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A midwives guide to Turner Syndrome (TS)

Abstract

Turner Syndrome (TS) is a cross cultural genetic disorder that affects around 1: 2500 women and is predominately associated with low growth rate and infertility. In *Classic TS* one sex chromosome is missing and is written as 45,X, whereas in *Mosaic TS* the whole chromosome is missing in some cells and not others. TS is characterised by short stature and non-functioning ovaries, which leads to impaired pubertal development and infertility. The majority of women with TS present as regular looking, have conventional personalities, and can be expected to live long and healthy lives. There is, however, a small cluster of physiological problems that can complicate the life of a woman with TS. Three occasions a midwife may encounter TS include: (1) Dealing with a childbearing woman who has received an in utero diagnosis of TS, (2) Recognising and managing a newborn with TS, and (3) Managing a woman with TS who as an adult has had successful assisted conception. To equip midwives with relevant knowledge, the objectives of this paper are to explain what TS is, how it may be recognized, and discuss medical and midwifery management of events.

Key words: Turner Syndrome, midwives, counselling, education, chromosomal anomaly

Keypoints

(1) Turner Syndrome (TS) is a cross cultural genetic disorder that affects around 1: 2500 girls and women.

(2) TS is predominately associated with low growth rate and infertility.

(4) The majority of women with TS live regular lives, with diagnosis moreoften becoming just one aspect of an otherwise normal life.

(5) With support from a knowledgeable and competent midwife, the childbearing woman, partner and family may be facilitated to adapt to their new situation.

A midwives guide to Turner Syndrome (TS)

Turner Syndrome (TS) is a cross cultural genetic disorder that affects around 1: 2500 women and is predominately associated with low growth rate and infertility (Carel et al, 2006).

Women with TS

Typically the nucleus of every human cell contains 46 chromosomes that cluster into 23 pairs. In *Classic TS* one sex chromosome is missing and is written as 45,X, whereas in *Mosaic TS* the whole chromosome is missing in some cells and not others. To view the chromosome organization of a woman with *Classic TS* (see *Fig 1).*

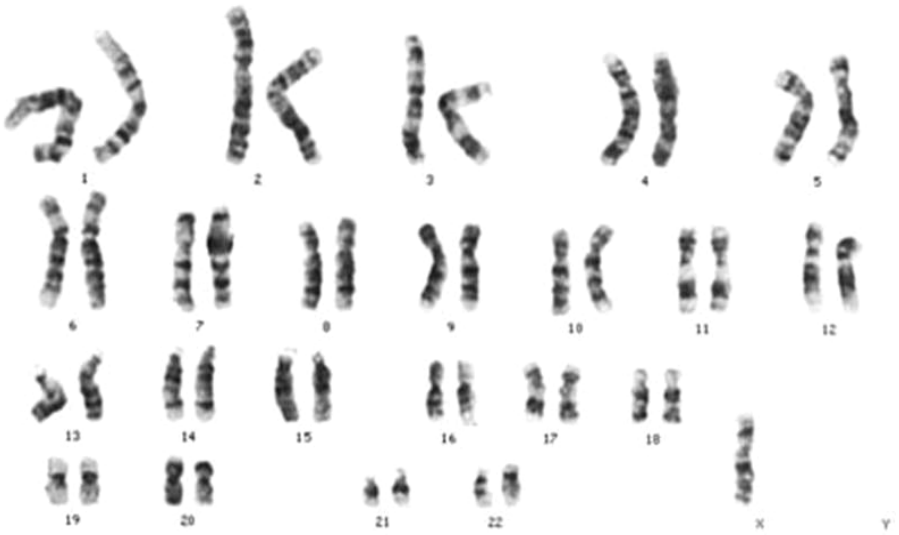

Fig 1: Chromosome organization in Classic TS (Smida, 2011)
(note second 23rd chromosome is absent)

