Puberty, sexual development and fertility

Most baby boys are born with a normal sized penis and testicles that have already descended into the scrotum. One boy, however, was born with an undescended testicle; in another boy, both the testes and the penis were small. Sexual development usually proceeds normally and puberty is generally unaffected by the extra Y chromosomes.
In some boys, however, signs of sexual maturation fail to develop fully at puberty. These boys may have a low testosterone level and boys with XYYY syndrome should be evaluated by an endocrinologist at puberty to determine if monitoring or treatment is needed.

Fertility

In five out of six reported adult males, infertility was present. Men with XYYY should be evaluated for fertility prior to attempting to have children as they may have low sperm counts.

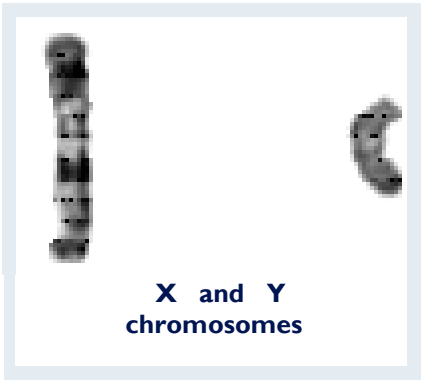
Why did this happen?

XYYY syndrome arises from the fertilisation of a woman's egg with a sperm containing an atypical number of Y chromosomes. Normal male sperm contain either one X or one Y chromosome, but in XYYY syndrome the sperm contains two or three Y chromosomes.

Sperm containing additional Y chromosomes can arise in a number of ways. Usually, it is due to a series of mistakes in sperm development where copies of the Y chromosome fail to separate from each other, giving rise to YY or YYY sperm. Sperm containing two Y chromosomes are found in over one per cent of samples from normal adult male sperm donors, so they are fairly common. The father himself usually has normal chromosomes but a mistake has occurred when his sperm were forming.

Another possibility is that the father has additional Y chromosomes as well, either having XYY chromosomes in all of his cells (XYY syndrome) or in just some of his cells that give rise to the sperm.

When YY sperm are involved in XYYY syndrome, the third Y chromosome is most likely to result from a mistake in the very earliest days when the embryo is developing after conception (when the baby is made). Typically when the mistake happens after conception, mosaicism is the result, so there may be cells with a single Y, no Y's, two Ys or four Ys as well.



X and Y chromosomes

Inform Network Support

Understanding chromosome disorders Unique

Rare Chromosome Disorder Support Group,
G1 The Stables, Station Road West, Oxted, Surrey RH8 9EE, UK Tel/Fax: +44(0)1883 723356
info@rarechromo.org | www.rarechromo.org

AXYS
info@genetic.org | www.genetic org

When you are ready for more information, *Unique* can help. We can answer individual queries and we also publish a full leaflet about XYYY syndrome.

This information is not a substitute for personal medical advice. Families should consult a medically qualified clinician in all matters relating to genetic diagnosis, management and health. The information is believed to be the best available at the time of publication and the full leaflet on which this text was based was checked by Nicole Tartaglia, MD, fellow in Developmental-Behavioural Pediatrics, UC-Davis Medical Center MIND Institute and Professor Maj Hulten, Professor of Medical Genetics, University of Warwick, 2005.

Copyright © Unique 2005

Rare Chromosome Disorder Support Group Registered in England and Wales
Charity Number 1110661
Company Number 5460413

Understanding chromosome disorders Unique

XYYY syndrome

rarechromo.org

What is XYYY syndrome?

XYYY syndrome, also called Triple Y syndrome or 48,XYYY syndrome, is a sex chromosome variation that only affects boys and men. It is extremely rare and there is little information available, partly because there may be boys and men who have XYYY syndrome who develop normally or with minimal problems and never come to the attention of the medical profession. This is believed to be true for 85 per cent of males with XYY syndrome, who have a single extra Y chromosome.

It seems that most boys and men with XYYY do not have major birth defects or handicaps and their long term outlook is fairly good. Nonetheless, the rate of behaviour, educational and medical problems among those who have been diagnosed is higher than average.

Chromosomes are the microscopic structures in the nucleus of the body's cells that carry our genetic information. In addition to 22 pairs of chromosomes numbered 1 to 22, boys and men usually have one X chromosome and one Y chromosome, making 46 in all. The Y chromosome is distinctive for males and is inherited from their father. Instead of a single Y, boys and men with XYYY syndrome have three Y chromosomes. They have 48 chromosomes: 22 pairs of ordinary non-sex chromosomes (called autosomes), making 44, plus four sex chromosomes (XYYY), making a total of 48 chromosomes.

Some boys and men have some cells with XYYY and other cells with different numbers of X and Y chromosomes. This condition is called mosaicism and does make it harder to be sure about the effects of the XYYY chromosome make up.

Common features

- **Vulnerability to behaviour difficulties**
- **Vulnerability to social difficulties**
- **Possible slight delay in mobility skills**
- **Possibly, slight effect on learning**

Development

Growth

Birth weight is typically in the normal range. Stature is usually tall normal with average height around six foot (180-185 cm), but this also depends upon family height.

Learning

Boys may need some support with their learning. Typically, they appear to have mild learning difficulties and can access the curriculum but work through it slowly. In terms of a measured IQ, the range found so far has been from 65 to 86, which would put most boys into the low normal category. Performance IQ is typically higher than verbal IQ. Most boys do better at manual, structural and conceptual work, such as computing, art, design and practical activities than at tasks that require high verbal fluency. Boys are also typically delayed in their hand control and use which undermines their practical skills.

Mobility and activity

A slight delay in sitting, crawling and walking is common but not universal. Boys typically first sit at 9-14 months, become mobile at 8-12 months and walk alone at 15 months to two years. Boys then may go on to be active and enjoy sports.

Muscle tone may be low (making the body feel floppy) and joints may be unusually mobile and loose. Supporting splints, braces or boots may then be helpful.

Speech and communication

Some speech delay is fairly consistent and speech and language difficulties are associated with other X and Y chromosome variations. Typically, first words emerge at 2 - 4 years. Many boys are helped by ongoing speech therapy. Speech delay may be overcome during the primary school years but some speech difficulties may persist. Despite this, adults can express their ideas and carry on conversations.

Behaviour

Typically, boys are vulnerable to social, emotional and behavioural difficulties. The nature of the behavioural difficulties varies, but boys may be immature, shy or lack emotional judgment. They may be prone to outbursts of temper. Boys with behavioural difficulties should be evaluated by a psychologist, psychiatrist, and/or developmental paediatrician, who can help provide guidance, intervention strategies and medications if necessary.

18 months old

Medical concerns

A 48,XYYY chromosome make-up does not appear to cause any major medical problems, but has been associated with some common medical and dental problems as described below.

Chest infections, asthma, coughs and colds

An unusual frequency of upper respiratory tract infections has been noticed in boys with XYYY syndrome. Coughs and chest infections appear to be more common in young children with chromosome disorders and in children with sex chromosome variations. Among older boys in *Unique* families, there is a high prevalence of asthma and atopy.

Teeth

Dental development is often imperfect in children with chromosome disorders and specifically sex chromosome variations. In boys with XYYY syndrome, poor enamel formation and discoloration have been reported as well as irregular or very large teeth. Boys should qualify for community dentistry and should practise excellent dental hygiene with regular brushing and flossing, and have regular dental check-ups.

Other features

These include radioulnar synostosis (fusion of the two long arms of the forearm near the elbow, limiting twisting movements of the lower arm and sometimes fully straightening the elbow); flat feet or bony foot deformities, minor kidney anomalies; acne.