SEX CHROMOSOME PROBLEMS DISCOVERED THROUGH PRENATAL DIAGNOSIS

Turner Syndrome
45,X

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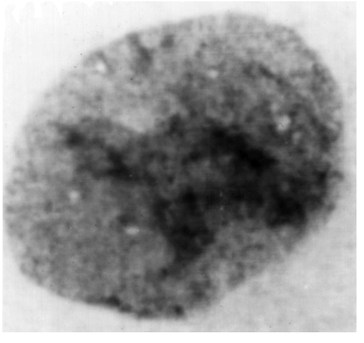
Introduction

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

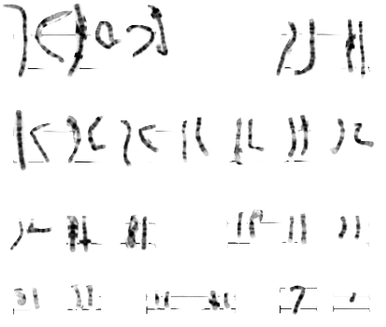
There are usually 46 chromosomes in each cell. They are like a set of cookbooks. Each chromosome or cookbook contains thousands of recipes that are pieces of information or instructions. These instructions or recipes are called "genes." Therefore, the chromosomes are packages of genes, which direct the 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:

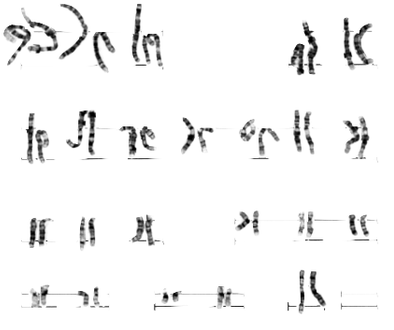
III. The Karyotype

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 picture shows a normal male karyotype:



This picture shows 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.

Turner Syndrome

You have recently learned that your fetus has Turner 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 Turner syndrome?

Turner syndrome is the name given to a set of physical features by a doctor, Henry Turner, MD. He named the condition before the underlying cause was known. We now know that the features observed by Dr. Turner are caused by a change in the sex chromosomes. Approximately 1 in 2000 baby girls has Turner syndrome.

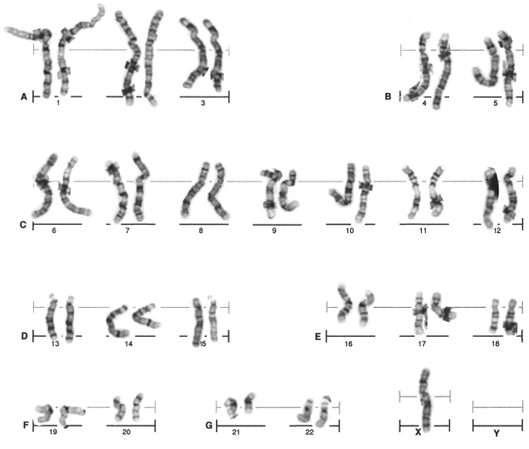
What causes Turner syndrome?

Girls and women with Turner syndrome have only one normal X chromosome instead of the usual two X chromosomes. The second sex chromosome is either missing or abnormal. When there is no second sex chromosome, the chromosome change is written as "45,X." This means there are 45 chromosomes, instead of the usual 46, and there is one X chromosome, instead of the usual two X's or one X and one Y. The missing sex chromosome was lost either during the formation of the egg or sperm that later came together to form the fetus, or during early fetal development soon after conception. The missing chromosome can never be regained. Turner syndrome happens by chance. The parents did nothing to cause it nor could they have done anything to prevent it.

Some females with Turner syndrome have one normal X chromosome, and one unusual sex chromosome. There are many possible variations. Sometimes the unusual sex chromosome is inherited from a parent. Other times a mistake happened during the formation of the egg or sperm that came together to form the fetus. Ask your health care provider or genetic counselor for more information about your fetus's chromosomes.

