SEX CHROMOSOME PROBLEMS DISCOVERED THROUGH PRENATAL DIAGNOSIS

47,XXX Syndrome

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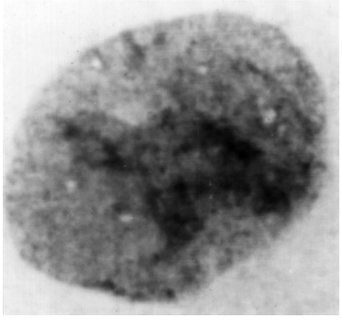
Introduction

The purpose of this booklet is to provide families with information about prenatal testing results that indicate a change in the sex chromosomes. This can be a very anxious time for families, and often little information is available to people who are not in the field of genetics. This booklet will provide you with some information, but it probably will not answer all of your questions. Please talk to a genetic counselor or a medical geneticist for more specific information about your situation. They are there to help. This booklet may also help you explain the test results to your family and health care providers.

I. Cells

The testing that was performed on your fetus was done by examining cells from either the amniotic fluid (if you had amniocentesis) or the placenta (if you had chorionic villus sampling). When the cells were studied, the packages of genetic information, called chromosomes, were examined. Your body, and your baby's body are made up of many billions of cells. Each cell contains a complete set of chromosomes. The cells of the fetus, placenta, and amniotic fluid, all came from the fertilized egg. Therefore, the chromosomes studied from amniotic fluid or the placenta match the chromosomes of the fetus.

This is a picture of one cell. It is so small that it can only be seen using a microscope:



II. Chromosomes

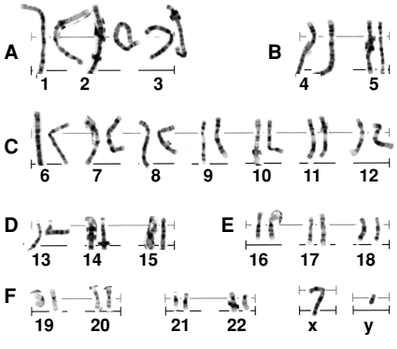
There are usually 46 chromosomes in each cell. They are like a set of cookbooks. Each chromosome or cookbook contains thousands of recipes that are pieces of information or instructions. These instructions or recipes are called "genes." Therefore, the chromosomes are packages of genes, which direct the body's development. For instance, there are genes that tell whether a person will have blue eyes or brown eyes. All of the information that the body needs to work is included in the chromosomes. The chromosomes contain the blueprint for growth and development. Scattered over the 23 pairs of chromosomes are about 30,000 genes. Even a very small piece of a chromosome contains many different genes. The precise location—or even the exact number—of all the genes is not known. Chromosome studies do not include a detailed examination of each gene.

Chromosomes come in pairs. One member of each pair comes from the father's sperm cell and the other comes from the mother's egg cell. In other words, the baby receives half its genetic material from the mother and half from the father. When the chromosomes are examined in the laboratory under the microscope, they look like this:

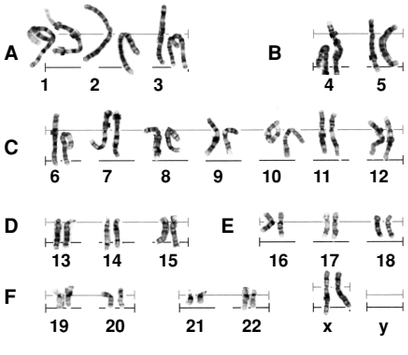
III. The Karyotype

In order to study the chromosomes, the cells obtained from amniocentesis or chorionic villus sampling are prepared in the laboratory so they can be seen under the microscope. The cells are photographed, the chromosomes are cut out of the photograph, and lined up by their sizes and characteristic light and dark banding patterns. Usually, a boy has an X and a Y chromosome and a girl has two X chromosomes. This picture is called a "karyotype."

