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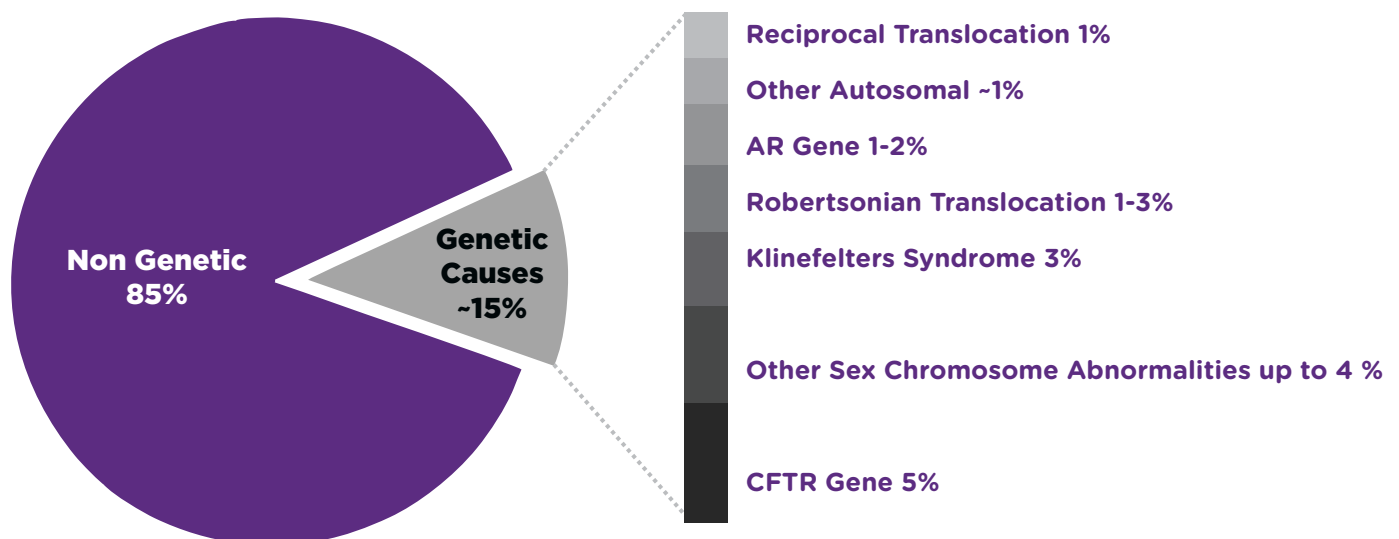
CENTER FOR
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Male Infertilty **Testing**

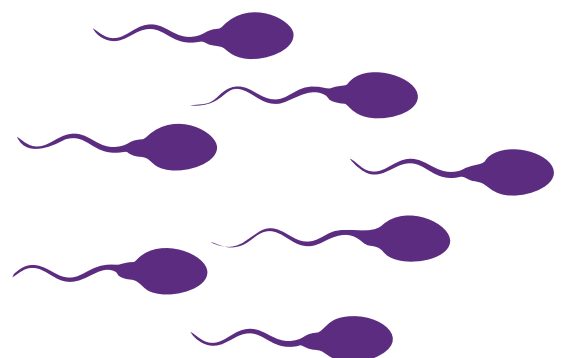
- ▶ According to the World Health Organization and the International Committee Monitoring Assisted Reproductive Technologies (ICMART), failure to get pregnant is defined as clinical infertility if pregnancy is not established after 12 months or more of regular unprotected sexual intercourse
- ▶ While either one or both of the partners may contribute to the reproductive challenges of the couple, male infertility, like female infertility, is a clinical diagnosis that can only be determined after formal assessment and testing
- ▶ According to WHO, 48 million couples and 186 million individuals live with infertility globally
- ▶ A male factor contributes in up to 50% of these couples
- ▶ Among this, 15% males and 10% of females are predicted to have genetic causes

Causes Of Male Infertility



- ▶ Etiology of male infertility ¹can be broadly divided into :
 - Endocrine Disturbance
 - Quantitative/qualitative
 - Spermatogenic Defects
 - Distal Obstruction

- ▶ Known genetic anomalies underlie ~15% of male infertility cases²



Why Genetic Testing Is Important For Male Infertility?



