The Turner Woman: A Patient Guide


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CONTENTS

TURNER SYNDROME

The lack of, or abnormality of, the Second X chromosome produces Turner Syndrome. It affects only females. Such women are likely to be short and lack ovaries that function correctly. There are other features that are common to the condition, but rarely do all occur together in one individual.

Introduction

This booklet compliments one entitled "Turner Syndrome: A Guide for Patients and Parents". Although that booklet was written mainly for children and adolescents with Turner Syndrome, there are many other important points that are relevant to adult women with Turner Syndrome and it may therefore be helpful to read both booklets. Although many of the medical problems that arise in Turner Syndrome have their origins in childhood, some do not arise until adult life.

Much of the medical management of Turner Syndrome in childhood is to do with improving growth and final adult height. There are many adult women with Turner Syndrome, however, who have not had the benefit of treatment, either because it was not available during their childhood or because their diagnosis was made too late for them to benefit from any treatment that might have helped with their growth. Of course, the upper limit for stature in women with Turner Syndrome is 5'2", which is well within the normal range. However, in contrast, there can be enormous difficulties for an adult with a height of only 4'3".

"Of course, I would prefer to be taller, and I am pleased that younger Turner girls may benefit from growth hormone treatment. Sometimes I wished it had been available for me when I was younger".

It is important that women with Turner Syndrome continue to have medical support and this can involve different specialties in medicine, particularly endocrinology and gynaecology, as well as for other aspects such as cardiovascular problems, high blood pressure, thyroid function and diabetes. It is recommended that there should be one physician who co-ordinates a patient's care. The importance of this being a physician who has both an understanding of the associated conditions and interest in women with Turner Syndrome is emphasised in the "Patient's Story" which is included towards the end of this booklet.

Most women with Turner Syndrome should be able to live a fully active life in all aspects. It is important when reading this booklet to realise that not all the problems are applicable to each individual. The aspects which relate to you should be discussed with you by your specialist.

"Until recently there was very little chance, of advancing final height, or improving fertility for Turner Syndrome women. Now the focus seems to be changing from the impossible to the possible".

What is Turner Syndrome?

The Turner Syndrome was first fully described by an American Dr Henry Turner in 1938. Turner Syndrome (TS) is a chromosomal condition affecting approximately 1 in 2,500 live female births. The diagnosis is confirmed by examination of the chromosomes from a blood sample (karyotype). Turner Syndrome is usually characterised by short stature and non-functioning ovaries leading to impaired pubertal development and infertility. There is no increased risk of mental retardation in Turner Syndrome girls whose intelligence spans the normal range. The physical features associated with Turner Syndrome may include webbing of the neck (extra folds of skin); nail abnormalities; puffy hands and feet; coarctation of the aorta (constriction or narrowing of the main artery from the heart which can be corrected with surgery). Feeding problems may occur in early childhood and sometimes there are learning or behavioural difficulties which may require professional help.

It must be emphasised that some girls may have only one or two mild features of the syndrome while others may have several easily recognisable ones.

Care of the Turner Woman

During childhood, girls with Turner Syndrome are usually looked after by either a general paediatrician or a paediatric endocrinologist (a specialist in disorders of hormone secretion and growth in children). The co-ordinating paediatrician may also involve other specialists who are concerned with problems of the ear, nose and throat, eyes, and the heart, as required. Hopefully, the diagnosis of Turner Syndrome will have been made at birth or in early childhood, which makes understanding and acceptance of her condition easier. As the child with Turner Syndrome grows older, her parents, with the specialist, will gradually be able to answer their daughter's questions and supply the necessary information about the implications of this condition. When the diagnosis is made in the teenage years, it is unlikely that treatment for her growth will significantly improve height, and the impact of the diagnosis at this age is hard to cope with for the parents as well as for the adolescent. Some of the difficulties which may occur with late diagnosis are highlighted in the "Patient's Story" at the end of this booklet. At the age of about 16 years, girls with Turner Syndrome are usually transferred from the care of a physician who specialises in children's medicine (paediatrician) to a physician who cares for adults. Unfortunately, in many areas, there is often a large gap between services for children and adults and it may be advisable that you ask your paediatric specialist for a referral to a specific adult physician with a special interest in Turner Syndrome.

Adolescence is a critical time in life, and this can be especially so when there is a condition such as Turner Syndrome which makes the individual feel "different" from her peer group, either through lack of height or lack of sexual development. The support and transfer of medical care during adolescence is extremely important and can be helped if it is done as a gradual process with dual responsibility between paediatrician and adult physician for a few years. It is important that the adult physician is someone who is knowledgeable about Turner Syndrome, especially with regard to oestrogen replacement and fertility as well as the other associated medical conditions. Although adult services may be provided by a general physician, endocrinologist or gynaecologist, the most likely choice would be a reproductive endocrinologist.

