

**For EXtra Special BoYs**



**A Guide to 47,XXY,  
Klinefelter syndrome**



# Acknowledgements

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association for X and Y variations.

Additional information available at [www.axysinfo.org](http://www.axysinfo.org).

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## For parents

This booklet is written to help you understand and explain your son's genetic condition, 47,XXY, also known as Klinefelter syndrome. Emphasize to your son is that this genetic condition is quite common. One boy in 600 is born with an extra X chromosome. XXY is not an illness and it is not life-threatening, but it can have a number of impacts on his life. He will need to know how the extra X chromosome may affect his learning, his ability to play sports, and his growth and sexual development.

As the parent, you will need to support him if he has any of the challenges outlined in this book. While some boys with XXY have few symptoms, many boys do find that XXY affects some elements of learning or growth. Assure him that despite having two X chromosomes he is a boy.

It is best to begin the explanation of this difference in his body early in his life, even in pre-school, if he has required speech therapy or any other special service. For young children, you may say that his body has a few extra "messages". Speech and other therapies may be necessary to help him develop as other children do. You do not need to tell a child younger than 8 or 9 the name of the genetic condition, but you should always be truthful when talking with your son.

A boy with XXY should see a pediatric endocrinologist at around age 9 to begin monitoring his progress toward and through puberty. By this age, he needs to know enough about XXY to understand that it is a genetic condition, but not an illness. Otherwise, he may begin to worry when he goes for medical exams and lab work. You can begin to bring up possible issues with delayed puberty, if they become evident.

It is possible that he will require supplemental testosterone

during his teens. Try to learn about the indications for hormone treatment so that he can discuss this with you as well as his physician. You can also begin to discuss sexuality and human reproduction at a time appropriate to his development.

If he has learning disabilities or emotional difficulties, your role as a parent in supporting him is critical. Help him to advocate for any special education services he may need. Assist him with finding hobbies and sports that he enjoys and that make him feel competent. And let him know that even though he may be maturing more slowly than his siblings or peers, he is a valuable member of your family and that you will help him to become the most successful person that he can be.

This booklet is meant to be used along with the guide, *Living with Klinefelter Syndrome, Trisomy X and 47,XXY*. Pages 32–42 of the guide provide a detailed discussion about disclosure of the diagnosis and talking with your child about XXY. The guide will provide you with current information on 47,XXY. As your son matures, read portions to him, or let him read sections on his own and discuss them with you.

As he approaches high school, you will need to begin discussing the fact that his fertility may be lowered. Many boys with XXY mistake infertility for being unable to have sexual relations, so reassure him in this area. Let him know that he can certainly become a father, but that he may need to use an assisted reproduction method, such as in vitro fertilization, or donor sperm. He can consider adoption. Because XXY impacts sexual development, this booklet does discuss sexuality and reproduction. You may not want to read the section about puberty and reproduction to him when he is very young, but it is important to begin to discuss this material with him before puberty starts, at a level that he can understand.

# What is Klinefelter Syndrome?

47,XXY, also known as Klinefelter syndrome, is the name of something called a *genetic condition*.

In Klinefelter syndrome, which is easier to say as “KS”, a boy has an extra “X message” in each cell of his body. XXY is common. One boy out of every 600 boys has XXY. If you were in a big school, with 1200 students, 600 would probably be boys and 600 would be girls. You could expect to find at least one boy with XXY.

What makes XXY a chromosomal variation is that most boys have one X chromosome and one Y chromosome. You have two X chromosomes and one Y chromosome.

XXY is not an illness and it will not make you sick. It is caused by an extra X chromosome, which is one of the many messages in your body that tells your body how to grow and develop.

# What are chromosomes and genes?

The tiny structures in your body that determine how you develop and grow are called *chromosomes* and *genes*. They are so small that you need a microscope to see them.

Most boys have one X chromosome and one Y chromosome. You have two X chromosomes and one Y chromosome, so you have one extra chromosome.

Each chromosome has many genes and each gene has a separate function, helping to determine how tall you will be, how you speak, how you see, how your muscles function, and how your brain works. We can think of genes as holding messages for the body, telling it how to work and how to grow.