This is a picture of a normal male karyotype:



This is a picture of a a normal female karyotype:



IV. Differences in Sex Chromosomes

Although most people have chromosomes that look like these photographs, some people have a different number of sex chromosomes. Others have a sex chromosome with an unusual structure. Most people with changes in their sex chromosomes are healthy, and look like other members of their families. Sex chromosome abnormalities are rarely diagnosed at birth. Unless the mother has an amniocentesis or chorionic villus sampling, it may be many years before the chromosome change is diagnosed. Sometimes it is never diagnosed. A sex chromosome change may be discovered when a child does not go through puberty normally, or much later, has trouble having children.

The most common reasons for studying a fetus's chromosomes are: the mother's age, a positive maternal serum screening test result, or an abnormal ultrasound evaluation. In these situations, the laboratory is mainly looking to make sure the baby does not have an extra "autosome." "Autosomes" are the chromosomes other than the sex chromosomes. As an example, babies with Down syndrome (trisomy 21), formerly known as "mongolism" have an extra chromosome 21 in their cells. Since all of the fetus's chromosomes are examined during a prenatal chromosome study, changes in the sex chromosomes are also detected.

There are several different types of sex chromosome changes: an extra or missing copy of the entire X or Y chromosome, an extra or missing part of the X or Y chromosome, or a rearranged X or Y chromosome. Changes in the number of chromosomes result from an error in the formation of the egg or sperm cell. When this happens, the other cells in the parents' bodies are normal. Neither parent needs to have a chromosome study of their own cells.

An extra, missing, or rearranged sex chromosome may be inherited from a parent, or have occurred for the first time during the formation of the egg or sperm. If a rearranged chromosome is discovered, your geneticist may recommend testing the parents' chromosomes.

Parents often feel guilty when their fetus has a chromosome change. However, nothing either parent did "wrong" caused the sex chromosome change.

V. What can these results mean for your fetus?

Most babies with changes in their sex chromosomes are healthy at birth, without serious birth defects. The sex chromosome change may not cause any obvious problems in your baby's early growth and development. Some people with sex chromosome changes never even learn they have a sex chromosome abnormality. But, there is a chance that there will be problems. Learning difficulties and emotional problems are more common in people with a change in their sex chromosomes. These problems cannot be identified prenatally or early in life. Other problems can occur, and depend on which chromosome change is present. Your physician or genetic counselor may recommend additional studies of your fetus. Since it is not possible to identify all birth defects before any baby is born, there will still be a risk for physical problems even if all the follow up studies are normal.

Parents often wonder if a change in the sex chromosomes means that the baby will be born with both male and female sex organs (ambiguous genitalia), or that their child will be homosexual. These conditions are no more likely to happen in a child with a change in the sex chromosomes than in someone with the typical sex chromosomes.

For some people, the additional risk for problems in the fetus is too high, and they choose to end the pregnancy. Others choose to continue the pregnancy. The decision whether to stop a pregnancy is a very personal one. It is usually a very difficult decision, and may take time to make. Each family must consider their own feelings and the risks. Your genetic counselor or health care provider is available to help you make the best decision for your family. Let them know how they can help.

47,XXX Syndrome

You have recently learned that your fetus has 47,XXX syndrome. You had probably never heard of this condition before you had prenatal diagnosis. You may now be making important decisions about how to proceed. This booklet was created to answer some of the many questions you may have.

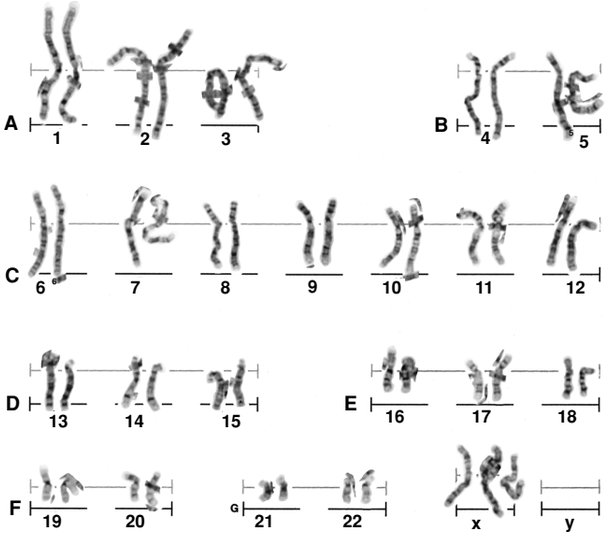
What is 47,XXX syndrome?

