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Clinical & Embryology
Academy of ART
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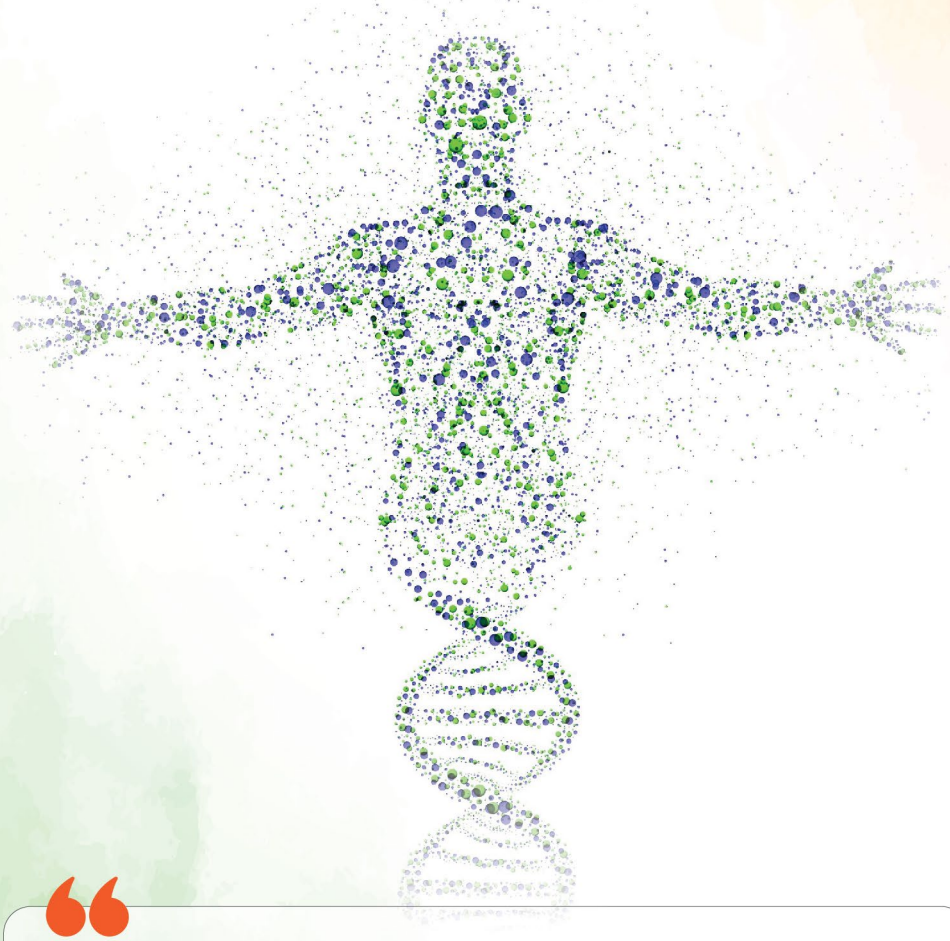
i-Ceat RESONANCE

GENETICS IN MALE INFERTILITY: THE ERA OF GENOMICS



"Everybody is a genius. But if you judge a fish by it's ability to climb a tree, it will live it's whole life believing that it is stupid

- Albert Einstein

Preface

To accomplish it's goal of providing a basic understanding of ART technologies and it's clinical relevance, **i-Ceat academy** comes out with it's **INDEPENDENCE DAY SPECIAL** bulletin of **RESONANCE** retaining it's unique approach of combining economy with concept building.

The essence of it's supremacy lies in the simplified approach in providing a meritorious playground that even the toughest game (topic) feels like a cake walk. i Ceat is the academy of students where they drink up all the concept building basics of ART and get practical tips and tricks for effective and ethical practice in the field. We thank all our student assets in INDIA and across the borders, their unparalleled enthusiasm have made us stand elite on the globe in such a short span of time.

On the marvelous occasion of INDEPENDENCE DAY, i-Ceat team congratulates all it's participants and wish them good luck.

