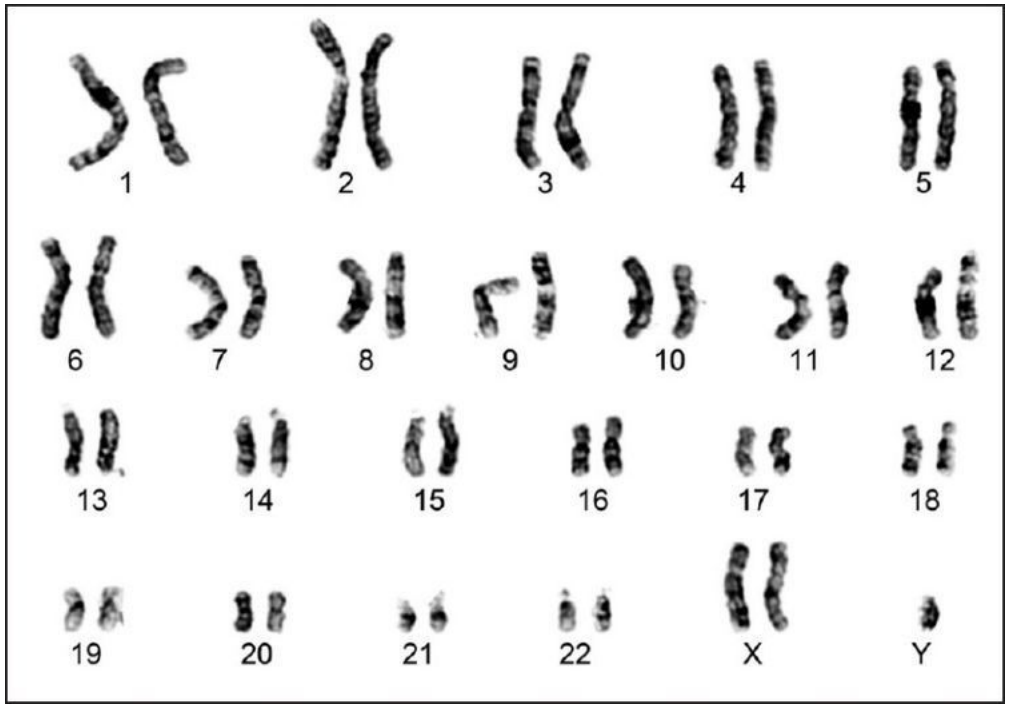
MEDICAL BIOLOGY

HEREDITARY DISEASES

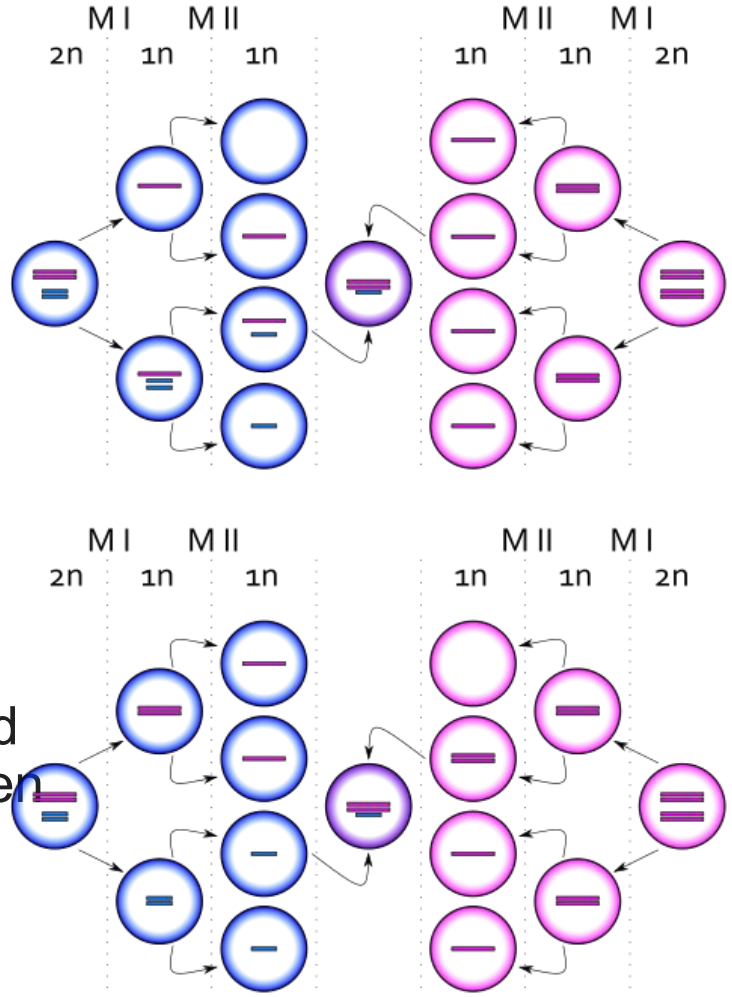
Klinefelter syndrome



- **Klinefelter syndrome** (**KS**), also known as **47,XXY** is the set of symptoms that result from two or more X Chromosomes in males.
- The primary features are infertility and small poorly functioning testicles.Often, symptoms are subtle and subjects do not realize they are affected.
- Sometimes, symptoms are more evident and may include weaker muscles, greater height, poor Coordination , less body hair, breast growth , and less interest in sex.
- Often it is only at puberty that these symptoms are noticed. Intelligence is usually normal; however, reading difficulties and problems with speech are more common.
- Symptoms are typically more severe if three or more X chromosomes are present (48,XXXY syndrome or 49,XXXXY syndrome).
- Klinefelter syndrome occurs randomly.The extra X chromosome comes from the father and mother nearly equally.

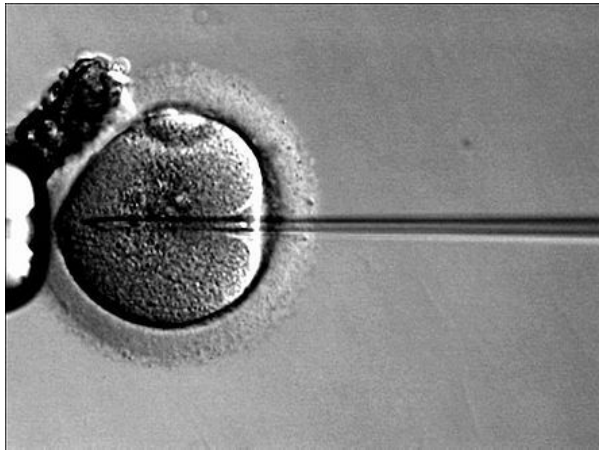
COGNITIVE AND DEVELOPMENTAL

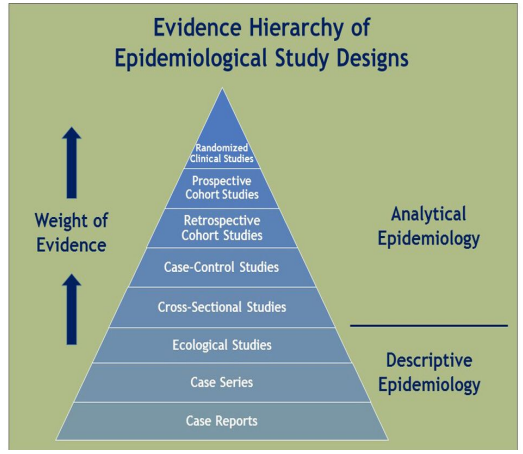
