Turner Syndrome: A Guide for Families

What is Turner Syndrome?

Turner syndrome happens when a girl is born missing part or all of one X chromosome. This is a change in the genes that happens by chance. It happens in about 1 out of every 2,000 to 2,500 baby girls born. It is not passed down from parents, so it is unlikely to happen again in the same family.

Girls typically have two X chromosomes in their cells. Girls with Turner syndrome have only one X chromosome, or part of the second one is missing. Some girls have "mosaic" Turner syndrome. This means some of their cells have two X chromosomes and some have only one.

What Are the Signs?

Turner syndrome can cause different problems. The signs depend on which cells are affected.

Signs in Babies:

- Swollen hands and feet
- Extra skin folds on the neck
- Heart problems
- Kidney problems

Some girls are not found to have Turner Syndrome until they are teenagers. Sometimes doctors find it before birth. Doctors can find Turner Syndrome before a baby is born in two ways:

- **Prenatal Ultrasound:** May identify certain organ issues seen in people with Turner Syndrome particularly involving heart, kidneys and neck
- **Blood test during pregnancy:** A simple blood test from the mom that looks at tiny pieces of the baby's DNA to check for certain genetic conditions

Common Signs in Older Girls:

- Short height (this happens in almost all girls with Turner Syndrome)
- Webbed neck or extra skin on the neck
- Low hairline at the back of the neck
- High roof of the mouth
- Increased angle at the elbows when arms are straight
- Curved fingernails or toenails
- Frequent ear infections
- Hearing problems
- Lack of puberty changes

Most girls with Turner Syndrome have normal intelligence and good reading skills, but some girls with Turner Syndrome have problems with math, especially shapes and geometry. Other girls with Turner Syndrome may feel shy or nervous around other people.

How Do Doctors Find Turner Syndrome?

Doctors commonly use a special blood test called a karyotype to diagnose Turner's Syndrome. This test looks at the chromosomes in the blood, specifically, the X chromosome.

How is Turner Syndrome Treated?

There is no cure for Turner Syndrome. But doctors can help with many of the differences it causes. Your child may need to see different doctors who work together.

For Short Height:

Without treatment, girls with Turner syndrome grow on average to be 4 feet 8 inches (56 inches) tall. Growth hormone shots can help girls grow taller. Growth hormone treatment may be used in girls with Turner syndrome whose height is below the fifth percentile on a growth curve.

For Puberty and Periods:

Many girls with Turner syndrome do not go through puberty on their own because their ovaries do not work the way they should. This means:

- Little or no breast growth
- No monthly periods
- Infertility

Many girls with Turner syndrome are treated with estrogen and progesterone to help with:

- Breast growth
- Starting monthly periods
- Strong bones

The doctor will start this medicine at the age when girls normally go through puberty.

Women with Turner Syndrome who desire pregnancy should discuss the safety and possibility of pregnancy with their fertility and heart doctors.

Other Important Care:

Your child needs regular checkups with:

- A heart doctor to follow up or monitor for heart problems or high blood pressure
- A kidney doctor if they have kidney issues

- A hearing doctor periodically to check for hearing
- A learning specialist if they need help with school and social skills

The Most Important Things to Know

- Turner Syndrome happens by chance. It is not your fault.
- Treatments can help your child grow taller and go through puberty.
- Your child will need to see different doctors throughout her life.
- With good medical care, girls with Turner Syndrome can live healthy, full lives.