



British Society for Paediatric Endocrinology and Diabetes

TURNER SYNDROME

The essential facts: Turner syndrome (TS) is a condition occurring exclusively in girls, and results from loss or abnormality of the second X chromosome from the body's cells. TS is named after Dr Henry Turner, an American doctor who described it in 1938. The main features affecting most girls with TS are short stature and underdevelopment of the ovaries. Other medical problems such as thyroid conditions, ear problems and certain heart defects are also associated with TS but it is very unusual for an individual girl to have all of them. Girls with TS can be considered 'normal' girls who can have more than their fair share of everyday medical problems.

Cause

Chromosomes are the strand-like structures contained in every cell, or 'building block', in the body and carry the genetic information that determines how an individual looks, feels and is made. Two of these chromosomes (X and Y) determine the sex of a baby when a woman's egg is fertilised by a man's sperm; male babies have one of each (XY) and female babies have two Xs (XX). Turner syndrome (TS) occurs when one of the two X chromosomes is lost or is abnormal so that it affects girls only. The cause is not known – it occurs randomly and if you already have a baby with TS, there is very little increased risk of future pregnancies being affected.

Major features

The two features affecting most girls with TS are:

1. Short stature: babies with TS tend to be small at birth, growth is slow during childhood and there is no pubertal growth spurt. Without medical treatment, the average height of UK women with TS is 143cm (4ft 8in). However, with growth promoting treatment during childhood, growth and final adult height can be improved.
2. Ovarian dysgenesis (abnormal development): in most girls with TS, the ovaries are only small streaks of tissue which produce neither eggs that can be fertilised by sperm to achieve pregnancy, nor the female hormone, oestrogen, which is responsible for puberty - the process when a girl changes into a woman. Oestrogen replacement therapy is therefore needed for puberty to happen in most girls with TS and throughout adult life to maintain these changes.

Other features

There are a number of other features of TS which may or may not be present.

1. Abnormalities of internal organs may occur before birth; some are more significant than others:
 - a. One of the main blood vessels in the body, the aorta, may have a narrowing (coarctation). This is seen in about 1 in 10 girls, is usually identified shortly after birth, and is corrected with surgery.
 - b. One of the valves in the heart may have two instead of the usual three 'flaps' although this is not usually associated with symptoms and no treatment is required.
 - c. In some cases, the kidneys are joined instead of being two separate organs; this is known as horseshoe kidney. It does not usually cause any symptoms and no treatment is required.
2. Some problems may develop during childhood or later in life
 - a. During childhood, many girls with TS have recurrent attacks of middle ear infection (otitis media) and 'glue ear'. Surgery to insert 'grommets' (ventilation tubes) may be required and any suspicion of hearing loss, which is more common in these girls and women, should be investigated promptly.



British Society for Paediatric Endocrinology and Diabetes

- b. Girls and women with TS have a tendency to develop high blood pressure (hypertension) so this should be checked regularly. Treatment may be required.
- c. The thyroid gland may become underactive. This is checked with a blood test and, if necessary, treated with thyroxine tablets.
- d. The bones of some women with TS may be thinner and weaker than unaffected women and, therefore, more likely to fracture (osteoporosis). This can be addressed with nutritional and lifestyle changes in the first instance.
- e. Some girls with TS have structural foot problems, such as short, broad feet and ingrowing toenails that can make it difficult to buy well-fitting shoes. Early foot-care advice from a podiatrist is helpful.
- f. Minor eye problems including squints are sometimes seen and referral to an eye doctor (ophthalmologist) may be needed.
- g. Some girls develop obesity and should be encouraged to eat a healthy, balanced diet and take plenty of exercise.

3. Physical features

A number of physical features are associated with TS. These include drooping of the eyelids, low-set ears, small chin, high palate (roof of mouth), neck 'webbing', broad chest with widely-spaced nipples, increased angulation of the outstretched arms at the elbows, narrow finger and toenails and multiple small dark birthmarks ('moles'). However, few girls are severely affected by several of these; in most cases, only a few are present, they may be subtle and not immediately obvious to the untrained eye. Overall, girls with TS don't look unusual.

4. Education & psychosocial characteristics

Intelligence in TS is 'normal' and like that of any other group of girls; some are high achievers, some struggle and need extra help and most fall somewhere in between. Some girls do have specific problems with number work, mathematics, and spatial tasks and it may be helpful to make teachers aware of this in case extra support is needed. However, many go on to further or higher education and enjoy a full range of job opportunities.

Girls with TS can be more socially vulnerable than other girls, having greater difficulty mixing with peers and making friends. They may stay living with their parents for slightly longer than average but most go on to live independently as adults, with or without a partner.

Treatment

• Growth promoting therapy

a. Growth hormone (GH)

GH therapy increases childhood growth and final adult height and is, therefore, standard treatment for most girls with TS in the UK. If possible, treatment is started at around five years of age and continues until growth is complete, usually around the age of 16 years. Treatment is given as a daily injection and is generally well tolerated. Current preparations have an excellent safety record.

b. Oxandrolone

The addition of a weak anabolic steroid tablet, oxandrolone, from 9 years of age has been shown to further increase final adult height. If given in a small, controlled dose, it is not associated with side-effects.



British Society for Paediatric Endocrinology and Diabetes

It is not possible to predict precisely how much height will be 'gained' by any individual girl in response to growth promoting treatment but, in general, a final adult height of 150cm (4ft 11in) is now considered an achievable target, with many girls exceeding this.

- **Oestrogen replacement therapy**

In the UK, this usually starts with a low dose tablet between 12 and 14 years of age and increases over approximately 3 years to mimic the normal process of pubertal development. An adult replacement dose is then given as a tablet or a patch throughout adulthood to maintain these developmental changes and to keep the womb (uterus), heart and bones healthy. Depending on the choice of treatment, most women will have a monthly 'period', where the lining of the womb is shed. Females with TS have a uterus and a vagina and can, therefore, expect to have normal sexual relations in adult life. However, because the ovaries do not produce eggs, most will require egg donation for pregnancy to be achieved and adequate oestrogen treatment is essential to prepare the uterus for this.

Management

During childhood, most girls with TS are cared for by a children's doctor specializing in growth and hormones called a paediatric endocrinologist. He or she will monitor growth, hormone replacement, blood pressure and will arrange referral to other specialists such as heart doctors (cardiologists), ear/nose/throat doctors, hearing specialists (audiologists) as required.

Girls with TS usually begin the transition to adult medical services once growth promoting treatment has stopped. It is very important that regular medical surveillance continues throughout adulthood so that any problems can be identified and treated early. This is usually coordinated by either a gynaecologist (a doctor specialising in women's health) or an endocrinologist (a doctor specialising in hormones). What discipline they come from matters little; what is important is that the person coordinating adult care is fully aware of the range of potential problems and has links with other specialists (for example, a heart specialist and someone with an interest in bone health).

Conclusion

TS affects many different areas of life and although it is associated with a number of medical problems, these can generally be addressed. Girls and women with TS live successful and fulfilling lives and, in many ways, should be considered no different from any other females.

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