

When is the diagnosis made?

Turner Syndrome is sometimes suspected when an ultrasound scan is performed during pregnancy. When a scan suggests Turner Syndrome, it can be confirmed by checking the baby's chromosome pattern. This can be done during pregnancy by either an amniocentesis test or a chorionic villus sampling (CVS) test.

Usually, however, the diagnosis is made later. It may be suspected shortly after birth because a baby has an unusually wide neck ('neck webbing'), puffy hands and feet or occasionally a problem with the heart. Often a girl with Turner Syndrome is not diagnosed until childhood because she is not growing as well as expected. Some girls are diagnosed as teenagers when they are taken to the doctor because their periods or puberty seem to be late in starting.

Features of Turner Syndrome

Turner Syndrome can have effects on many parts of the body. Some of the most common features are listed below. Most girls with Turner Syndrome would have some, but not all, of these things:

- Short stature (not so tall as other girls)
- Widening of the neck ('webbed neck')
- Ovaries that do not function and infertility
- An underactive thyroid gland
- A broad chest with widely spaced nipples
- A heart murmur, sometimes associated with narrowing of the aorta (the main blood vessel that comes out of the heart)
- A difference about the way the kidney is formed

Aspects of Turner Syndrome

Growth

Girls with Turner Syndrome tend to be short. Their growth rate may be normal for the first 2 or 3 years before slowing down. There are several ways to try and improve the growth of girls with Turner Syndrome and a girl will usually need to be referred to a child growth specialist so that her individual needs can be assessed and the treatment options discussed. Although girls with Turner Syndrome do not have growth hormone deficiency, growth hormone is often used to increase their final height.

Developmental progress

Girls with Turner Syndrome usually have normal intelligence and their progress at school is generally good. However, a small proportion of girls with Turner Syndrome may have specific learning difficulties. Sometimes they have particular difficulties with mathematics and geometry, but their reading age may be advanced. Activities involving dexterity eg, fine finger movements and co-ordination can occasionally be a problem.

Puberty

In girls with Turner Syndrome, the eggs in the ovaries degenerate and disappear in early childhood and the ovaries stop functioning properly well before the age that puberty would normally begin. The ovaries normally produce the sex hormones oestrogen and progesterone and it is oestrogen that is needed to start puberty. When the ovaries do not function, puberty will only occur if replacement oestrogen therapy is given. The great majority of girls with Turner Syndrome do not start their periods or develop the adult female body shape without the help of some hormone treatment. Oestrogen is used to start off breast development and progesterone and oestrogen together help produce regular periods.