Full or *Classic TS* always conceives a girl, whereas with *Mosaic TS* sex depends upon whether the cell line is 46,XX/45,X or 46,XY/ 45,X. Diagnosis of TS is confirmed through karotyping chromosomes from blood or skin cells obtained from a “scratch test”. If *Mosaic TS* is diagnosed, the first approach is to scan the baby and look for the apparent phenotypic sex. Only then parents should be informed as to whether they have a boy or girl. Due to absence of functioning ovaries *Classic TS* is a non hereditary condition (Charney, 1987).
If a women with TS becomes pregnant, it is either because she has Mosaic TS and has

managed to conceive in the conventional way or because donor eggs have been successfully implanted.

Women carrying a fetus with TS.

Ninety percent of women who conceive a fetus with TS spontaneously abort in the first trimester. The remaining 10% may develop utero hydrops which is associated with broad neck and puffy hands and feet that later develop spoon shaped nails. It is important for midwives to be vigilent to potential features of TS in newly born neonates, because in 10% of cases coarctation of the aorta occurs. Coarctation of the aorta involves narrowing of the aorta and can be corrected through surgery. Early recognition and diagnosis will facilitate access to experts who can diagnose complications and initate early treatment and remedies. The most common features to look for are webbing of the neck (extra folds of skin), poor feeding, oedematous hands and feet, low set ears, small jaw, arched palate, low hairline, droopy eyelids, short fourth toe, short fingers, broad chest and widely spaced nipples. To view clinical features and problems associated with TS see *Table 1.*

Table 1: Total array of clinical features and problems associated with Turner Syndrome

(1) Non-functioning ovaries associated with halted sexual development and infertility.
(2) Short stature (range 4 ft 3 inches to 5 ft 2 inches).
(3) Low set ears
(4) Webbing of skin around the neck (can be corrected with surgery).
(5) Small jaw and narrow high arched palate, which can source feeding and dental poblems.
(6) Arms that turn out slightly at the elbows (cubitus valgus)
(7) Soft spoon shaped fingernails that turn up at the end.
(8) Low hairline.
(9) Droopy eyelids.
(10) Short fourth toe.
(11) Short fingers.
(12) Broad chest.
(13) Widely spaced nipples.
(14) Puffy hands and feet.
(15) Appearance of non malignant moles, especially post growth therapy.
(16) Being overweight.
(17) Squints and premature aging of the eyes.
(18) Predisposition to middle ear infections and consequent problems with hearing.
(19) Heart defects occur in 10% of cases (e.g., coarctation of the aorta)
(20) Abnormalities of the kidneys and ureters increase risk of urinary tract infectons and hypertension.
(21) Increased risk of underactive thyroid.
(22) Increased risk of coeliac disease.

Some girls with TS have only one or two mild features, whilst others have several recognisable ones. Medical treatments can be used to alleviate many of these, with the overall outlook good. During childhood, TS is characterised by two main features: (1) short stature, and (2) ovarian dysgenesis. As a consequence of their regular appearance (see *Image 1*) and conventional personalities, many girls with TS remain undiagnosed until nonappearance of puberty and absence of menstruation has been acknowledged.

Image 1: Group of women with TS taken at a Turner Syndrome Support Society Meeting (TSSS) in the UK in October 2008 by Christine Charlton

Lack of breast development and initiation of menstruation consistent with peers typically incites disquiet in the young adolescent and arouses parental suspicion that something is amiss. Intelligence of girls with TS is usually within normal range, although some can experience difficulties with educational achievement and social adjustment (Williams, 1995). Children with TS have characteristic cognitive patterns, which present with nonverbal skills that are slightly subsidiary to verbal skills (Bender et al, 1989; Gordon and Galatzer, 1980; Silbert et al, 1977), with many capable of graduating from college. The majority hold down regular jobs, with their TS diagnosis just one aspect of an otherwise normal life.