It is extremely important to emphasise to the adolescent that there are many aspects of the medical care of a woman with Turner Syndrome that may need attention and it is very important that, with the gradual gaining of an adolescent's independence, they do not "drop out" from medical care. This is particularly important because of the change of emphasis from growth, which predominates during childhood, to oestrogen replacement, maintenance of bone strength and fertility, which are the important medical features of adult life.

"Being smaller than average is boring. People sometimes make assumptions about me purely because of my height. Everyday situations are naturally geared to the 'norm', so for me, many things are just that little bit out of reach – either too high or too deep. (I am tired of being rescued from chest freezers!). Clothing is one area that seems to be improving. Many more stores now carry Petite ranges so that it is perfectly possible to find clothes in proportion.These petite clothes however, hang on rails impossible for the "petite" person to reach, unless able to pole vault."

General Problems

These are discussed in the booklet "Turner Syndrome: A Guide for Patients and Parents". Many of the problems encountered with Turner Syndrome are there at birth and have usually been dealt with during childhood and adolescence. These include:

- **Hearing** (middle ear infections).
- **Eyes** – squints. Also, the normal aging of the eyes may occur at an earlier age than usual and regular eye checks are therefore advised.
- **Heart defects** (about 10%).
- **Short stature.**
- **Nails** – spoon shaped and can be difficult to cut.
- **Webbing of the neck**, (which may require corrective surgery).
- **Moles** are more common in Turner Syndrome (especially after growth hormone therapy). However, these moles do not seem to have an increased incidence of being malignant.
- **Weight** – being overweight is often a problem for women with Turner Syndrome and

this is made more obvious by their short stature, If excessive weight gain occurs, it is important to be certain that the thyroid gland is working normally. There is an association with other conditions such as the thyroid gland working abnormally (usually under-secreting thyroid hormones) and poor absorption of food (Coeliac disease).

- **Glucose intolerance** – there is an increased risk of developing diabetes mellitus and it is therefore recommended that glucose levels in urine are monitored annually.
- **Abnormalities of the kidneys** and the drainage tubes to the bladder may be present and seen on an ultrasound scan. This may be associated with an increased number of urinary tract infections and/or an increased risk of hypertension (high blood pressure).

It should be emphasised that it is the overall care of the woman that is important in Turner Syndrome and there are many aspects of the condition that occur during childhood which may become more important during adult life. Understandably, one of the problems that some women with Turner Syndrome find hardest to come to terms with relate to the failure of the ovaries to work satisfactorily. This means that they do not produce the female hormones or eggs which means that they will not be fertile. The lymphatic system (small drainage channels like veins) may be inadequately formed in the hands and feet so that in adulthood the feet may swell and accumulate fluid. The common abnormalities in the kidneys and/or the heart (coarctation of the aorta – narrowing of the large blood vessels leaving the heart) may result in high blood pressure, particularly when treatment with oestrogen is started. Also, there may be problems with diabetes mellitus and with thyroid function.

Educational difficulties may often be a problem during childhood, although intelligence falls across the normal range. Difficulties with hand-eye co-ordination (so that writing is a much more difficult problem than reading) will continue into adult life. Although verbal skills are good, there may be difficulties with abstract thoughts, planning skills and visual-spatial tasks, which become apparent in mathematics. Women with Turner Syndrome are just as capable of a university education as anyone else, but they are unlikely to chose mathematics as a main subject. For those women who feel they may not have achieved their potential academically, help from a Community Adult Education Service could be of great value in improving academic performance. If you would like to know more about this, then you should discuss this with your specialist, and/or contact your local Adult Education Department.

Intelligence is across the normal range in Turner Syndrome and it is important that such girls or women are given the help and support they need to fulfil their potential.

The Chromosomal Abnormality

Humans have forty six chromosomes which consist of twenty three pairs. A chromosome is a collection of genetic material (called DNA) which carries all the information that

enables humans to be the individuals they are, One pair of these chromosomes are called the sex chromosomes. If both sex chromosomes are an X (ie. 46XX) then this codes for a female. If the pair of sex chromosomes contain an X and a Y chromosome (46XY) then this codes for a male. Loss of one of the X chromosomes in a female (45X0, the 0 representing the missing chromosome) causes Turner Syndrome and such girls/women may have many, or only some, of the problems that are listed in this and the previous booklet "Turner Syndrome: A Guide for Patients and Parents". 45X0 Turner Syndrome is called "classical Turner Syndrome" and accounts for approximately 50% of girls with Turner Syndrome. The other 50% have some cells which have the two X chromosomes and are normal, but other cells which are either missing all X chromosome or where there is a partial abnormality of one of the X chromosomes. In practical terms, it makes little difference as to the exact abnormality of the chromosomes, except in one instance as discussed below.

