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Original Article

Deletion and Testicular Expression of DAZ (Deleted in Azoospermia) Gene in Patients with Non-Obstructive Azoospermia

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Abstract:

Background: Deletions of the DAZ (deleted in azoospermia) genes within the human Y chromosome's AZFc region are the most common cause of spermatogenesis failure. These deletions are usually assessed by analyses of genomic DNA extracted from peripheral leukocytes. DAZ genes are expressed in male germ cells. In this prospective study, we investigated DAZ expression and deletion in 102 consecutive infertile men presenting with non-obstructive azoospermia in Avesina Research Institute, Tehran, Iran during 2005-6.

Methods: In this prospective study, we extracted genomic DNA from peripheral blood leukocytes for detection of DAZ deletions and testicular biopsies for histopathological assessment and analyses of DAZ expression level by reverse transcription polymerase chain reaction. DAZ levels were normalized to expression of the housekeeping *Phosphoglucomutase 1* gene.

Results: In four out of 102 patients (3.9%), we found DAZ deletion. DAZ expression was observed in 60 (61.2%) of 98 other patients. Expression was not detected in patients with Sertoli cell-only syndrome, but observed in 37 of 40 (92.5%) patients with maturation arrest and 20 of 26 (76.9%) with hypospermatogenesis.

Conclusion: The absence of DAZ expression could result in quantitative reduction of germ cells and might be observed despite of normal genomic DNA constitution. We recommend to check DAZ testicular expression and genomic DNA deletion, in non-obstructive azoospermia. This is more recommended to avoid transmission of genetic abnormalities which might lead to infertility in male offspring, when assisted reproductive techniques (ART) are performed.

Keywords:

Infertility ، *Deleted in Azoospermia Gene* ، *Y Chromosome* ، *Azoospermia* ، *Iran*

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