Medical management of TS

During childhood, girls with TS are medically managed by either a general paediatrician or a paediatric endocrinologist who specialises in disorders of hormone secretion and growth in children. The co-ordinating paediatrician may also involve additional specialists when problems of the ear, nose, throat, eyes, feet and heart are detected.

In childhood, medical management involves improving growth and final adult height. Recombinant human growth hormone (Somatropin) is the contemporary hormone prescribed to promote growth (National Institute of Clinical Excellence, 2002). There are a few adult women with TS who have not had the benefit of treatments, because growth hormone was not available during their childhood or because diagnosis was made too late for them to benefit from hormone replacement therapy. With modern treatments focus has shifted from making the impossible possible, with final height often pulled up to the lower limits of normal range. The upper limit for stature in women with TS is around 5 feet 2 inches, with adolescents treated with growth hormone still on average 9-13 centimetres shorter than their peers (Allen, 2006). In contrast, without treatment a woman with TS will reach a maximum height of around 4 feet 8 inches.

Due to ovarian dysgenesis, oestrogen and progesterone are prescribed to induce puberty, to maintain uterine health and to instigate development of secondary sexual characteristics (National Institute of Clinical Excellence, 2002). It is potentially possible for a woman with TS to host and deliver an infant. Solutions involve egg donation and fertilisation using the partner's sperm, which is technically known as In-Vitro Fertilisation (IVF). The donated eggs and sperm are amalgamated in a test tube, with typical fertilisation resulting 12-15 hours later. The fertilised eggs are referred to as pre-embryos and an agreed number (usually 2-3) are transferred through the cervix into the uterus using a file. In preparation, specified hormone treatment is given to prepare the lining of the uterus for embryonic embedding.

Another method of assisted conception is Gamete Intra-Fallopian Transfer (GIFT). This procedure involves the donated egg and partner's sperm being transferred into one of

the fallopian tubes where fertilisation proceeds naturally. Once the embryo has been implanted into the uterus, the placenta will produce oestrogen hormones and additional hormone therapy can be abandoned for the duration of the pregnancy. Techniques like IVF and GIFT are expensive, stressful, time consuming and the waiting lists can be long. In some parts of the UK, IVF is not available on the NHS, and there may be a shortage of egg donors. A few women with TS (< 0.5%) do have egg producing menstrual cycles and the potential to be fertile. However, if a pregnancy is decided upon, there is an increased chance of the baby developing TS. Accordingly, genetic counselling is always recommended before embarking on a pregnancy. For this reason it may be considered preferable for a woman with TS to receive an egg donation and have IVF using her partner's sperm. Once conception and embedding have been successful, the pregnancy is managed by the maternity services in the customary fashion.

Midwives management of TS

There are three occasions where a midwife may encounter TS: (1) Dealing with a childbearing woman who has received an in utero diagnosis of TS, (2) Recognising and managing a newborn with TS, and (3) Managing a woman with TS who as an adult has had successful assisted conception.

(1) <u>Dealing with a childbearing woman who has received an in utero diagnosis of TS</u>

When an intrauterine diagnosis of TS has been made, the parents will require information from which they can construct a meaningful picture of what their baby will be like. The genetic counsellor will fulfil this role. The goal of the midwife is to help parents gain understandings about what having a baby with TS means and let them ask questions and make informed choices about their future. A series of pamphlets and booklets about TS should be kept in the maternity unit to accommodate events such as an intrauterine diagnosis. There are several organisations that produce information packs for childbearing women in this situation. For example, *The Child Growth Foundation* produces a free booklet called *The Turner Woman: a Patient Guide,* which hosts a great deal of relevant information.

There is also the *Turner Syndrome Support Society* (TSSS), which maintains an extremely facilitative website (www.tss.org.uk). They produce many accessible booklets about TS and host events and chat groups where women with TS and their significant others can share experiences.