- Some degree of language learning or reading impairment may be present, and neuropsychological testing often reveals deficits in executive functions , although these deficits can often be overcome through early intervention.
- delays in motor development may occur, which can be addressed through occupational and physical therapies.
- XXY males may sit up, crawl, and walk later than other infants; they may also struggle in school, both academically and with sports. It's estimated that 10% of men with Klinefelter syndrome are Autistic.

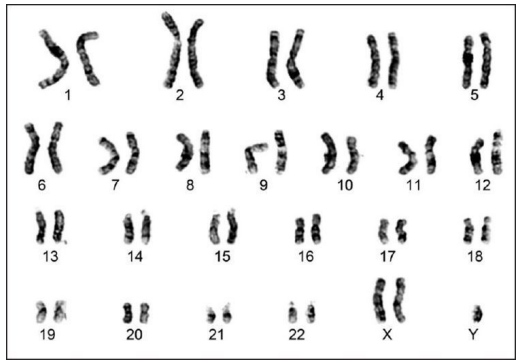


Treatment

- The genetic variation is irreversible, thus there is no causal therapy. From the onset of puberty, the existing testosterone deficiency can be compensated by appropriate hormone replacement therapy.
- Testosterone preparations are available in the form of syringes, patches or gel. If gynecomastia is present, the Surgical removal of the Brest, may be considered for both the psychological reasons and to reduce the risk of breast cancer.
- The use of Behavioral therapy can mitigate any language disorders, difficulties at school, and socialization. An approach by Occupational therapy is useful in children, especially those who have dyspraxia .

