



Klinefelter syndrome is a **genetic condition** in **males** characterized by an **extra X chromosome (47, XXY)**, resulting from **meiotic non-disjunction**. It leads to **reduced testosterone production** and **abnormal sexual development**. The condition is **not inherited** and affects ~ 1 in 500 to 1,000 newborn males.

47 XXY

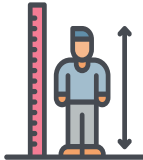


Meiotic non-disjunction

## CLINICAL FEATURES

### Growth:

- Infants appear normal at birth
- Tall stature**
- Long limbs
- Low upper-to-lower segment ratio



### Genitourinary:

- Hypogonadism**
  - Micro-orchidism
  - Micropenis
  - Hypospadias



### MSK:

- 5<sup>th</sup> finger clinodactyly
- Cubitus valgus**

### Neurologic:

- IQ below average to average**
- Intention tremor
- Behavior problems:**
  - Difficulties forming peer relationships
  - Poor judgement
  - Immaturity
  - Insecurity
  - Shyness



### Endocrinology:

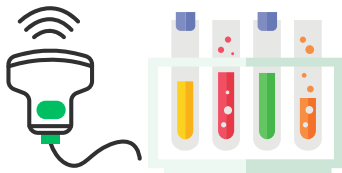
- Delayed puberty**
- Inadequate testosterone production
- Gynecomastia**
- Osteoporosis**
- Infertility**

### Oncological:

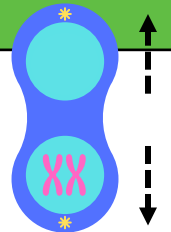
- Extragenital germ cell tumors
- Increased risk of breast cancer



## DIAGNOSIS



- Karyotype:** confirms an **extra X chromosome (47,XXY)**.
- Hormonal testing:** measures **testosterone, LH,** and **FSH** to assess testicular and pituitary function.
- Imaging studies:** ultrasound may assess testicular size and structural abnormalities.



## MANAGEMENT

- Hormone replacement therapy:** can help address low testosterone levels, improving physical development, energy levels, and mood.
- Psychological support:** counseling and support groups can help with emotional and psychological challenges.
- Educational support:** for learning difficulties.
- Physical therapy:** may help with muscle strength and coordination.
- Regular monitoring:** routine follow-ups to monitor health issues, including metabolic syndrome and osteoporosis.
- Fertility treatment:** assisted reproductive technologies, such as intracytoplasmic sperm injection (ICSI), may be considered later in adolescence or early adulthood.



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