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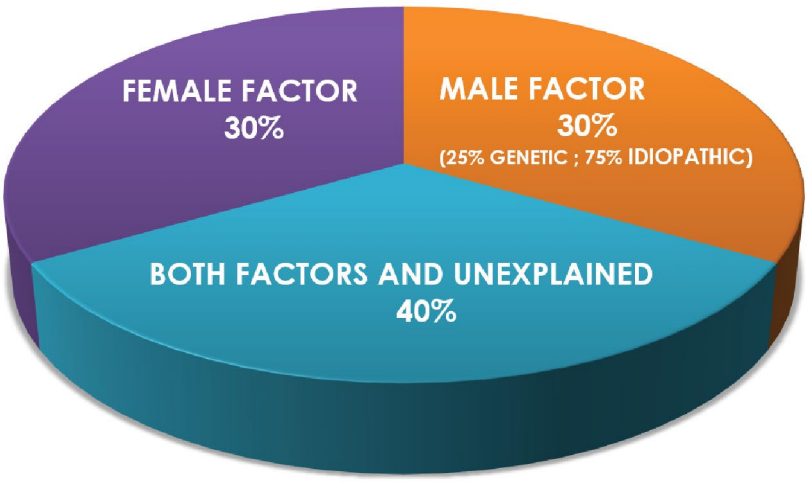
'If you can't explain it simply,
you don't understand it well enough'
- Albert Einstein

GENETICS IN MALE INFERTILITY: THE ERA OF GENOMICS

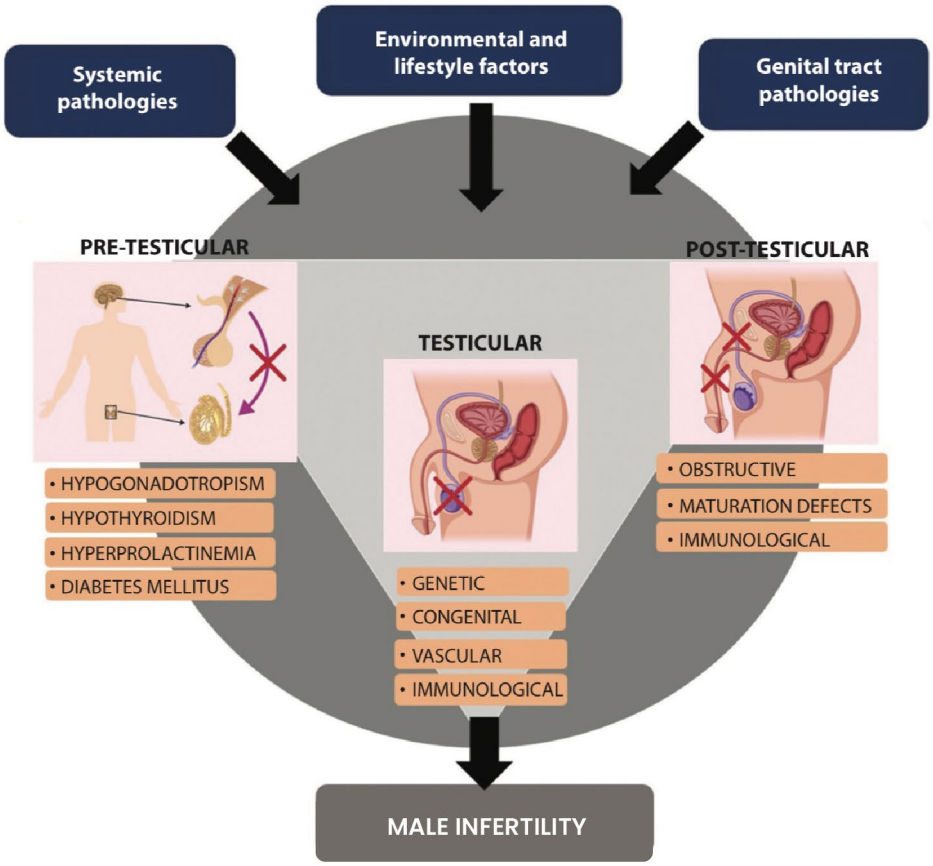
- Male infertility means that a man has a problem with his reproductive system and is unable start a pregnancy with his fertile female partner.
- Infertility effects 10-15% couples

GENDER FACTORS INVOLVED IN INFERTILE COUPLES



CAUSES OF INFERTILITY IN MALES

(25% genetic; 60-75% idiopathic; idiopathic causes mostly are genetic too)