This syndrome is caused by an extra X chromosome in a female individual. Approximately one in 1,000 to 1,200 females has 47,XXX syndrome.

What causes 47,XXX syndrome?

Girls and women with 47,XXX have three X chromosomes instead of the usual two X chromosomes. This chromosome change is written as "47,XXX." This means that there are 47 chromosomes instead of the usual 46, and there are three X chromosomes instead of the usual two. The extra X chromosome was gained either during the formation of the sperm or egg that later came together to form the fetus, or during early fetal development soon after conception. The extra chromosome can never be removed. 47,XXX syndrome happens by chance. Nothing the parents did caused it to happen, nor could they have done anything to prevent it from happening.

This is a karyotype from a girl with 47,XXX:



What physical features are to be expected with 47,XXX syndrome?

Newborns and children with 47,XXX syndrome look like other girls their age. They are usually taller than other girls in their family and may be less coordinated. Women with 47,XXX syndrome are usually able to have children (fertile).

Each feature seen in 47,XXX syndrome can also be seen in girls and women with the usual number of chromosomes.

What mental or social features are to be expected with 47,XXX syndrome?

Of all the sex chromosome conditions, 47,XXX syndrome is the one most often associated with thinking and behavioral problems. An increased chance for speech and language problems can lead to delays in social skills and learning. Therefore these girls usually need additional help to succeed in school. In one small study of 11 girls who were diagnosed at birth and followed to see how they developed, fewer than half graduated from high school. Although these girls had friends in high school, they tended to act younger than other children their age. They did not like to participate in group activities and were more likely than their sisters to suffer from depression. In this small group, one girl went to college.

Is there any cure for 47,XXX syndrome?

The sex chromosome change that causes 47,XXX syndrome can never be repaired. A supportive family and help at school may reduce learning and behavioral problems.

Should I expect any complications during the rest of my pregnancy?

No. You have no higher chance of complications than the next person. The delivery and newborn period should be normal as well.

Will this happen again in future pregnancies?

Probably not. Your chance of having another baby with 47,XXX syndrome (or any other chromosome abnormality) is unlikely to be much greater than your age-related risk. Your other children, brothers and sisters, and other family members have no higher chance to have a child with 47,XXX syndrome or any other chromosome abnormality. Once a woman has a baby with a chromosome abnormality, she is often worried in the next pregnancy. Prenatal diagnosis will be available in any of your future pregnancies.

Other questions you may have for your genetic counselor or health care provider:

- How do I know the prenatal diagnosis results are accurate?
- What are my options in this pregnancy?

Ask them about anything that concerns you.

Are there support groups available?

Yes, and talking with others who have "been there" can be very helpful. Try:

Klinefelter Syndrome and Associates
PO Box 119
Roseville, CA 95678-0119
Phone: 916-773-2999
E-mail: ksinfo@genetic.org
Website: http://www.genetic.org

Children with sex chromosome problems are frequently followed through a genetics clinic. Call one of the phone numbers below to locate a clinic in your area.

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1999
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In the Pacific Northwest

There are genetics clinics and prenatal diagnosis clinics in Alaska, Idaho, Oregon, and Washington

Anyone can call for more information...
To find the clinic nearest you call:

Alaska:	907-269-3430
Idaho:	208-334-2235 x261
Oregon:	1-800-SAFENET 7233638
Washington:	253-395-6741

Or look at the PacNoRGG Directory of Genetic Services and Support Groups on the PacNoRGG web site:
http://mchneighborhood.ichp.edu/pacnorgg

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Copies of this booklet can be obtained from the PacNoRGG web site:
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