Occasionally, only some of the baby's cells are missing a sex chromosome; the other cells have two normal sex chromosomes. This is called "mosaicism." Babies with mosaicism can have some, many, or none of the features of Turner syndrome.

This is a karyotype from a woman with Turner syndrome:



What physical features are to be expected with Turner syndrome?

The features of Turner syndrome are variable from one person to another. Sometimes the features are noticeable at birth; other times they are not. Girls and women with Turner syndrome can have such mild features that the diagnosis is not made until adolescence or adulthood.

The diagnosis is sometimes suspected before birth due to certain ultrasound findings. Newborns with Turner syndrome may have physical features that make health care providers suspect the condition right away. There can be puffiness of the hands and feet, smaller than average overall size, and extra skin (or "webbing") at the neck. Some girls with Turner syndrome have heart or kidney abnormalities that need treatment. In childhood, girls with Turner syndrome usually remain smaller than other girls and may be more prone to ear infections and thyroid problems. Girls who are not diagnosed in childhood usually come to medical attention when the normal late childhood growth spurt fails to occur, and they do not start their menstrual periods or have breast development. Adult women with Turner syndrome are shorter than other women. The average adult height is 4'6" for women not treated with growth hormone during childhood.

Women with Turner syndrome have normal sexual function but most are unable to have children (infertile) because their ovaries do not develop completely.

Each feature seen in Turner 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 Turner syndrome?

For girls with 45,X or 45,X/46,XX karyotypes

The intelligence of girls with TS varies just like that of the general population and usually is in the normal range. Those with 45,X karyotypes, or with 45,X/46,XX mosaicism do not have a significantly increased chance of mental retardation. Girls with TS are more likely than other children to have specific learning disabilities, especially in nonverbal areas (sometimes called nonverbal learning disabilities or NLD). Commonly observed weaknesses include: difficulty imagining objects in relation to each other (this can affect driving), trouble appreciating subtle social cues such as facial expressions, problems with nonverbal problem-solving like mathematics, and clumsiness. Given early educational and social support, however, educational and career expectations for girls with Turner syndrome should be similar to those for their siblings.

For girls with other karyotypes

The prognosis varies if there are other chromosome changes. Ask your doctor or genetic counselor for information about your situation.

Is there any cure for Turner syndrome?

The sex chromosome change that causes Turner syndrome can never be repaired. However, some therapies are available. Girls can be given human growth hormone to increase final adult height. Two other hormones, estrogen and progesterone, are usually given in the early teen years to help girls with Turner syndrome develop a more feminine appearance. The infertility cannot be cured, but families can make a point of discussing alternatives to biological parenthood such as adoption or the use of an egg donor. With new advances in technology, some women with Turner syndrome are able to carry pregnancies using donated eggs. A supportive home environment may reduce learning and behavioral problems.

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

Possibly. Sometimes babies with Turner syndrome have extra fluid build up in areas of their bodies before birth. If this fluid build-up gets bad enough, the baby may not survive. The mother may also develop problems, such as high blood pressure. Also, certain heart and kidney abnormalities are more common in babies with Turner syndrome. Information on whether your baby has any of these problems may be important in managing the rest of the pregnancy.

Will this happen again in future pregnancies?

Probably not. If your fetus's karyotype is 45,X, your chance of having another baby with Turner 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 do not have an increased chance to have a child with Turner syndrome or any other chromosome abnormality. If the fetus's karyotype is different than 45,X, ask your health care provider to tell you more about your recurrence risks. 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:

14450 TC Jester, Ste. 260
Houston, TX 77014
800-365-9944
832-249-9988
Fax: 832-249-9987
Website: http://www.turner-syndrome-us.org
tssus@turner-syndrome-us.org

Children with sex chromosome problems are frequently followed through a genetics clinic or by a pediatric endocrinologist. Call one of the phone numbers on the last page to locate a clinic in your area.

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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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