GENETIC CAUSES OF INFERTILITY IN MALES

CYTOGENETIC OR CHROMOSOMAL CAUSES

1. Y chromosome defects
2. Other chromosome defects

GENETIC TESTING

1. Karyotyping
2. Microarray
3. PCR
4. FISH
5. MLPA

SINGLE GENE DEFECTS

1. CFTR Gene mutations
2. AR gene mutations
3. Hypogonadotropic hypogonadism
4. Infertility as part of genetic syndrome

GENETIC TESTING

1. Sanger
2. Panels
3. Exome

OBJECTIVES OF GENETIC TESTING IN MALE INFERTILITY

- To establish the etiology and diagnosis of associated comorbidity (e.g. vasal aplasia with unilateral renal agenesis, immotile cilia syndrome predisposition of recurrent chest infections).
- Prognostic for surgical sperm retrieval, leading to success of IUI / ICSI procedure.
- Rule out inheritance of genetic disorders in off springs.
- Personalizing therapy & counselling.

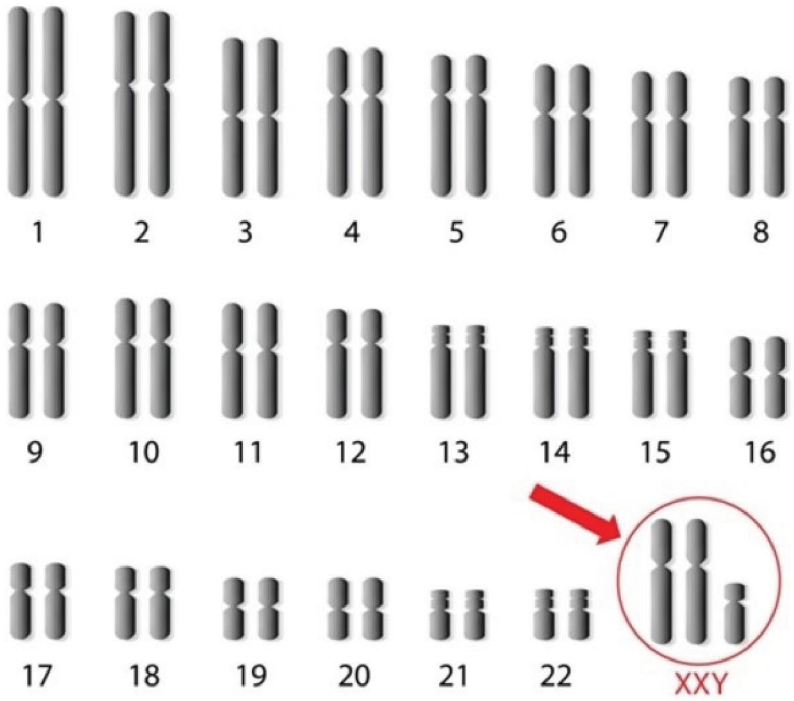
GENETIC DISORDERS : MALE INFERTILITY

1. Klinefelter syndrome (KS) - 47,XXY
2. Hypogonadotropic hypogonadism (HH) gene mutation.
3. Y chromosome microdeletions.
4. CFTR (7q.31.2)mutations - CBAVD (Congenital Bilateral Absence of Vasa Differentia).

KLINEFELTER SYNDROME (KS)- 47,XXY

- ✓ Extra X-chromosome
- ✓ Meiotic imbalance
- ✓ Overexpression X-Y genes
- ✓ Successful sperm recovery in KS patients(50%)
- ✓ ICSI success (50%)