It is important to emphasise that women with Turner Syndrome should have no doubt of their femininity – physically, behaviourally and sexually.

Although the diagnosis of Turner Syndrome can often be made by an expert who recognises the physical characteristics, the diagnosis has to be confirmed by assessing the chromosomes in the blood (karyotype). The main reason is to allow your physician to make certain that there is not a small fragment of a Y chromosome included with a fragment of an X chromosome. The presence of a Y fragment has no effect on femininity, or the physical appearance of a girl with Turner Syndrome, or her behaviour, but it is important for the development of her ovaries. When a Y chromosome fragment is present, the ovaries should be removed soon after the diagnosis has been made as they may undergo a cancerous change. It is often difficult for parents of a girl with Turner Syndrome, or indeed an adult woman with Turner Syndrome, to understand why her ovaries have to be removed when they are non-functioning. This needs considerable and careful explanation and discussion and, possibly, counselling.

Turner Syndrome occurs in approximately 1 in 2,500 women. Although relatively uncommon in the general population, it is a common disorder to an endocrine specialist and your specialist should be one who has considerable experience in dealing with the problems that women with Turner Syndrome have.

Natural History of the Ovary in Turner Syndrome

Early in the pregnancy a baby with Turner Syndrome has the normal number of eggs in her ovaries. However, from three months of foetal life, and because of the lack of the missing X chromosome, the eggs have difficulty dividing, die off, and are not replaced. Thus, at birth there are considerably fewer eggs in a female baby with Turner Syndrome than would normally occur in the ovaries of girls and this rapid decline continues throughout childhood. Therefore, in most girls with Turner Syndrome, there is an insufficient number of eggs (and hence ovarian activity) at the age of eleven to produce

enough oestrogen to trigger the start of puberty, When looking at the ovaries using ultrasound, they will either appear as "streaks" of tissue or as normal sized ovaries which contain very few eggs. This process is similar to that which occurs in older women at the menopause except that in girls with Turner Syndrome it happens before puberty. Most women with Turner Syndrome are not able to produce the female hormones (oestrogens and progestogens) which are needed to stimulate the development of the sexual characteristics such as breast development nor are they able to produce eggs and so they are not fertile. However, the other internal reproductive organs such as the fallopian tubes, the uterus and vagina, are entirely normal.

Puberty

As discussed in the previous section, most girls with Turner Syndrome do not enter puberty. They need to be treated with oestrogen which induces all the secondary characteristics of puberty, including breast development, maintenance of pubic hair, change in body shape, moistness of the vagina in order to allow comfortable sexual intercourse, and the associated psychological changes of puberty.

Treatment:
In order to mimic the normal changes that happen in girls going into puberty, low dose oestrogen therapy is started at around the age of eleven years. The timing of starting oestrogen may be earlier than II years and you will need to discuss this with your specialist.

A starting dose would be in the order of 2 micrograms ethinyloestradiol (a synthetic oestrogen) per day. The dose is gradually increased during pubertal development. For the first year or two only oestrogen treatment is given (that is, oestrogen not combined with another hormone, progestogen). After the dose of oestrogen has been built up to a level sufficient for when the first period should occur, or the first period has occurred (this may be seen as a spotting of blood), progestogen is added each month to the treatment.

This combined treatment pattern may be continued until it is appropriate for a higher dose of oestrogen to be given. At this point the combination of oestrogen and progestogen can be given in the form of a combined pill. Initially this may be a low dose "contraceptive pill" containing progestogen and 20 micrograms of oestrogen. In the later teenage years this may be increased to a "contraceptive pill" containing 30 to 35 micrograms of oestrogen. The tablet (pill) is usually taken for three weeks. There is then a week off from treatment so that there is a "period" (a uterine withdrawal bleed). It is important to realise that there are a number of variations in how the tablets (pills) are presented but they produce the same effect. Your specialist will discuss with you what is most suitable for you.

The withdrawal bleed is not a period in the true sense as there has been no egg released from the ovary. However, it is important to have this withdrawal bleed in order to shed the lining of the uterus and so keep the uterus healthy.

