

Turner Syndrome

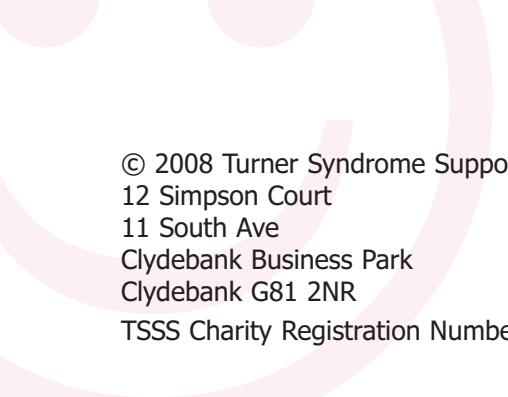
lifelong guidance & support



2nd
edition



Turner Syndrome Support Society



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Acknowledgments

Many people have worked long and hard on this book. The Turner Syndrome Support Society would like to thank everyone whose suggestions, support and encouragement have enabled this book to be published.

In particular, our sincere thanks and gratitude go to Arlene Smyth, Ruth Kimbell, Lynne Morris, Helen Lawn, Jane Smalls, Christine Charlton, and John Short, whose contributions to this book have been invaluable.

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Dr Lih Mei Liao, Psychologist, Middlesex Hospital, London



Foreword

1st Edition Foreword

The Turner Syndrome Support Society can be justifiably proud of this publication. It provides detailed and appropriately balanced advice for individuals who have been diagnosed with Turner syndrome and for their parents, not only in the form of detailed information, but also from a practical standpoint.

Advances in medical diagnostic techniques mean that the syndrome can now be diagnosed with more certainty and at a very early stage. This allows decisions to be made from the very early days on how best to plan treatment in an individual case – each of which will have its own specific and varying needs.

The agreement by the National Institute of Clinical Excellence (NICE) for England and Wales to approve the availability of growth hormone treatment in Turner syndrome has been, for the most part, the result of the Society's hard work in education and lobbying over many years. This treatment will help address one of the major concerns which arise as girls with Turner syndrome approach puberty – that is how to maximise the growth spurt and endeavour to achieve as optimal a height as possible.

However, it is the knowledge and understanding of the wide variety of other associated problems experienced by individuals who have Turner syndrome, which this book contains, that provides excellent guidance and supportive advice.

Few general practitioners, and even the majority of general paediatricians and gynaecologists, unless they have a particular interest in the condition, will be aware of all of the potential special needs of girls and women with Turner syndrome. The guidance in this book provides a wealth of information and is recommended as essential reading for any professional who provides care.

However, it is for individuals with Turner syndrome, and their parents, that the Society primarily produced this book. It sets out to explain how recent advances in medical treatments and understanding of Turner syndrome can now best be used to improve quality of life and, as such, I know it will be immensely informative. We hope you will find the book of value and would welcome comments on how to improve future editions.

Robert Shaw CBE

Professor of Obstetrics and Gynaecology
Patron, Turner Syndrome Support Society

2nd Edition Foreword

This is a super book crammed full of knowledge and understanding. It is a credit to the Turner Syndrome Support Society and encapsulates its raison d'être, namely to bring that knowledge and understanding to others so that life is improved for girls and women with TS.

It can be read with advantage by the parents of a child newly diagnosed with TS and equally by the professionals who encounter a girl or woman with TS. Most questions about TS can be answered in this book or a lead may be given as to where the answer lies.

The accounts of personal experiences bring real understanding. As Arlene puts it in her introduction, the real experts in TS are the girls and women with TS themselves and their parents and friends. It is as instructive and helpful to the professional to read those accounts as it is enlightening to those living with TS to be aware of the facts around this condition that has so many facets and implications.

This second edition reflects the fact that each year brings more information and understanding of Turner Syndrome. Let there be many more editions in the future.

Tony Price

Patron, Turner Syndrome Support Society,
Consultant Paediatric Endocrinologist (retired)

Preface

1st Edition Preface

I am delighted as Chairman of the TSSS to add my own special thanks to all who have contributed to this comprehensive overview. This book has been needed for so long and I believe that in it we have a superb review available to experts and beginners alike. As a father of a daughter with TS, whose diagnosis was confirmed over 23 years ago, I can only marvel at the progress that has been made to our understanding of the life problems that have been so clearly presented in this volume.

Keith Masters MB FRCOG

Chairman, Turner Syndrome Support Society

2nd Edition Preface

Affectionately known as the 'TS bible' this publication gives an informative and unbiased view on Turner Syndrome and related issues. The education and greater awareness of TS is central to addressing the challenges of the condition and this book successfully achieves that. It has been deliberately written and edited in a manner that transcends boundaries – it is equally useful for girls and women with TS, their parents, friends and relatives – as well as medical specialists and professionals.

As both Chairman of the Turner Syndrome Support Society and a father of a teenage daughter with Turner syndrome I am delighted to offer my sincere thanks and heartfelt appreciation to everyone who has contributed to the production of this second edition of Turner Syndrome, Lifelong Guidance and Support.


Malcolm Lee

Chairman, Turner Syndrome Support Society

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This book is dedicated to the girls and women
with Turner syndrome whose spirit and
determination are an inspiration to us all.

Introduction

1st Edition Introduction

This book covers all aspects of Turner syndrome and top UK specialists have endorsed the medical information it contains. Most importantly, the book gathers together the experiences of girls and women with TS, and those of their families and friends. They live with the condition on a daily basis and are the real experts on TS.