KLINEFELTER SYNDROME



HYPOGONADOTROPIC HYPOGONADISM GENE MUTATION

Influences genes controlling hypothalamo-pituitary axis

↓

Impairment of GnRH production

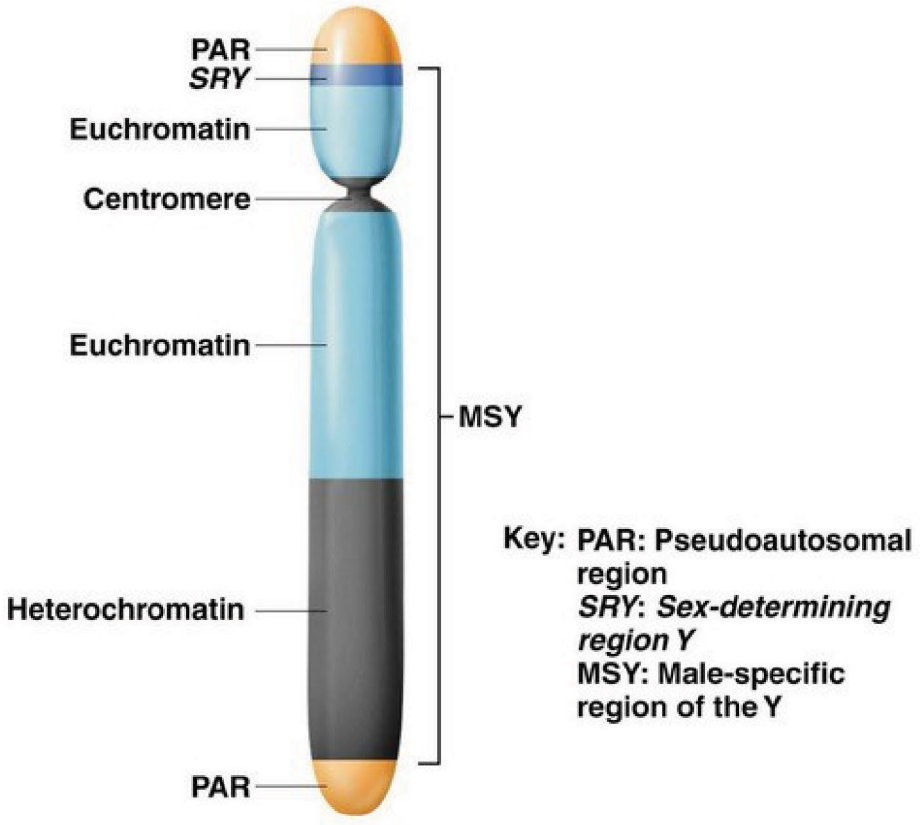
↓

Spermatogenesis negatively influenced

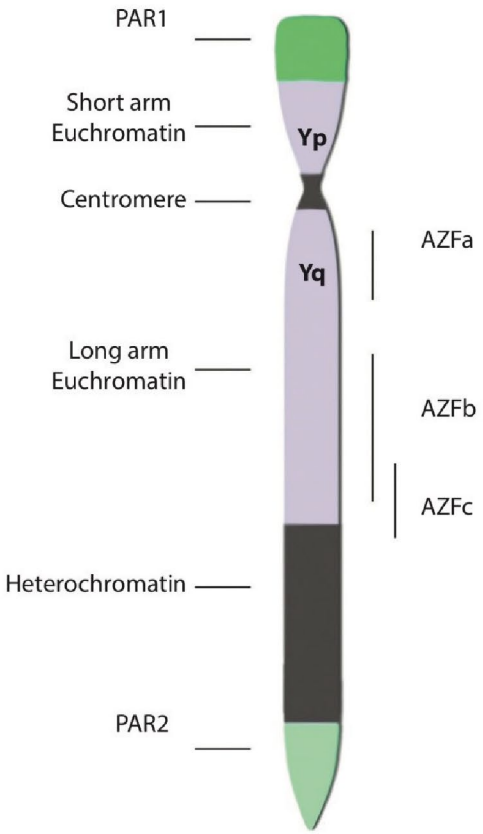
Y CHROMOSOME MICRODELETIONS (Yq11.2)

- ✓ The human Y chromosome contains many genes that are essential for male sex determination and spermatogenesis.
- ✓ Y chromosome long arm (q) has:
  - Azoospermia factor region (AZF a,b,c)- It is vital for normal spermatogenesis.
  - AZF Deletion ➡ Male infertility

A schematic representation of the Y Chromosome.



A schematic representation of the Y Chromosome (Yq11.2). The long arm Yq contains azoospermia factor (AZF) regions



CYSTIC FIBROSIS TRANSMEMBRANE CONDUCTANCE REGULATOR (CFTR) GENE

- ✓ CBAVD patients require surgical sperm retrieval and ICSI.
- ✓ Should have their female partners screened for CFTR mutations.
- ✓ PGD required

CFTR gene is present on chromosome 7q.31.2 → ✓Codes for CFTR protein → ✓Influences viscosity of epithelial secretions and development of male reproductive structures → ✓CFTR gene mutations → ✓Cystic fibrosis (CF) associated with CBAVD