About 4% of girls with Turner Syndrome have enough ovarian function to enter puberty without any oestrogen treatment. However, only about 1% complete normal development on their own and have spontaneous menstrual cycles. These initial menstrual cycles are usually not egg producing (anovulatory) which is anyway quite normal for the first eighteen months or so following the first period in any female. Only 0.5% of Turner Syndrome girls have egg producing (ovulatory) cycles. However, if spontaneous puberty and menstrual cycles do occur in women with Turner Syndrome, it is likely that they will have their menopause early.

Maintenance of Sexual Development

Unless you are one of the few women with Turner Syndrome whose ovaries continue to work in adult life, you will need to take oestrogen therapy in order to maintain your general well being. This treatment is not just to maintain breast development, but is important for psychological reasons, for the vaginal secretions that enable sexual intercourse to be comfortable and medically maybe most important of all, to help prevent coronary artery disease and to maintain the strength of your bones (see below). It is mainly in order to supervise oestrogen therapy that we advise you to stay with an adult specialist with a knowledge of Turner Syndrome. It is important that you remain on an adequate dose of oestrogen in order to maintain the strength of your bones and the most critical time for this is in late adolescence. With the increasing independence of girls in their late teenage years, medical care is often thought to be of no further importance and may even be seen as an inconvenience. It is important that this does not happen as a woman may then find that she is no longer under the specialist care that she needs and does not know where to seek it.

Oestrogen medication is usually given as tablets in the early teenage years to trigger the start of puberty. However, unopposed oestrogen (oestrogen on its own without a progestogen) is an unsafe treatment once periods have started. Treatment with oestrogen alone will stimulate the build up of the lining of the uterus and this lining will not be shed in a normal way without the introduction of progestogen. This monthly bleed is stimulated by the progestogen treatment and is necessary to keep the uterus healthy. Without progestogen, after a time, cancerous changes of the uterus may develop. It must be remembered that this monthly bleeding is dependent on oestrogen and progestogen therapy and is not associated with being fertile as the ovaries are not producing eggs.

Sometimes there are advantages in giving oestrogen in adult women by a route that is not via the mouth. If oestrogen is given either through a patch on the skin or by a small implant (insertion of a small pellet just under the skin from which the oestrogen is released slowly) it may have less effect on blood pressure and perhaps blood clotting. Non oral oestrogens/progestogens are therefore worth considering in women who have high blood pressure during oral oestrogen therapy. Your specialist will discuss what is most appropriate for you.

In older post menopausal women who have hormone replacement therapy (HRT– which is oestrogen and progestogen treatment) one of the common reasons for not continuing treatment is because of the inconvenience of periods and because of the minor side effects during the progestogen treatment phase which are similar to premenstrual syndrome. This could also apply for women with Turner Syndrome. One way to help with this is to have continuous combined low dose oestrogen and low dose progestogen therapy which allows enough oestrogen to be given to protect the bones without causing a build-up of the lining of the uterus. There are treatments available which have a combined oestrogen and progestogen content and these can protect the bones without the need to have periods. This treatment may be regarded by some doctors as experimental and so the decision about which type of treatment would best suit you will need to be discussed with your specialist.

Osteoporosis (Thin/Brittle Bones)

Osteoporosis is characterised by a reduction in the amount of bone thickness and an increase in bone fractures (eg through only minor injury). It is a major cause of pain, disability and suffering in elderly women and in view of the increased risk of major fractures, may be the cause of death in elderly women. It is now realised that the main bone mass, and thereby strength of the bones, is made during adolescence through the action of the sex hormones (oestrogens and progestogens). Thus, any condition in which the ovaries do not produce enough oestrogens, as occurs in Turner Syndrome, causes a woman to be more likely to develop osteoporosis. In addition. in Turner Syndrome the bones are probably more likely to develop osteoporosis as part of the underlying condition. Indeed, it is not unusual to be able to diagnose "pre-pubertal osteoporosis" on a hand x-ray during childhood. For these reasons it is particularly important that both girls and adult women with Turner Syndrome continue with oestrogen/progestogen treatment throughout life until the expected age of the menopause and then to continue this as HRT, probably into their sixties.

As women with Turner Syndrome are at risk of developing osteoporosis, they should have the appropriate screening by measurement of bone density in the late teenage years or early twenties in order to have an accurate baseline measurement for serial measurements in later life. Although compliance with HRT has proven disappointing in post-menopausal women, it is very important that a woman with Turner Syndrome continue oestrogen/progestogen therapy until her specialist advises that it is unnecessary. The common anxieties of women about HRT therapy include a fear of gaining weight, developing cancer, developing thrombosis and of cardiovascular disease. Certainly the fears of cancer and cardiovascular disease are unwarranted and indeed there is a considerable amount of evidence that there is a dramatic reduction in mortality from heart disease with HRT therapy.

