



## OPEN ACCESS

## EDITED AND REVIEWED BY

Maxim B. Freidin,  
Queen Mary University of London,  
United Kingdom

## \*CORRESPONDENCE

Xiaojin He,  
✉ hxj0117@126.com

## SPECIALTY SECTION

This article was submitted to Human and Medical Genomics, a section of the journal Frontiers in Genetics

RECEIVED 16 March 2023

ACCEPTED 27 March 2023

PUBLISHED 05 April 2023

## CITATION

He X, Yang S and Lv M (2023), Editorial: Genetic factors in male infertility. *Front. Genet.* 14:1187445. doi: 10.3389/fgene.2023.1187445

## COPYRIGHT

© 2023 He, Yang and Lv. This is an open-access article distributed under the terms of the [Creative Commons Attribution License \(CC BY\)](https://creativecommons.org/licenses/by/4.0/). The use, distribution or reproduction in other forums is permitted, provided the original author(s) and the copyright owner(s) are credited and that the original publication in this journal is cited, in accordance with accepted academic practice. No use, distribution or reproduction is permitted which does not comply with these terms.

# Editorial: Genetic factors in male infertility

Xiaojin He<sup>1\*</sup>, Shenmin Yang<sup>2</sup> and Mingrong Lv<sup>1</sup>

<sup>1</sup>Reproductive Medicine Center, Department of Obstetrics and Gynecology, The First Affiliated Hospital of Anhui Medical University, Hefei, China, <sup>2</sup>Center for Reproduction and Genetics, Suzhou Municipal Hospital Suzhou, Gusu School, The Affiliated Suzhou Hospital of Nanjing Medical University, Nanjing Medical University, Suzhou, China

## KEYWORDS

male infertility, teratozoospermia, azoospermia, genetic factors, gene variants

## Editorial on the Research Topic Genetic factors in male infertility

Our Research Topic is “Genetic Factors in Male Infertility” and the goal of this Research Topic is to collect more papers on the aspect of identifying different gene variants and various types of RNAs mainly focused on male infertility including azoospermia, oligozoospermia, teratozoospermia, and asthenozoospermia. In order to achieve this goal, we carefully reviewed every submitted manuscript and screened for highly qualified reviewers. Eventually, we accepted and published seven articles including four “Original Research” articles, one “Brief Research Report” article, one “Case Report” article, and one “Review”. These articles covered the genetic factors of non-obstructive azoospermia and teratozoospermia and the clinical outcomes of men with non-mosaic Klinefelter syndrome (KS). Xie et al. identify a homozygous missense variant in *DND1* that causes non-obstructive azoospermia in humans (Xie et al.). Aprea et al. identify the pathogenic gene variants in *CCDC39*, *CCDC40*, *RSPH1*, *RSPH9*, *HYDIN*, and *SPEF2* that cause defects of sperm flagella composition and male infertility. They find that immunofluorescence microscopy in sperm cells is a valuable tool to identify flagellar defects related to the axonemal ruler, radial spoke head, and the central pair apparatus (Aprea et al.). Yuan et al. finds a Gly684Ala substitution in the androgen receptor that causes azoospermia (Yuan et al.) and Fang et al. reports the phenotypic findings and genetic considerations of congenital absence of the vas deferens with hypospadias or without hypospadias: (Fang et al.), which might advance the genetic diagnosis and clinical genetic counseling for male infertility. Zhu et al. also verifies that FISH could analyze numerical chromosomal abnormalities in the sperm of Robertsonian translocation der (13; 14) (q10; q10) carriers (Zhu et al.). Moreover, Wang et al. review various phenotypes resulting from different pathogenic genes, including sperm ultrastructure and encoding proteins with their location and functions as well as

assisted reproductive technology outcomes, providing additional clinical views and broadening the understanding of this disease (Wang et.al). Finally, Xu et al. reports a case of the birth of a boy after intracytoplasmic sperm injection using ejaculated spermatozoa from a non-mosaic KS man with normal sperm motility, which increases our knowledge of non-mosaic KS (Xu et al.). The findings in these studies broaden our understanding of genetic factors in male infertility and were also included in our Research Topic scope.

## Author contributions

XH generally directs and modifies the manuscript of the editorial. SY and ML wrote it.

## Conflict of interest

The authors declare that the research was conducted in the absence of any commercial or financial relationships that could be construed as a potential conflict of interest.

## Publisher's note

All claims expressed in this article are solely those of the authors and do not necessarily represent those of their affiliated organizations, or those of the publisher, the editors and the reviewers. Any product that may be evaluated in this article, or claim that may be made by its manufacturer, is not guaranteed or endorsed by the publisher.