Turner's Syndrome

Turner's syndrome is a congenital syndrome caused by a genetic abnormality in girls. Girls with Turner's syndrome are short and do not go through normal sexual changes at puberty. Intelligence is usually normal, but there is an increased risk of certain health problems. Hormone therapy can increase height and promote sexual development. However, most women with Turner's syndrome are unable to have children.

What is Turner's syndrome?

Turner's syndrome is a relatively common genetic (inherited) disorder, affecting about 1 in 2500 girls. Girls with Turner's syndrome have a chromosomal abnormality that interferes with the normal development of the ovaries.

Turner's syndrome results in certain physical abnormalities, most noticeably short height (stature). Other malformations may be present as well, including heart defects. Turner's syndrome may not be recognized until the early teen years, when affected girls don't go through the normal changes of puberty.

Growth hormone therapy may allow your daughter to achieve near-normal height. Treatment with the hormone estrogen can allow development of normal female sex characteristics but cannot restore fertility. Although close medical follow-up is needed to detect health problems, most girls with Turner's syndrome have a normal life span.

What kinds of problems occur with Turner's syndrome?

Physical problems. Girls with Turner's syndrome have certain physical characteristics, some of which may be present at birth:

- Swelling of the hands and feet.
- Loose skin at the base of the neck.
- Low birth weight/small size.
- Later in childhood, other signs may develop:
  - Short stature; this is sometimes the only abnormality until puberty.
  - Webbed neck; it looks wider at the base.
  - Low hairline at the back of the head.
  - Small jaw; prominent ears.
  - Broad chest.
  - Narrow fingernails.

For some girls with Turner's syndrome, the condition goes unrecognized until the early teen years.

- Most girls with Turner's syndrome don't go through the normal sexual changes at puberty (for example, breasts enlarging, menstrual periods starting).
- Females with Turner's syndrome are usually infertile (unable to bear children).
- *Medical problems.* Certain medical problems are possible, either in childhood or later in life:
  - *Heart problems.* Some girls with Turner's syndrome are born with heart defects, especially an abnormality of one of the heart valves (the aortic valve). A defect called aortic coarctation (narrowing of the aorta, the body's main artery) is more serious but less common. Aortic coarctation is more common in girls who have the typical "webbed neck" appearance of Turner's syndrome.
  - *Kidney problems.* Various kidney defects can be present at birth. These may cause problems with kidney function or contribute to problems with high blood pressure (hypertension).
  - *Thyroid problems.* Girls with Turner's syndrome can develop hypothyroidism (not making enough thyroid hormone).
  - *Ear and hearing problems.* Middle ear infections (otitis media) may be a recurrent problem. Risk of hearing loss is higher as your daughter gets older.
- *Developmental difficulties.* Certain types of learning disorders may occur in girls with Turner's syndrome. However, intelligence is usually normal.

What causes Turner's syndrome?

- Normally, girls have two X chromosomes (XX). In most girls with Turner's syndrome, one of those X chromosomes is missing. Less often, just part of the X chromosome is missing. Other girls have some cells in their bodies with two X chromosomes and others with one (mosaicism).
- In most cases, the chromosomal abnormality occurs at the time of conception; it is not passed on from the parents. In this situation, there is not a high increase in the risk of Turner's syndrome in future children.

How is Turner's syndrome diagnosed?

In most cases, the doctor first suspects Turner's syndrome because of abnormalities on physical examination, such as short stature or delayed puberty. A specific genetic test

called karyotyping can show the absent or missing X chromosome that causes Turner's syndrome. It may also provide important information for medical treatment and genetic counseling.

How is Turner's syndrome managed?

Medical testing and follow-up

- When Turner's syndrome is diagnosed, your child undergoes tests to detect other possible birth defects and abnormalities. These tests usually include an echocardiogram ("echo"), which uses sound waves to take pictures of the heart, and ultrasound scans to look for possible abnormalities of the kidneys and ovaries.
- We will probably recommend visits to some medical specialists, such as a medical geneticist (specialist in genetic diseases), a cardiologist (heart specialist), and an endocrinologist (specialist in hormone problems).
- Your child will have regular checkups to look for medical problems that can occur with Turner's syndrome, such as hearing problems, high blood pressure, high cholesterol, or scoliosis (abnormal curvature of the spine).

Hormone therapy

- For most girls with Turner's syndrome, treatment with human growth hormone can help them to reach a more normal adult height. For very short girls, growth hormone treatment may start in early childhood.
- Around the time of normal puberty, treatment with the female hormone estrogen may be recommended. This will help to provide a more normal female appearance (such as normal breast development). However, it cannot restore fertility.
- The endocrinologist will discuss issues with you related to when these two types of hormone therapy should be started.

Support

- Support groups and other forms of support can be helpful for girls with Turner's syndrome. Contact the Turner Syndrome Society at 1-800-365-9944, or on the Internet at *www.turner-syndrome-us.org/.*

When should I call your office?

Call our office, or the specialist managing your child's care, if you have any questions about Turner's syndrome or your child's treatment.

Please type your custom instructions and/or office contact information here.