Osteoporosis is a difficult disease to treat once it is established, and often the best that

treatment can achieve is to slop any further deterioration of the bones. The treatments that are available are many and varied but among the most important are regular exercise, calcium supplements and possibly vitamin D supplements. It is extremely important to replace any deficiencies in oestrogen hormones. Other treatments which are available include bisphosphonates and calcitonin although such treatments are more specialised. The use of fluoride treatment (like fluoride in toothpaste) is controversial. If you have had treatment to improve your growth during childhood and adolescence, using an anabolic steroid (such as oxandrolone) or growth hormone, then this was probably beneficial to your bones, as well as improving your overall growth. Both of these treatments can increase bone density.

Screening for osteoporosis and monitoring the condition of your bones, are best done using bone densitometry. Computerised tomography is sometimes used as well as specialised ultrasound techniques. Your specialist will discuss with you what assessments you should have as well as where and when such tests should be done. Certainly, measuring bone density at intervals to monitor the strength of your bones is a useful investigation, although there is not yet enough information available to make definite recommendations.

"I find the possibility of developing osteoporosis extremely threatening. I have always been fairly supple and active and the thought of being restricted in movement seems a harsh possibility to face. I certainly feel that the monitoring of bone density should be a standard procedure for Turner Syndrome women. The tests I have undergone have been purely at my own instigation. Despite my concerns, it seems difficult to prise out specific answers to my queries. Just how much at risk am I? Sometimes I feel I am battering myself against an extremely polite brick wall!"

Ankle Swelling

In Turner Syndrome there is often a problem with the development of the lymphatic vessels which drain away fluids and are rather like blood vessels in the skin, This can result in swelling of the hands and feet which is commonly seen in newborn babies with Turner Syndrome. The swelling usually disappears within the first year of life, but may continue or return in adolescence or adult life. Once oestrogen replacement therapy has started, this can increase the retention of fluid in the tissues. The use of a treatment which makes the kidney pass more water is often helpful (and such drugs may have the added advantage of also protecting against osteoporosis). If simple remedies, such as raising the end of the bed are unhelpful, you will need to discuss treatment options with your specialist.

Blood Pressure

Women with Turner Syndrome are at risk of developing high blood pressure (hypertension). Your blood pressure should be measured regularly, particularly when you are on oestrogen therapy. There are several reasons why blood pressure in Turner Syndrome may rise including the possibility of a constriction in the large blood vessel leaving the heart (coarctation of the aorta), as well as problems with the kidneys or in the

blood vessels within the kidney. However it is so important that a woman with Turner Syndrome receives adequate oestrogens for all the reasons we have mentioned above that it may be necessary to additionally treat the hypertension while continuing oestrogen medication. As discussed in the section on "Maintenance of sexual development", it may be appropriate to use non oral oestrogens.

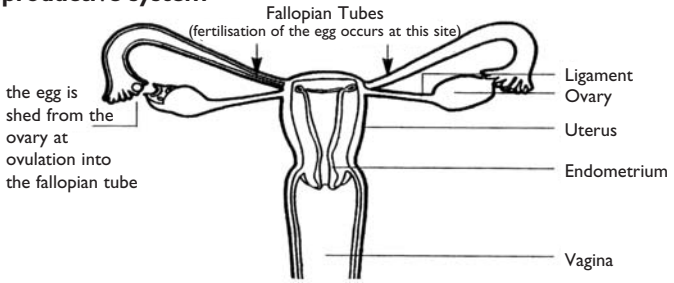
Because of the coarctation (narrowing) of the large blood vessel leaving the heart, which can occur in women with Turner Syndrome, it is important to measure blood pressure in the right arm, as measurement of blood pressure in the left arm may be misleading. The problem of high blood pressure will probably require further investigations and your specialist will discuss this with you.

Fertility

Very few women with Turner Syndrome are able to have children without medical help despite having regular uterine bleeds from cyclical oestrogen/progestogen therapy which is needed to help keep the uterus healthy. This often causes the most severe anxieties associated with the diagnosis of Turner Syndrome as is well illustrated in the "Patient's Story" at the end of this booklet Infertility is difficult for anyone to accept. Women with Turner Syndrome have a normal uterus which responds normally to hormone treatment. It is therefore possible to have a child through egg donation and fertility treatment. An egg can be donated from another woman and fertilised with your partner's sperm. This can take place using the technique of in-vitro fertilisation (IVF) whereby the egg and sperm are put together in a test tube and fertilisation takes about twelve to fifteen hours and is confirmed under a microscope. The fertilised eggs are referred to as pre-embryos and an agreed number (usually 2 to 3) will be replaced into the uterus about two days after egg collection. The pre-embryo(s) are transferred using a fine tube through the cervix into the uterus. During this time, hormonal treatment is required in order to make the lining of the uterus able to receive the embryo.