Genes are composed of DNA, which you can think of as an instruction book telling our bodies how to grow and develop. Genes are packaged in our body as chromosomes. The drawing below shows a chromosome composed of many different genes.

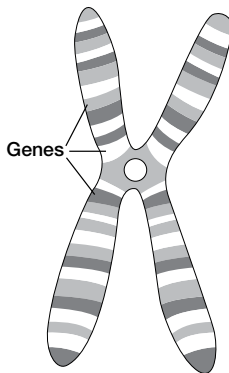


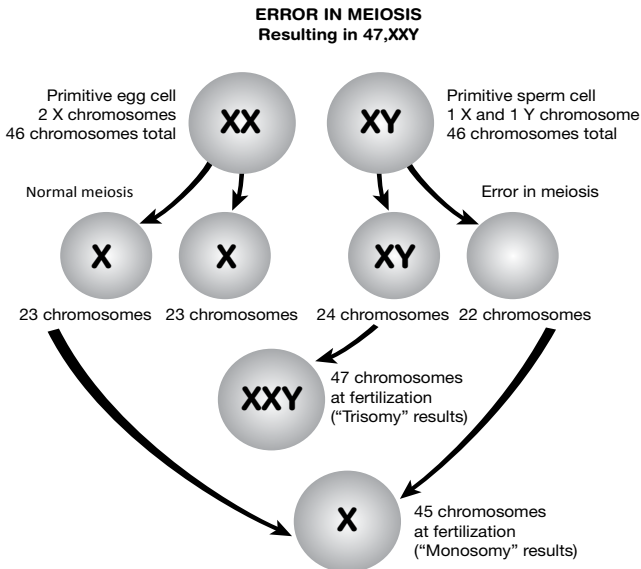
Illustration of an X chromosome. There are more than 1500 genes on the human X chromosome.

# How did this extra X chromosome happen?

The extra chromosome happens when the chromosomes divide to create either the egg (from the mother) or the sperm (from the father). The egg and the sperm combine in fertilization. This creates the tiny embryo that becomes a baby.

No one knows why the chromosomes can sometimes become “sticky” and instead of mom or dad passing down one chromosome from each, a second chromosome also was passed on to the embryo. Your mother or father could not have prevented the extra chromosome. The extra X chromosome can also happen when there is an error in cell division after fertilization.

This is an illustration of an error called non-disjunction that results in an extra X chromosome.



## How does the extra X chromosome affect growth?

In many boys with XXY, the extra chromosome may cause them to be taller than average. Sometimes they are taller than many of their classmates, or taller than their fathers.

But this is not always the case. There are also boys with XXY who are short.





# How does the extra X chromosome affect development?

All boys with XXY are different, just as each person is different. Some boys with XXY do not have any signs or symptoms. They learn to speak and crawl and walk at the usual times. Other boys, however, may speak at a bit later age. If you find that it is hard for you to say what you mean, you may be able to get additional help in school or from a speech therapist who can help you form words and make sentences.

When school starts, many boys with XXY will do well, but for some, it may be more difficult to learn to read, write or do arithmetic and algebra. This is because XXY can sometimes affect the way that your brain processes words and numbers. It is not because you are lazy or not trying hard enough! It just means that you and your parents and teachers need to find another way for you to learn. It may take longer for you to learn to do some things.

Schools are required to make a plan to give extra help to you if you need it. An example would be a 504 plan to give you extra time on tests. Another type of assistance is in an Individualized Education Plan, called an IEP.

As you move into higher grades, you may encounter difficult subjects where you find yourself struggling to keep up. If this happens, speak with your parents or teachers about additional help.

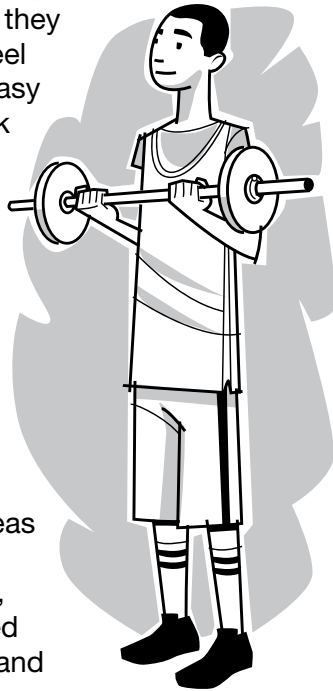
Many boys with XXY have reported that they do particularly well with music, art, computers, and work requiring a good sense of shapes and distances, such as graphics or carpentry. As you go through school, find those subject areas that you like and where you think that you would be happy working as an adult.