It is important to emphasise that midwives should not give women detailed genetic counselling about TS. It is the midwives role to know and understand enough to empathise if a TS diagnosis is suspected or confirmed. An appointment with the genetic counsellor or fetal medicine specialist are required. The job description of the genetic counsellor is to help the woman, partner and family understand and adapt to the medical, psychological and familial implications of TS to disease. This process integrates interpretation of family and medical histories to educate about inheritance, testing, management, prevention, resources and research, and provide counselling to promote informed choices and adaptation to the condition.

For a childbearing woman receiving an alien diagnosis, it is important for the midwife to reinforce the views of specialists by painting an accurate picture of what may be expected. It is really important for the midwife not to construct a picture of TS that portrays a baby that consists of a collection of “deficits”. It is more meaningful to construct a picture of “individual differences”. The key points to emphasise are that some women without TS experience infertility and possess short stature. An emphasis on “difference” can obscure the fact that an individual with TS and those without are more similar to one another than different.

Women with TS are a very diverse group with a wide range of skills and abilities. It is important to explain to the couple that their child may have only a small number of features. Also, it is important to reasure parents that nothing they did instigated TS to develop. It was purely random chance that caused the chromosonal anomaly. How detailed an explanation of karotyping is given is dependent upon the genetic counsellors assessment of the couples interest and ability to comprehend what is being told. A list of key points that the counsellor should cover with the woman and partner are provided in *Table 2*.

Table 2: Key points to inform parents about TS

What TS is?
What causes TS?
How common TS is?
How TS is diagnosed?
What the main features of TS can be?
How TS is treated and managed?
Names and contact numbers of support groups.
Name and contact number of a genetic counsellor.

Having a pre-prepared pack of accessible information to take home would be tremendously helpful to both the couple and maternity care staff.

(2) Recognising and managing TS in the newborn

Many infants born with TS pass through the system unnoticed. However, a few present with complications that require immediate attendance. Where suspicion of a TS diagnosis is aroused the midwife should call the paediatrician who will take a maternal blood sample for karotyping. It is imperative to keep the parents informed of the processes leading up to diagnosis and to provide them with information about TS prior to diagnosis being confirmed. Once the diagnosis is secured, the genetic counsellor is called and important information leaflets and contact numbers given to parents.

It is also the role of the midwife to be involved in care of the neonate with TS. Dependent upon complications that transpire, the neonate may be transferred to the Special Care Baby Unit (SCBU) where paediatric care plans may be written. For example, a feeding regimen or cardiac care may be required.

(3) Managing a woman with TS who has had successful assisted conception

Once conception has been successful, antenatal care should be delivered according to the prescribed pathway of care depending upon whether it is a singleton or multiple pregnancy.

Conclusion

This paper has concentrated on providing midwives with information to prepare them for the role of working with parents of a baby with TS and the neonate itself. Due to the word limit,

some aspects of TS have been omitted; for example, providing more information about physical and psychological aspects of females with TS as young children, teenagers and adults. Examples of these omissions include subjects such as fear of romantic rejection and social isolation due to infertility which has been identified in some women with TS (Rolstad et al, 2007; Sutton et al, 2005). As a consequence, women with TS are generally older when they have their first sexual encounter (Boman et al, 1998; Carel, 2006; McCauley et al, 1994; Pavlidis et al, 1995; Rolstad et al, 2007). They are also less inclined to establish steady relationships with men and are generally less sexually active than their non TS peers (Boman et al, 1998; Naess et al, 2010; Pavlidis et al, 1995; Rolstad et al, 2007). Pavlidis et al (1995) reported that out of (n = 80) women with TS, only 55% reported having sexual relations by the age of 34. Carel et al (2006) testimonies that by 20 years of age only 30% of women with TS have had sexual experience compared with 85% of the general population (Spira et al, 1993). This evidence informs us that women with TS sometimes develop attachment avoidance strategies and that they may benefit from counseling in early adulthood. Although it is not the role of the midwife to discuss these themes, further reading will extend knowledge and horizons. Support provision from a knowledgeable and competent midwife will facilitate the childbearing woman, partner and family to adjust to their new situation.

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