

Klinefelter Syndrome-XXY

Definition

Klinefelter syndrome is a chromosomal disorder that affects only males. People with this condition are born with at least one extra X chromosome. The syndrome was first identified and described in 1942 by Harry Fitch Klinefelter Jr., an American physician.

Cause/Etiology

Klinefelter syndrome is a condition in which one or more extra X chromosomes are present in a male. A problem very early in development results in an abnormal number of chromosomes. 60% of the embryos with Klinefelter's syndrome do not survive the fetal period. Mosaic Klinefelter syndrome occurs when some of the cells in the body have an extra X chromosome and the others have normal male chromosomes. These males can have the same or milder symptoms. Klinefelter syndrome is not considered an inherited condition. The risk of Klinefelter syndrome reoccurring in another pregnancy is not increased about the general population risk.

Males with Klinefelter syndrome appear normal at birth and have normal male genitalia. From childhood, males with the syndrome are taller than average with long limbs. Approximately 20-50% have a mild intention tremor, and uncontrolled shaking. Many males have poor upper body strength and can be clumsy. Approximately 1/3 of males have gynecomastia or breast growth, some requiring breast reduction surgery.

Incidence

Klinefelter syndrome is one of the most common chromosomal abnormalities. About 1 in every 500 to 800 males is born with this disorder; approximately 3000 affected boys are born each year in the United States. About 3% of the infertile male population have Klinefelter syndrome. The condition appears to affect all racial and ethnic groups equally.

Characteristics

Not all males with XXY actually develop the syndrome or its symptoms. In fact, many males show no abnormalities at all. However, for those who have developed KS, the following characteristics have been identified:

- Sterility (normal sexual function, but inability to produce sperm)
- Breast Development
- Incomplete masculine build; round body type
- Undersized testes
- Social difficulties (may be less confident, more immature, shy, passive, apathetic, sensitive, dependent, and have a fragile self-esteem)
- Learning difficulties
- Restless sleep patterns, yet difficult to awake in the morning

- Lower level of activity
- Lower level of endurance
- Hand tremors
- Frustration-based outburst
- Decreased growth of facial hair
- Large stature (average height is 6'1/2")
- Overweight
- Speech and language problems (receptive skills are higher than expressive)
- Difficulty learning to read and write

IDEA Category

Boys with Klinefelter syndrome may receive services under:

- Other Health Impaired
- Speech or Language Impairment
- Specific Learning Disability

DSM-IV-TR Category

There is no DSM-IV-TR diagnosis.

Long-Term Developmental Outcomes

Many men with Klinefelter syndrome go on to live normal lives, nearly 100% of them will be sterile. There is an increased risk of several systemic conditions including epilepsy, osteoporosis, such autoimmune disorders as lupus and arthritis, diabetes, and breast and germ cell tumor.

Assessment Approaches

Klinefelter syndrome is diagnosed by examining chromosomes for evidence of more than one X chromosome present in a male. This can be done during pregnancy or after birth with a small blood or skin sample

Interventions & Treatments

There is no treatment available as of the early 2000s to change a person's chromosomal makeup. Children may benefit from speech therapy for speech problems or other educational interventions for learning disabilities. Testosterone injections started around the time of puberty may help to produce more normal development including more muscle mass, hair growth and increased sex drive. Testosterone supplementation will not increase testicular size, decrease breast growth or correct infertility. Psychiatric consultation may be helpful when the boys reach adolescence.

Contributions of the School Psychologist

A school psychologist should be aware of the physical and emotional implications that these boys may experience. This disorder may manifest through speech or learning difficulties. Interventions may need to be put into place to help develop both academic and social aspects of these student's lives.

Additional Resources

American Association for Klinefelter Syndrome Information and Support (AAKSIS) -

<http://www.aaksis.org/>

Klinefelter Syndrome Support Group Home Page-

<http://klinefeltersyndrome.org/>

Klinefelter and Associates-

<http://www.genetic.org/ks/index.html>

National Institute of Health-

<http://www.nih.gov/health/chip/nichd/klinefelter>

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