Another method of assisted conception is Gamete Intra-Fallopian Transfer (GIFT). In this technique the egg and specially prepared sperm sample are transferred into one of the fallopian tubes and this allows fertilisation to take place naturally within the fallopian tube.

Female reproductive system



Successful IVF has been reported in women with Turner Syndrome. Once the embryo has been implanted into the uterus, the placenta will produce oestrogen hormones and so, for once, additional hormone therapy will not be required during the pregnancy. Such techniques as IVF are difficult and time consuming. The waiting lists for such treatment are long and it is therefore important that you discuss your plans for assisted fertility with your specialist as early as possible. In some parts of the United Kingdom IVF treatments are not available under the NHS and there is a great shortage of egg donors. Both these factors seriously restrict the number of patients who can benefit from assisted fertility treatment.

A few women with Turner Syndrome (less than 0.5%) have egg-producing menstrual cycles and are potentially fertile. However, it should be remembered that if a pregnancy is decided upon, the chances of having a baby with either Turner Syndrome or a major heart defect are extremely high. It is probably preferable to receive a donor egg and have in-vitro fertilisation.

Social Concerns and Psychological Support

Some women find coping with the implications of Turner Syndrome harder than others. This may depend on their particular situation, or the attitude shown to them when they are growing up. Parents may have been over protective and not allowed their daughter the experience of achievement, and the ability to "cope" is therefore often not well developed. Sometimes girls or young women who are relatively small for their age and who look immature are treated according to their size and not their age. This can cause feelings of loss of achievement and self-esteem which may result in a sense of being unable to succeed at anything.

Many of the problems which are associated with Turner Syndrome are seen first in childhood but continue into adulthood. They include difficulties with planning skills, problem solving and a certain lack of flexibility of thinking about day to day concerns. There is often a need for constant reassurance which can be frustrating for the woman herself as well as being difficult for those around her. To compensate for these problems, reasons are often found for not attempting challenging tasks. Daily life can become a routine that cannot be altered. If it is disrupted, even by a trivial event, panic and confusion are created which can often result in a show of uncontrolled temper.

There are also the emotional difficulties of coming to terms with being small as well as the lack of functioning ovaries, both implications of Turner Syndrome. Feeling "different" because of these reasons and because of the often rigid way of thinking means a woman may require professional help and advice. Comprehensive psychological care may often be necessary for those adult women with Turner Syndrome who have a markedly impaired sense of self esteem, as well as an inability to cope with daily events. Physicians who have the care of Turner Women need to be sensitive in exploring the very real problems some of these women have, which include being very dissatisfied with themselves and their lives, and should refer to an appropriate specialist if needed.

A Patient's Story

This is the story of a woman in her thirties who has Turner Syndrome. It illustrates many of the problems involved in coming to terms with the condition, especially those related to late diagnosis, as well as the important place of the specialist who co-ordinates her care.

"I was diagnosed at fifteen as having Turner Syndrome, and had little idea of what that meant. My father took me every so often to a London hospital. I had tests, received medication and went home, where it was never discussed. My consultant must have explained the implications of Turners Syndrome to me, yet I still believed, because I bled every month, I would be able to have children, perhaps we only hear what we can cope with. At eighteen, when I finally understood I was infertile, I wept, and the world was suddenly full of pregnant women.

I went through a period of denial so intense that I convinced my GP to refer me to a gynaecologist who performed a laparoscopy. He was thrilled to find I had a uterus – even I could have told him that! My endocrinologist, unaware of all this, was not best pleased when he found out I had taken my custom elsewhere! I was lucky enough to have the same endocrinologist for nearly fifteen years. Medication changed, blood tests came and went, but the benefit was in the mutual trust we established. I felt he knew me as a person, not just a patient. I felt I mattered. When eventually he left to take up another post I mourned and went through a real period of bereavement. It was as if he had died. I saw another consultant, but there was little rapport (no doubt I made little effort) and I stopped attending hospital as an outpatient. After nearly six years, my GP still hasn't quite realised – but that is another story!

There have been comic interludes – the GP trainee playing "spot the syndrome" who announced my diagnosis as "cretin" (obviously not one of my better days!) I told her gently perhaps she ought to re-think in view of the numerous diplomas, Bachelor of Arts and possible imminent MA!