CFTR GENE MUTATION: CBAVD

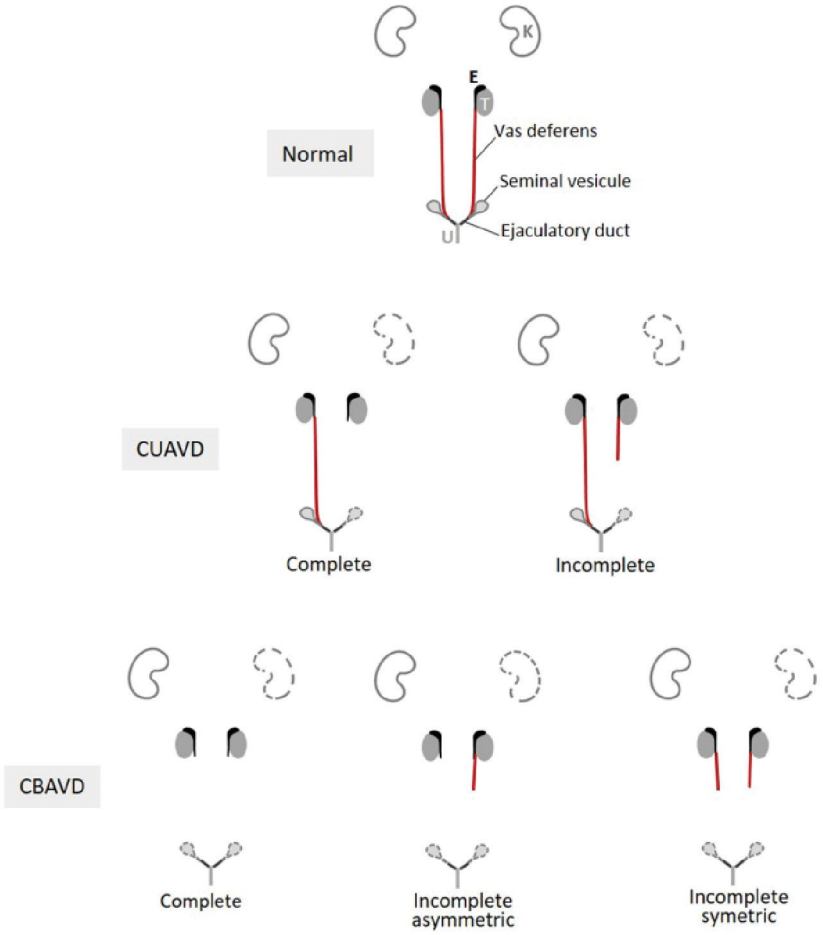


Fig. 1

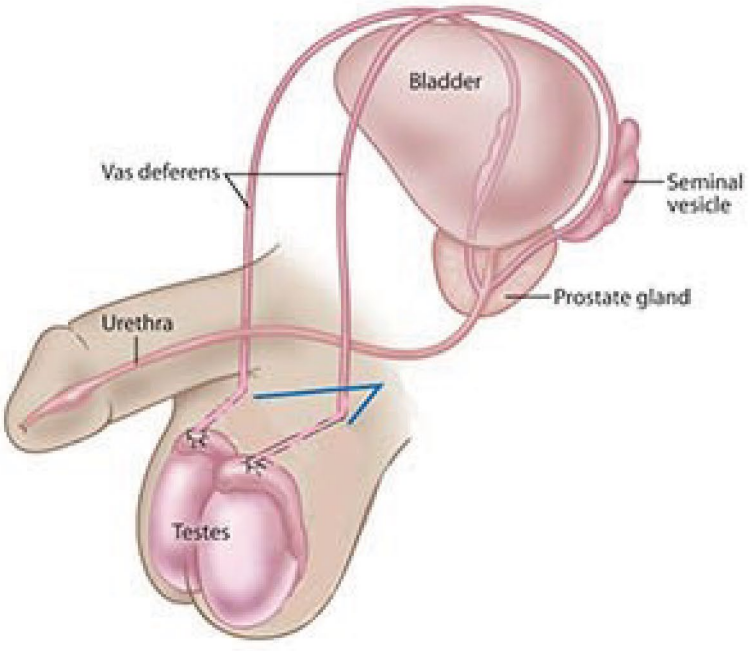


Fig. 2

SUMMARY

- Genetic factor is responsible for upto 25% of men with severe oligospermia and azoospermia.
- Counts less than 5-10 million/ml - Karyotype abnormalities suspected.
- Counts less than 5 million/ml - Y chromosome microdeletions suspected.
- Screening for specific gene mutations depends on the clinical picture e.g. as in vasal aplasia, hypogonadism and specific sperm disorders.

Scientific Committee

Dr Prof (Col) Pankaj Talwar, VSM

Dr Gunjan Bhatnagar | Dr Neeti Bansal

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