## XXY can also impact other areas of your life

Some boys with XXY find that they are not very good at certain sports, such as fast team sports like soccer or hockey. They can choose to enjoy other things, such as bike riding, or golf, or sailing. It is important for you to explore a number of activities to discover sports and exercise that you enjoy. Some boys with XXY find that their muscles and balance are not as good as other boys. Weight training and martial arts are good ways to help develop strength and balance, and can help to give you confidence.

Another area where some boys with XXY have concerns is that they may feel shy. Some may feel that they do not have an easy time making friends. Speak with your parents if this is a concern. You can get involved in groups that help you with learning what are called “social skills” so that you can have an easier time making friends and getting along in a group.

If you are worried about school, friends or other areas of your life, it is important for you to tell your parents, a teacher or another trusted adult. Don't keep it inside and worry about it on your own.



The extra X in no way affects the fact that you are a boy. Because of the Y chromosome, you have developed as a male with a penis and testicles. You will approach puberty between the ages of 10 and 14. Puberty is when your body begins to make male hormone, testosterone. All boys' bodies begin to change during puberty. Testosterone causes you to develop hair on your body. Your voice will begin to deepen. You will also begin to grow a beard. And your penis will also begin to grow in size.

With puberty comes sexual interest. Boys may begin to make sperm and will sometimes have ejaculations, often during night time dreams. All of this is completely normal and is preparing your body for adulthood.

The extra X chromosome, however, can cause puberty to be delayed or absent. Or puberty may not be complete. For this reason, your parents will have you see a physician. If puberty does not start or proceed normally, a physician can prescribe male hormone (testosterone) for you. Testosterone can be given as an injection or you can use a testosterone cream or gel.

Your doctor will do blood tests to determine if you are making adequate testosterone and how well your bones are growing. The doctor will also ask how you are feeling, how you sleep, and other questions.

Many older teens and adults will take supplemental testosterone because their bodies do not produce enough once they have gone through puberty. Testosterone helps them to build muscle. It can help prevent a problem later in life, called osteoporosis, in which bones lose strength, resulting in a greater risk of fractures. Not all boys or adults with XXY will need testosterone.

Another thing that you will want to discuss with your parents is that XXY usually dramatically lowers fertility. Fertility is your body's ability to make a baby with a woman that you love. Most boys with XXY produce sperm in their teen years, but as they age into adulthood, the number of sperm diminishes. Although adults with XXY are capable of normal sexual relations, they usually cannot become fathers through ordinary sexual relations with a woman. It will take the assistance of a doctor who specializes in helping men with lowered fertility.

Fortunately, there have been many improvements in assisted reproduction that now allow men with XXY to become fathers. These include freezing and preserving sperm for later use, in vitro fertilization procedures, and use of donor sperm to achieve a pregnancy. Men can also adopt to create their families.

These are important things to discuss with your parents and with your doctors. Materials are available through the website, [www.axysinfo.org](http://www.axysinfo.org), that provide information on advances in achieving pregnancies and parenthood.

Remember, most adults with XXY will tell you that they have happy lives. They can fall in love. They can also become parents, if they want to. Your parents and your doctors will help you to gain as much knowledge as possible about XXY, including fertility options.

You should also be aware that boys with XXY sometimes have a problem with teeth that decay easily. Brushing your teeth well twice a day and limiting sugary snacks and soda will help you to avoid tooth decay.

Genetic information is private. There is no reason for you to talk with anyone but your own family and your doctors about your extra X chromosome if you do not feel comfortable doing so.

The extra X chromosome is nothing to be ashamed of, and it is common. But it is a good idea to guard your genetic information, and to talk about it only to people with whom you are very close and whom you can trust. If you fall in love, you will want to tell this person about XXY. Don't give the information to people who may tease or hurt you, or try to bully you because of knowing that you have an extra X chromosome.





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