Sex Chromosome Abnormalities , Sex Chromosome Aneuploidy

It has been estimated that, overall, approximately one in 400 infants have some form of sex chromosome aneuploidy. A thorough discussion of sex chromosomes and sex chromosome abnormalities. Additionally, the diagnosis may create for a young adult a potentially unique and more delicate set of psychosocial issues, as this diagnosis may come at a time when the individuals already struggling with a developing sense of self and sexuality.

Klinefelter syndrome (47, XXY)

A genetic disorder that affects males. Klinefelter syndrome occurs when a boy is born with one or more extra X chromosomes.

Most males have one Y and one X chromosome.
Having extra X chromosomes can cause a male to have some physical traits unusual for males

- A pair of sex cells fail to separate during the formation of an egg or sperm
- Embryo ends up with three copies of chromosomes
- (XXY)
- A recessive trait

How is Klinefelter syndrome diagnosed?

- Klinefelter syndrome usually is not diagnosed until the time of puberty.
- At this point, the boy's testicles fail to grow normally and may start to notice other symptoms.

- **physical symptoms of Klinefelter Syndrom**
- Low testosterone levels in the body. The degree of symptoms differs based on the amount of testosterone needed for a specific age or developmental stage and the amount of testosterone the body makes or has available.
- During the first few years of life, when the need for testosterone is low, most XXY males do not show any obvious differences from typical male infants and young boys. Some may have slightly weaker muscles, meaning they might sit up, crawl, and walk slightly later than average. For example, on average, baby boys with KS do not start walking until age 18 months.

- After age 5 years, when compared to typically developing boys, boys with KS may be slightly:
- Taller
- Fatter around the belly
- Clumsier
- Slower in developing motor skills, coordination, speed, and muscle strength
- Puberty for boys with KS usually starts normally. But because their bodies make less testosterone than non-KS boys, their pubertal development may be disrupted or slow. In addition to being tall, KS boys may have:
- Smaller testes and penis

- Breast growth (about one-third of teens with KS have breast growth)
- Less facial and body hair
- Reduced muscle tone
- Narrower shoulders and wider hips
- Weaker bones, greater risk for bone fractures
- Decreased sexual interest
- Lower energy
- Reduced sperm production

Turner syndrome (45 , X) : a condition that affects only females, results when one of the X chromosomes (sex chromosomes) is missing or partially missing. Turner syndrome can cause a variety of medical and developmental problems, including short height, failure of the ovaries to develop and heart defects.

Turner syndrome may be diagnosed before birth (prenatally), during infancy or in early childhood. Occasionally, in females with mild signs and symptoms of Turner syndrome, the diagnosis is delayed until the teen or young adult years.

Signs and symptoms of Turner syndrome may vary among girls and women with the disorder. For some girls, the presence of Turner syndrome may not be readily apparent, but in other girls, a number of physical features and poor growth are apparent early. Signs and symptoms can be subtle, developing slowly over time, or significant, such as heart defects.

Before birth

Turner syndrome may be suspected prenatally based on prenatal cell-free DNA screening — a method to screen for certain chromosomal abnormalities in a developing baby using a blood sample from the mother — or prenatal ultrasound. Prenatal ultrasound of a baby with Turner syndrome may show:

- Large fluid collection on the back of the neck or other abnormal fluid collections (edema)
- Heart abnormalities
- Abnormal kidneys

At birth or during infancy

Signs of Turner syndrome at birth or during infancy may include:

- Wide or weblike neck
- Low-set ears
- Broad chest with widely spaced nipples
- High, narrow roof of the mouth (palate)
- Arms that turn outward at the elbows
- Fingernails and toenails that are narrow and turned upward
- Swelling of the hands and feet, especially at birth
- Slightly smaller than average height at birth

- Slowed growth
- Cardiac defects
- Low hairline at the back of the head
- Receding or small lower jaw
- Short fingers and toes

In childhood, teens and adulthood

The most common signs in almost all girls, teenagers and young women with Turner syndrome are short stature and ovarian insufficiency due to ovarian failure that may have occurred by birth or gradually during childhood, the teen years or young adulthood. Signs and symptoms of these include:

- Slowed growth
- No growth spurts at expected times in childhood
- Adult height significantly less than might be expected for a female member of the family
- Failure to begin sexual changes expected during puberty
- Sexual development that "stalls" during teenage years
- Early end to menstrual cycles not due to pregnancy
- For most women with Turner syndrome, inability to conceive a child without fertility treatment

Causes

Most people are born with two sex chromosomes. Boys inherit the X chromosome from their mothers and the Y chromosome from their fathers. Girls inherit one X chromosome from each parent. In girls who have Turner syndrome, one copy of the X chromosome is missing, partially missing or altered.

The genetic alterations of Turner syndrome may be one of the following:

1- **Monosomy**: The complete absence of an X chromosome generally occurs because of an error in the father's sperm or in the mother's egg. This results in every cell in the body having only one X chromosome.
2- **Mosaicism**: In some cases, an error occurs in cell division during early stages of fetal development. This results in some cells in the body having two complete copies of the X chromosome. Other cells have only one copy of the X chromosome.