dbSNP) was identified. This variant segregated as expected in the family, with a calculated Lod score of 3.42. The variation was not present in 620 Bedouin controls. The variation is a frameshift mutation in a gene encoding a protein demonstrated to be essential for silencing of Line-1 retrotransposon in the male germline. Using a commercial antibody to the N-terminus of the encoded protein, immunofluorescent studies demonstrated it is produced in patients' testes, especially in spermatogonial cells (mainly in the cytoplasm) and in spermatocysts/round spermatids (mainly in the nucleus).

Discussion: The identification of the mutation causing azoospermia enables accurate diagnosis in the enlarged family and demonstrates the importance of repressing retrotransposon activation in the male germline in human.

Biography

Maram Arafat did her Master degree in Ben Gurion University in Israel. Now she is at the third year of my PhD in Genetics at Ben Gurion University in Israel. She published two papers, and presnting 3 posters in different conferences.

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