Coming to terms with TS began with my involvement with The Child Growth Foundation. Until then I had never met anyone else with Turners. It gave me access to wider information and a "sounding board". Suddenly I had experience to offer others and I no longer felt isolated. In all my years of hospital treatment I had never been offered any counselling or support. Coming from a naturally reticent family, we had never discussed the implication of TS. It was an unspoken taboo. Involvement with the CGF came as a profound relief, and with it a great leap in confidence. Any wound will not heal without cleansing, and the CGF provided the means for me to come to terms with Turners though contact with others.

One could see TS as a "Cinderella" syndrome – not rare enough to cause major ripples in the medical pool, not common enough for every GP to have a thorough understanding of what it entails. We are somewhat "betwixt and between" . If I am ever tempted by self pity I remind myself of those in far worse situations. Others also confront infertility or health problems. They need not always be insurmountable.

Ultimately, I feel good communication is essential. Consultants and GPs can only hope to offer effective help if we are honest about our concerns and needs as patients. I want to take responsibility for my health, not be passively "treated". I wish to participate in decisions about my well-being. I am also sure that all consultants realise that a patient with a little knowledge is not always a dangerous thing!

It can hurt to be "different", not to feel "normal" – not be able to always "blend in with the crowd". One can either retire from the fight, and withdraw from life, or stand firm against the "slings and arrows of outrageous fortune", to do the latter takes courage. Usually I find it."

Questions & Answers

1. **Q** Do I need to see a doctor as an adult woman with Turner Syndrome?

   A Yes, particularly to make certain that you have an adequate oestrogen replacement, without complications, and that you are screened for the development of osteoporosis.

2. **Q** Can I become taller as an adult?

   A This is very unlikely if you are older than your teenage years, however your specialist will be able to advise you about this. The specialised treatments now available to improve the growth of children with Turner Syndrome were not available prior to the mid 1980s. Hopefully, some of the problems associated with short stature in Turner Syndrome will be prevented in the future.

3. **Q** If I am having oestrogen replacement treatment, why must I have periods?

   A Taking oestrogen alone without a progestogen (unopposed oestrogen) results in a build-up of the lining of the uterus which can, after a considerable time, increase the risk of cancer of the uterus. It is therefore important to have regular withdrawal bleeds in order to keep the uterus healthy.

4. **Q** I have been told that I am infertile so why am I taking a "contraceptive pill"?

   A This is a situation which many women with Turner Syndrome find particularly difficult to come to terms with. The combined hormones oestrogen and progesterone are included in each daily pill used in hormone replacement therapy, which is needed by women with Turner Syndrome, The same combination of hormones is required for the contraceptive pill. The practical benefits of using the contraceptive pills are that they are produced in a package that is easy to use and that they are free from prescription charges.

5. **Q** What is HRT?

   A The initials stand for Hormone Replacement Therapy and this treatment involves the administration of oestrogen and progestogen in a way that mimics the body's normal secretion. A sequence of pills containing differing levels of oestrogen and progestogen are taken during the month which continues the beneficial effects of these hormones once their normal levels decrease after the ovaries have stopped working (the menopause).

6. **Q** Which method of HRT would suit me'?

   A This should be tailored to the individual and you will need to discuss this with your specialist.

7. **Q** Are there any other advantages to HRT other than the prevention of osteoporosis?

   A Yes, as well as the maintenance of female physical characteristics of puberty and moistness of the vagina, there is also a general improvement in well-being that is difficult to quantify. In addition, there is a significant reduction in the incidence of heart disease and probably of strokes as well.

8. **Q** Why do so many women stop HRT after only a short period of treatment?

   A This is usually due to the inconvenience of having withdrawal bleeds after the menopause and also to the occasional "pre-menstrual symptoms" which are related to the progestogen component of treatment.

9. **Q** I have diabetes mellitus (sugar diabetes). Will HRT interfere with this?

   A It may mean that your insulin treatment will need to be increased a little but it will make very little difference overall. Many women with Turner Syndrome have insulin intolerance which, in the majority of cases, is controlled by diet and HRT should not affect this.

10. **Q** Is oestrogen therapy related to breast cancer?

    A This is an extremely controversial subject and there have been some concerns in the past. However, if there is a risk, it is thought to be very small. If you remain concerned about this, ask your specialist for further information.

11. **Q** Do I need to be monitored tot the development of osteoporosis?

    A Yes. Having Turner Syndrome means that you will be more likely to develop

osteoporosis. The exact method of monitoring/assessment in order to assess your bone density, as well as how often this will be needed, will be discussed with you by your specialist.

12. **Q** If I have IVF with a donor egg, will my baby be normal?