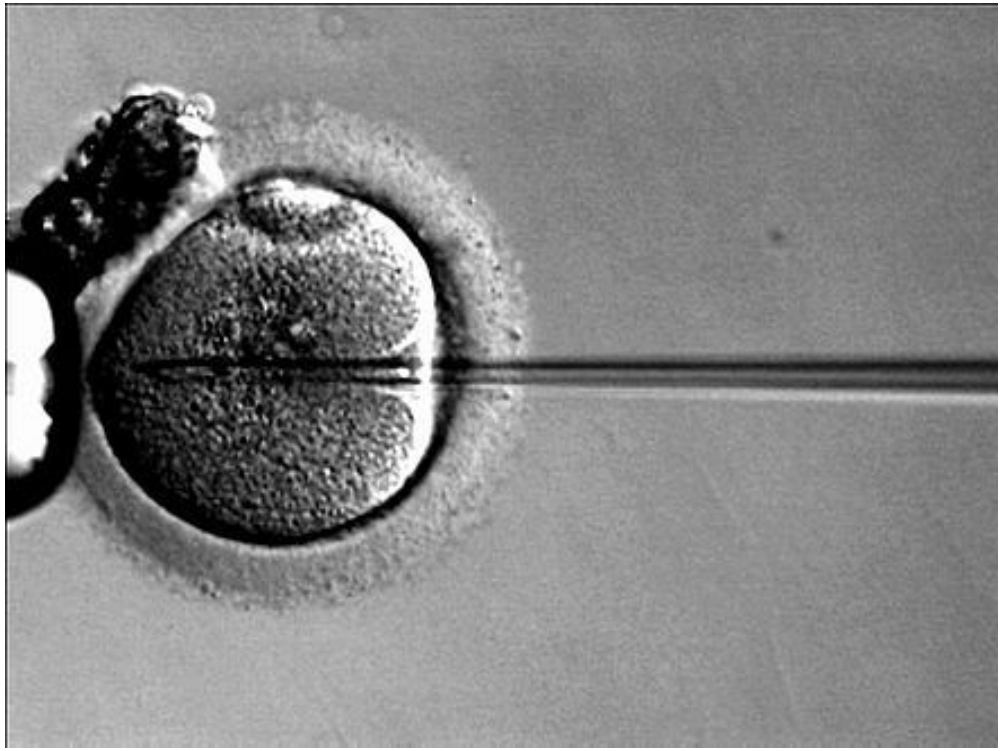






Infertility treatment

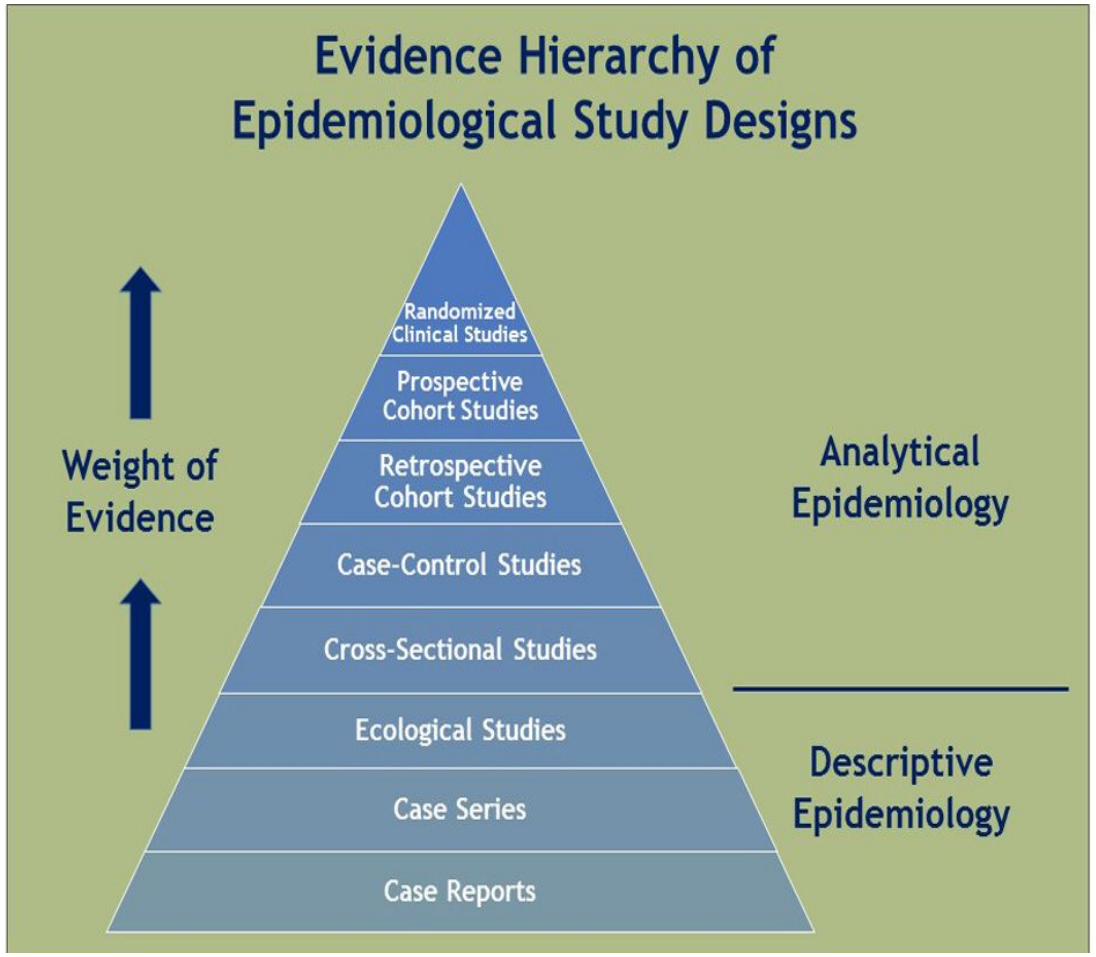
- **Intracytoplasmic Sperm injection**:
- Methods of reproductive medicine, such as intracytoplasmic sperm injection (ICSI) with previously conducted testicular sperm extraction (TESE), have led to men with Klinefelter's syndrome to produce biological offspring.
- By 2010, over 100 successful pregnancies have been reported using IVF technology with surgically removed sperm material from males with KS.



•Prognosis

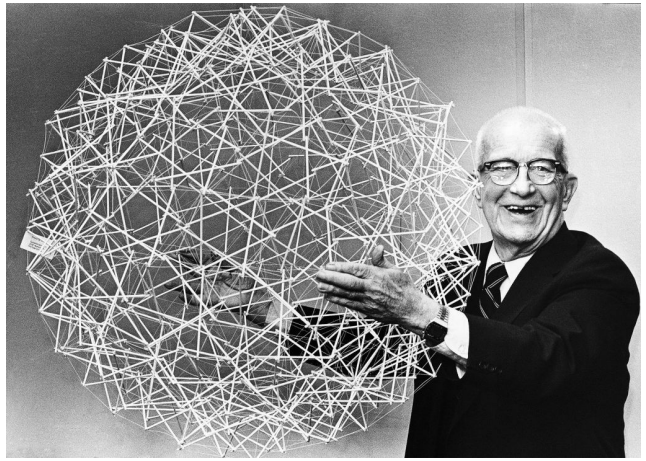
- The lifespan of individuals with Klinefelter syndrome appears to be reduced by approximately 2.1 years compared to the general male population.These results are still questioned data, are not absolute, and need further testing.

- **Epidemiology**
- This syndrome, evenly distributed in all ethinic groups, has a Prevalence of one to two subjects per every 1000 males in the general population.However, it is estimated that only 25% of the individuals with Klinefelter syndrome are diagnosed throughout their lives 3.1% of infertile males have Klinefelter syndrome. The syndrome is also the main cause of male hypogonadism



•History

- The syndrome was named after American endocrinologist :
- Harry Klinefelter, who in 1942 worked with Fuller.
- Albright and E. C. Reifenstein at Massachusetts General Hospital in Boston, Massachusetts.
- The account given by Klinefelter came to be known as Klinefelter syndrome as his name appeared first on the published paper, and seminiferous tubule dysgenesis was no longer used.



Questions for Other Members:

1. Explain Hemophilia?
2. Explain Neurofibromastosis ?
3. Explain Phenylketonuria Disorder?
4. Briefly explain Sick cell disease?
5. Define Turners Syndrome?
6. Briefly explain Color blindness ?

THANK YOU

- MEDICAL BIOLOGY{ GROUP 2}

THIS POWERPOINT PRESENTATION:

ЛАЗ-с-о-211(2) Карикуннан Шахзад