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Klinefelter's Syndrome

A Simplified Guide

by Katherine Cummings

Article appeared in Polare magazine: January 2011 Last Update: October 2013 Last Reviewed: September 2015

Klinefelter (or Klinefelter's) Syndrome is the most common chromosomal variation found in men. Named for Harry Klinefelter who published the first reports on the condition in 1942, it was not until the late 1950s that it was established that an extra X chromosome (XXY rather than XY) was involved. The condition occurs as often as 1 in 500 to 1 in 1,000 births. Many men live out their lives without realising they possess the extra chromosome. Because of the negative connotations of the term "Syndrome", many medical researchers now prefer the term "XXY males".

XXY males may have breast enlargement, may lack facial and body hair and have a more feminine body shape than other males. They are often taller than their fathers and brothers.

XXY males may have breast enlargement, may lack facial and body hair and have a more feminine body shape (more rounded, less angular) than other males. They are often taller than their fathers and brothers.

These physical characteristics are sometimes less important in the long run than a degree of language impairment. This is not to suggest mental deficiency but XXY males may learn to talk later than other children and language impairment can affect their success in school.

There is no certainty as to the cause, or causes, of XXY conception. There is an increase in likelihood of XXY in women of advanced maternal age but this is slight. Although, as stated above, a male may never be diagnosed as XXY, the chances are greater for this to happen before or shortly after birth, during early childhood, during adolescence or in adulthood following an infertility test.

Some parents are concerned about an appropriate age to inform a child of the XXY condition and when to inform relatives (grandparents for instance). The consensus seems to be that by the time a child is ten or eleven he may be told there are differences in his anatomy. By the time he is twelve he can be filled in on details, depending, of course, on the individual's maturity. He should be prepared for the likelihood of infertility despite the fact that he will almost certainly be able to enjoy sexual activity.

There are variations on the XXY format (also referred to as the 47,XXY karyotype). The 48,XXYY syndrome is sometimes considered to be a variation of Klinefelter's although there is some conflict over this classification.

In some cases the XXY subjects may be raised as females, or may prefer to be reassigned as females when they are informed of their condition. Because they are often tall and delicately built they can sometimes make successful careers in modelling or acting. Caroline Cossey (author of *Tula, I Am A Woman*), who claims to have XXXY chromosomes, is a case in point, and one of the episodes of "House" skirted the issue with one of its cases concerning a female model whose mood swings and aggression were attributed to internal testes supplying a masculine level of testosterone.

According to Andrology Australia, men with Klinefelter's Syndrome will benefit from lifelong testosterone treatment.

References:

Parker, James N. and Philip M. Parker (eds.) *The official parents' sourcebook on Klinefelter Syndrome; a revised and updated directory for the Internet Age*. Icon Group International, San Diego, 2002

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[Andrology Australia](#) 

[National Institute of Child & Human Development](#) 

Polare Magazine is published quarterly in Australia by The Gender Centre Inc., which is funded by the Department of Family & Community Services under the S.A.A.P. program and supported by the N.S.W. Health Department through the AIDS and Infectious Diseases Branch. Polare provides a forum for discussion and debate on gender issues. Unsolicited contributions are welcome, the editor reserves the right to edit such contributions without notification. Any submission which appears in Polare may be published on our internet site. Opinions expressed in this publication do not necessarily reflect those of the Editor, The Gender Centre Inc., the Department of Family & Community Services or the N.S.W. Department of Health.

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