A There is approximately a 1% chance in any pregnancy that the baby may have an abnormality. From the evidence available, it seems that the risk is no greater when having IVF or GIFT.

13. **Q** As an adolescent I was treated with growth hormone and an anabolic steroid – is there any long-term advantage in this?

A Yes, both treatments, as well as improving growth and final height, are important in the treatment of osteoporosis. We know that adolescence is the most important time for building up the strength of the bones and it may well be that these two treatments will have an important effect in protecting against osteoporosis in later life.

14. **Q** How do I find a specialist who deals with Turner Syndrome in adults?

A If you are not already under the care of a specialist you should discuss this with your GP, The Turner Society or the specialist you were under for your paediatric care if appropriate, who will advise you.

15. **Q** Do I need to be monitored for any hormone deficiencies?

A Yes, thyroid hormone deficiency (hypothyroidism) is common in all adult women but especially those who have Turner Syndrome. This will require a blood test at intervals that your specialist will discuss with you.

Summary

It is important to remember that Turner Syndrome is not a disease but a condition which is compatible with a normal life. In this booklet we have concentrated on the main problems in adult life, which relate to the ovaries not working. These include the requirement for oestrogen replacement, the prevention of osteoporosis and having children through in vitro fertilisation with a donor egg. However, there are many aspects of the management of women with Turner Syndrome which need consideration and your specialist will be able to both help and guide you. With this support, most Turner Syndrome women should be able to have a fulfilled life in all aspects including those physical, emotional and sexual.

Further Information

The Turner Society
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London W4 IPW
Tel. 020 8994 7625
020 8995 0257

The National Osteoporosis Society
P0 Box 10
Bradstock
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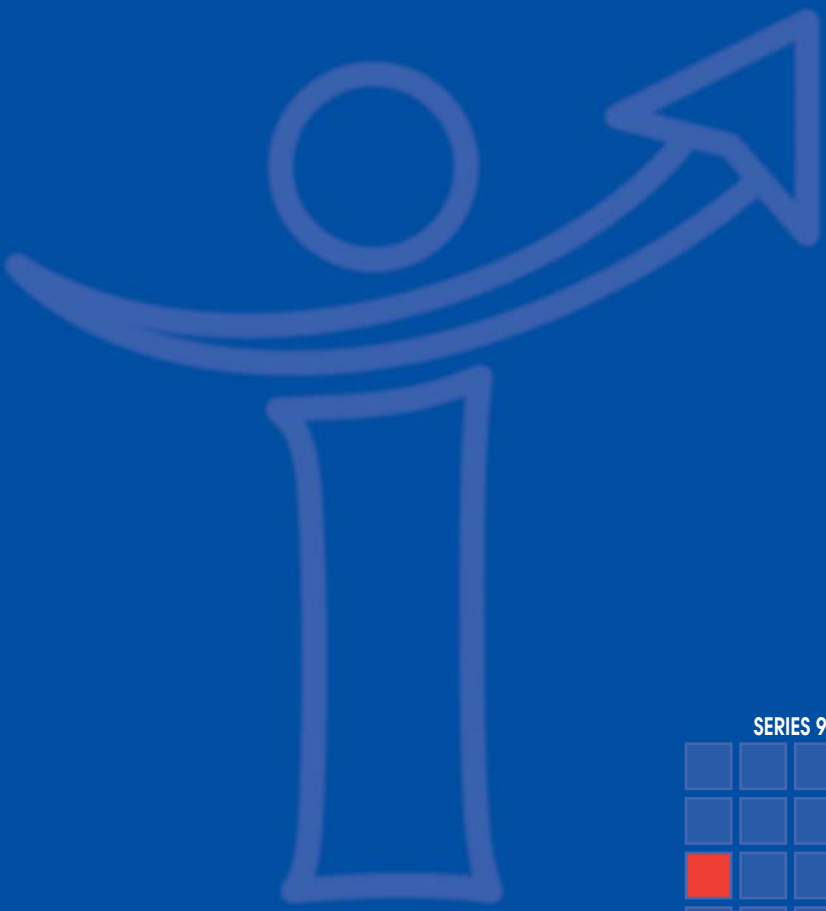
Tel. 01761 432472

Hormone Replacement Therapy: Your Questions answered. By M Whitehead & V Godfree. Published by Churchill Livingstone, 1992.

Societies for support of the infertile:

CHILD
(The National Infertility Patients Support Group)
Charter House
43 St Leonards Road
Bexhill-on-Sea
TN40 1JA
Tel. 01424 732 361

ISSUE
(The National Fertility Association)
114 Lichfield Street
Walsall
WSI ISZ
Tel: 01922 722666



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