
# Investigation of GRTH gene single nucleotide polymorphism in association with male infertility in Iranian population

Sharareh Hosseinzadeh Kashani, Elham Siasi Tarbiati, Zahra Sadat Talari, Parviz Pakzad

PII: S2214-5400(18)30007-0
DOI: https://doi.org/10.1016/j.mgene.2018.01.006
Reference: MGENE 392

To appear in: *Meta Gene*

Received date: 20 August 2017
Revised date: 25 December 2017
Accepted date: 18 January 2018

Please cite this article as: Sharareh Hosseinzadeh Kashani, Elham Siasi Tarbiati, Zahra Sadat Talari, Parviz Pakzad , Investigation of GRTH gene single nucleotide polymorphism in association with male infertility in Iranian population. The address for the corresponding author was captured as affiliation for all authors. Please check if appropriate. Mgene(2017), https://doi.org/10.1016/j.mgene.2018.01.006

# Investigation of GRTH gene single nucleotide polymorphism in association with male infertility in Iranian population

**Sharareh Hosseinzadeh Kashani[a], Elham Siasi Tarbiati[a], Zahra Sadat Talari[b],*,Parviz Pakzad[a]**

a Department of life Sciences, Islamic Azad University, North Tehran Branch, Tehran, Iran

b Faculty of Biological Sciences and Biotechnology, Shahid Beheshti University, Tehran, Iran

**Abstract**

**Introduction:** *GRTH* Gene is located on chromosome 11q24 and consists of 14 exons. *GRTH* codes for a protein product which is involved in testicular function and spermatogenesis. The absence of this gene can lead to gametogenesis arrest. The purpose of this study was to investigate the association of SNP IVS6+55G-->T (rs551373) in *DDX25 / GRTH* gene with male infertility.

**Methods:** in this case-control study, DNA samples were extracted from 180 men, including 90 fertile men and 90 infertile cases with azoospermia by Salting out method. PCR-RFLP was conducted to identify the SNP in *DDX25 / GRTH* gene.

**Results**: according to statistical analyses carried out in SPSS and SNPstats online software, the SNP rs551373 is significantly associated with azoospermic cases studied.

**Conclusion:** The results of our study suggest that positive association of rs551373 SNP probably reveals an important role in azoospermia male infertility. Further studies with larger sample size are required to support our findings. Investigation of association of this SNP with other types of non-obstructive male infertility is also suggested.

**Keywords**: single nucleotide polymorphisms; *DDX25/ GRTH* gene; male infertility; azoospermia.

## 1. Introduction

In spite of so many attempts made by numerous researchers for finding the underlying causes of male infertility, about half of the male infertility cases remain unexplained (Carrell et al., 2001; Nuti et al., 2008). Azoospermia, which is defined by the absence of sperm in the semen, affects nearly 1% of all men and is the cause of 10 to 15% of infertility in males (Jarow et al., 1989).Due to the complexity of the spermatogenesis and the large number of genes that are involved in this process, a sizeable proportion of infertility cases are believed to have a genetic cause (Kenneth et al., 2009). Since some SNPs were present at higher rates in some populations compared to others, scientists claimed that there is an association between certain SNPs and diseases. Applying this fact to infertility as a disease, particular SNPs might be used to estimate the probable risk of infertility. SNPs are the simplest and most common form of genetic polymorphism in human genome. SNPs are estimated to account for close to 90% of all human genome polymorphisms (Collins et al., 1998). DDX25 (DEAD-Box Helicase 25) gene, also named GRTH (Gonadotropin-regulated testicular RNA helicase) is located on chromosome 11q24.3 (-http://www.genecards.org). GRTH, which is a member of DEAD-box protein family, is a Gonadothropine / androgen and is present in miotic spermatocytes. All DEAD-box proteins are RNA helicases and contain an Asp-Glu-Ala-Asp motif. This protein family unwinds RNA tertiary structure using their specific motifs and ATP with their helicase and ATPase activities. GRTH protein is a post-transcriptional regulatory factor and its presence is necessary for the completion of spermatogenesis in germ cells (Tang et al., 1998; Sheng et al., 2003). DDX25/GRTH will be referred to as GRTH in this article hereafter. Male