Diagnostic Benefit

Genetic testing helps to identify the cause of the infertility which helps to understand implications in general health of the individual and his future generation



Prognosis

By detected the specific mutation or chromosomal aberration, the outcome of various reproductive technique such as TESE and ICSI can be predicted



Treatment & Management

It also helps to deliver specific treatment and management for a successful pregnancy outcome

Test Indication

- ▶ Failed to achieve pregnancy over one year
- ▶ Low sperm count (azoospermia or oligospermia) & increased FSH level confirmed by Semen Analysis
- ▶ Physical defects such as small testicles
- ▶ Congenital absence of vas deferens

Genetic test	Society		
	AUA	EAU	ASRM
Karyotyping	NOA or <5 million/mL	Sperm conc <10 million/mL	NOA or <5 million/mL
YCMD	NOA or <5 million/mL	Sperm conc <5 million/mL	NOA or <5 million/mL
CFTR	CBAVD	CBAVD or CUAVD without renal abnormalities	CBAVD, CUAVD without renal abnormalities, or bilateral epididymal obstruction
	Female partners should also be tested	Female partners should also be tested	Female partners should also be tested

Fig 1: Condition specific indication for testing

Biochemical Testing

Test name	Technique	Sample Type	Turn Around Time
????	Automated sperm analyzer with powerful visualization system	Semen	1 Day

Advanced Cytogenetic Testing

Test name	Technique	Sample Type	Turn Around Time
Karyotype	G-banding	Heparin Whole Blood	7 Day
Sperm Fish	—	Seminal Fluid	?

Advanced Molecular Testing

Test name	Technique	Sample Type	Turn Around Time
Y-chromosome microdeletion	Multiplex Real Time PCR	Extracted DNA, Whole Blood EDTA	7 Day
Sperm DNA Fragmentation	Gel Method	Seminal Fluid	5 Day
CFTR - Single Gene Sequencing	NGS	Whole Blood EDTA, Extracted DNA	36 Day
Chromosomal Microarray 315K	SNP Based Array	Whole Blood EDTA, Extracted DNA	12 Day
Sperm DNA Fragmentation	NGS	Whole Blood EDTA, Extracted DNA	36 Day

- ▶ All tests are recommended by The American Urologic Association (AUA) & American Society of Reproductive Medicine (ASRM)
- ▶ High Sensitivity
- ▶ Faster TAT
- ▶ Cost Effective
- ▶ Our group of renowned Clinical Geneticists and trained certified Genetic Counselors are available to provide :
 - Support in ordering test
 - Additional information about testing
 - Offers result interpretation
 - Helps to understand future prognosis and management

1. Pelzman DL, Hwang K. Genetic testing for men with infertility: techniques and indications. *Transl Androl Urol.* 2021 Mar;10(3):1354-1364. doi: 10.21037/tau-19-725. PMID: 33850771; PMCID: PMC8039607.
2. Ferlin A, Raicu F, Gatta V, et al. Male infertility: role of genetic background. *Reprod Biomed Online* 2007;14:734-45.
3. Schlegel PN, Sigman M, Collura B, De Jonge CJ, Eisenberg ML, Lamb DJ, Mulhall JP, Niederberger C, Sandlow JI, Sokol RZ, Spandorfer SD, Tanrikut C, Treadwell JR, Oristaglio JT, Zini A. Diagnosis and treatment of infertility in men: AUA/ASRM guideline part I. *Fertil Steril.* 2021 Jan;115(1):54-61. doi: 10.1016/j.fertnstert.2020.11.015. Epub 2020 Dec 9. PMID: 33309062.



Signs Of Male Infertility

- Having a decreased sex drive
- A decrease in growth of facial or body hair
- Suffering from respiratory infections repeatedly
- Continuous pain and swelling in the testicular area
- Difficulty in ejaculation or low amount of ejaculation



Our Services



Inherited Genetic Disorder



Reproductive Genetics



Cancer Genomics



Haemato Oncology



Transplant Immunology



Infectious Disorders



Pharmacogenomics



Research Services

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