47,XXX occurs when girls have an extra copy of an X chromosome in some or all of her cells. It does not cause them to look much different than their peers but does cause subtle physical differences including taller than average height, a curved little finger, a skin fold near the inner part of the eye and flat feet. Seizures, renal, genitourinary and cardiac abnormalities, and infertility can also be seen.

Trisomy X

Individuals with 47,XXX typically have intelligence in the normal range, although some specific learning disabilities and speech/expressive language delay are common and can be supported. Some individuals with Trisomy X syndrome have more significant health and developmental concerns, while others may have few if any notable features and do not even know that they have the condition.

What Causes 47,XXX (Trisomy X)?

47,XXX occurs when there are problems with formation of a parent's egg or sperm or when there are errors in cell division after an egg and sperm meet to form an embryo. The exact reason for these occurrences is not known. The risk of trisomy X increases with advancing maternal age.

What are the Health and Developmental Concerns Associated with 47,XXX (Trisomy X)?

It is important to remember that the signs and symptoms of 47,XXX can vary quite a bit from person to person. During pregnancy, the condition may be diagnosed through genetic testing such as a chorionic villus sampling (CVS) or amniocentesis or suspected through a

blood test such as cell free DNA analysis. Keep in mind that cell free DNA analysis cannot tell for certain if a baby has a variation in their number of X and Y chromosomes.

During the newborn period, girls with 47,XXX are often indistinguishable from other girls. During early childhood, low muscle tone and/or a delay in meeting developmental milestones may or may not be seen. During school years, a lag in language skills and academic difficulties may or may not be seen.

Intelligence is typically in the normal or low-normal range but mild intellectual disability can be seen. Psychological features including attention deficits, anxiety and mood disorders are not uncommon. Occasional medical issues can include genito-urinary or kidney malformations, seizures and infertility. Autoimmune problems such as thyroid disorders may be more common in women with Trisomy X.

It is believed that the majority of women with 47,XXX have gone their entire lifetimes without ever knowing they have an extra X chromosome. However, more people are learning that they or their baby have the condition due to prenatal genetic testing or during infertility testing.

If I have a baby with 47,XXX (Trisomy X), what is the chance I will have another baby with this condition?

In most cases, the chance that a woman will have another baby with an X and Y chromosome variation is thought to be less than 1% as the majority of cases of X and Y chromosome variation are not inherited.

Treatment for 47,XXX (Trisomy X)

There is no “cure” for 47,XXX as the genetic changes that cause the condition are typically present in some or all of the cells in the person’s body.

Individuals with 47,XXX often do not need any special treatment or intervention as infants. However, infants and children known to have triple X should have medical and developmental evaluations tailored to the features that can be associated with the condition for early detection if present. These include a renal ultrasound and cardiac evaluation as well as watch for staring spells or atypical movements. If any symptoms of 47,XXX such as learning disabilities or social/emotional difficulties arise during childhood, they should be managed with early intervention and support in school.

Teens and adult women with 47,XXX who experience menstrual irregularities or infertility

should be evaluated for hormonal abnormalities that could signal ovarian insufficiency. Other autoimmune problems including thyroid problems should also be considered.

How common is 47,XXX (Trisomy X)?

47,XXX (Trisomy X) is the most common variation is estimated to occur in one of every 1000 females.

Outlook

Most girls and women with 47,XXX will not be observably different from other girls and women. Some may be taller than average and some may have infertility. Some may experience difficulty in learning in some areas however, many individuals with 47,XXX have success in school, including higher education. Women with 47,XXX work in a variety of career settings. The majority of women with 47,XXX may have children on their own. Some may have children with the help of assisted reproductive technologies. Individuals with 47,XXX can expect to live full and productive lives.

Click here to learn more about scheduling a genetic counseling appointment for pregnancy-related questions.

Click here to learn more about scheduling a genetic counseling appointment for infertility or preconception questions.

Click here to learn more about scheduling a genetic counseling appointment for questions about pediatric or adult genetic conditions.

Resources

The Association for X and Y Variations (AXYS) https://genetic.org/

The Focus Foundation: http://thefocusfoundation.org/

Triple X Support Group https://rarediseases.org/organizations/triple-x-support-group/

Triple X Parent Support Group https://www.care.com/c/for/triple-x-syndrome