



What is Klinefelter Syndrome?

Klinefelter syndrome is a genetic condition that occurs in males of all racial and ethnic backgrounds. It affects sexual development and may lead to learning disabilities. Klinefelter syndrome occurs in about 1 in 500 to 1 in 1,000 boys.

Michigan Resources & Support

Children's Special Health Care Services

Family Phone Line
Toll-free: 1-800-359-3722
www.michigan.gov/cshcs

Early On® Michigan

Toll-free: 1-800-EARLY ON
www.1800earlyon.org

Michigan Birth Defects Program

Nurse Follow-up Coordinator
Toll-free: 1-866-852-1247
E-mail: BDRfollowup@michigan.gov

National Resources & Support

American Association for Klinefelter Syndrome Information & Support

Toll-free: 1-888-466-KSIS
www.aaksis.org

Knowledge Support & Action

Toll-free: 1-888-999-9428
www.genetic.org

Family Village

www.familyvillage.wisc.edu

Genetics Home Reference

www.ghr.nlm.nih.gov/condition=klinefeltersyndrome

National Institute of Child Health and Human Development

www.nichd.nih.gov/health/topics/klinefelter_syndrome.cfm

How may Klinefelter syndrome affect my child?

Learning: Most boys with Klinefelter syndrome have normal intelligence. They may have developmental delays in some motor skills like learning to walk. They may also have learning disabilities, especially with speech, language, and reading.

Behavior: While personality and behavior vary, many children are described as being quiet, passive or shy. They may be immature for their age, and have difficulty with social skills. As boys with Klinefelter syndrome grow up, they may have concerns with body image or self-esteem. Emotional problems can include anxiety or depression.

Physical: The features of Klinefelter syndrome are often so mild that a person may not be diagnosed until there are concerns about sexual development or fertility. During puberty, increased body fat around the torso and breast enlargement (gynecomastia) may occur. Facial and body hair is often sparse without hormone therapy and the testes are usually small. Adult height may be taller than other family members.

Medical: There is an increased risk for health problems such as diabetes, thyroid disease, weak bones (osteoporosis), and breast cancer. Most men have normal sexual function, but are infertile.

How does Klinefelter syndrome occur?

Klinefelter syndrome is caused by an extra copy of an "X" chromosome. Usually males have one Y chromosome and one X chromosome in all of the body's cells. Boys with Klinefelter syndrome have a Y and two X chromosomes. The child is usually the first and only family member affected. Genetic counseling is recommended for parents to learn more about Klinefelter syndrome in their family.

How is Klinefelter syndrome treated?

Klinefelter syndrome cannot be cured, but some symptoms can be treated. Infants and toddlers (birth to 3 years) should be connected with *Early On*® Michigan if there are concerns about learning, speech, or behavior; while children over 3 years of age should be referred for special education services if concerns arise. Around the time of puberty, testosterone hormone levels should be checked. Many boys have low levels and will benefit from hormone treatment to reduce physical signs of Klinefelter syndrome. Assisted reproduction techniques have helped some men become biological fathers. Children with Klinefelter syndrome and their families benefit from having a primary care physician who helps to coordinate their care with medical specialists and other community-based services.

For more information, call Michigan's Genetics & Birth Defects Program toll-free at 1-866-852-1247 or e-mail Genetics@michigan.gov

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