When my daughter was born and diagnosed with TS 15 years ago, I was frustrated by the lack of information available to me as a parent. When I was at last able to meet other families who had a child with TS, I learned that girls and women with TS face many medical and practical problems. I also learned that, in discussing the variety of ways in which they had overcome these problems, the families recognised many similarities in their daughters. This was when I discovered that the most valuable help and support comes from talking with others and sharing their experiences.

I feel privileged that over the years so many people have confided to me their deepest concerns. By listening to their personal stories I learned that it is possible to come to terms with TS, to understand it and to learn to cope. There is something special about every girl and woman with TS.

I am glad to have the opportunity, through this book, to realise my dream and share the knowledge that I have gained from everyone I have spoken to from all over the world. The publication of this book means that no-one faced with a diagnosis of TS, either for themselves or for a daughter, need feel the frustration and isolation that I did when my daughter was diagnosed. My hope is that this book will help girls and women with TS, as well as their families and those who care for them, to understand the condition better so they can lead happy, healthy lives.

Arlene Smyth

Founding member, Turner Syndrome Support Society

2nd Edition Introduction

I am immensely proud of the success of this wonderful book. Our first print run of five thousand books has been distributed to many families throughout the UK and around the world. We are delighted to have secured further funding for a second print run of another five thousand books. We were once again supported in part by Ferring Pharmaceuticals, phenomenal fundraising by members of the TSSS and the generous support of Harlow Printing Limited. We are very grateful to them all for their support. This book continues to be a valuable resource for any one who has TS or is involved in the care of someone who has TS. There have only been some minor changes made to this book updated introduction, preface & foreword and some photos.

Personally I feel a great sense of pride at the information the TSSS has produced in the last nine years. The society has a marvellous group of volunteers, trustees and committee members who work hard to keep all our information relevant and up to date. Together we make a great team continually working to improve understanding of Turner Syndrome throughout the world.

Having recently celebrated the twentieth anniversary of the Scottish group it is clear our knowledge and understanding of TS has come a long way. When I look now at my beautiful daughter who has grown into a wonderful young woman, I think what did I worry about she is doing great. I realise how lucky I am because, by having the knowledge in this book and others the TSSS produce. It gives you the power to make positive changes to help and support girls and women who have TS, allowing them to reach their full potential and blossom into the very special people they are.

As we approach our tenth year the TSSS have achieved many goals and realised a few dreams along the way. I am so proud to be part of the TSSS family and look forward to working towards even greater understanding in the next ten years.

Turner syndrome – making sense of the diagnosis

What is Turner syndrome?

Turner syndrome (TS) is a relatively common chromosomal disorder affecting only females, caused by complete or partial deletion of the X chromosome in some or all cells of the body. TS affects approximately 1 in 2000 live female births [1,2]. In Britain, it is estimated that there are about 10,000 girls and women with TS [3] and worldwide figures are estimated to be 1.5 million [4].

How is Turner syndrome diagnosed?

TS is sometimes suspected when an ultrasound is performed during pregnancy (one of the signs is fluid around the neck). Sometimes TS is diagnosed incidentally during pregnancy when chorionic villus sampling (CVS) or amniocentesis are performed for some other reason, the most common being advanced maternal age - although this in itself is not associated with increased incidence of TS [1,5]. Table 1.1 gives details of these tests. When an ultrasound scan suggests TS, the diagnosis can be confirmed by CVS or amniocentesis for chromosome analysis to check the chromosome pattern, known as karyotype testing (Table 1.2). The chromosome patterns in TS are discussed in detail later in this chapter.



Figure 1.1: This little girl, Iona, has many characteristic features of TS, for example, a short, thick neck, low set ears and epicanthic folds at the inner corners of the eyes.



It is more usual for diagnosis of TS to be made later however, after the baby is born. TS may be suspected shortly after birth because the baby girl has certain features, for example a short wide neck, epicanthic folds at the inner corner of the eyes (Figure 1.1), puffy hands or feet (oedema) (Figure 1.2), widely spaced nipples (Figure 1.3), feeding difficulties, or occasionally a heart problem. Sometimes the problem with the neck is very severe (webbed neck). Table 1.3 sets out the characteristic features that suggest a diagnosis of TS in a newborn baby.

Table 1.1: Pre-natal tests and how they may reveal a diagnosis of Turner syndrome

Ultrasound

A method of imaging the body based on the reflectivity of sound which uses very high frequency (ultrasonic) sound wavelengths. In obstetrics, ultrasound can show the position of the placenta, reveal twins at an early stage, show the size and sex of the foetus and detect some (but not all) foetal abnormalities. One such abnormality is the translucency of tissue around the baby's neck (nuchal translucency or NT, sometimes called cystic hygroma) that is characteristic of Turner syndrome. Other findings on ultrasound characteristic of Turner syndrome are raised foetal heart rate (tachycardia), and restricted intrauterine growth early on in pregnancy.

Chorionic villus sampling (CVS)

This test involves removing a small amount of placental tissue using a fine tube or needle. It is usually performed at about 10-12 weeks of pregnancy. The placental tissue contains the same chromosomes (karyotype) as the developing embryo and genetic analysis will reveal the presence of chromosomal abnormalities of the type seen in Turner syndrome.

Amniocentesis

In this test, a sample is taken of the amniotic fluid in the womb using a fine needle. This fluid contains cells from the foetus which can be analysed to determine the sex of the baby and its chromosome karyotype. Amniocentesis is often performed to look for Down's syndrome (another chromosomal abnormality). Occasionally Turner syndrome is detected unexpectedly when an amniocentesis (or CVS) is performed for some other reason.



Figure 1.2: Often, the first sign that raises the suspicion of TS in a newborn baby girl is swelling (oedema) of the feet and hands.



TS is sometimes not diagnosed until the early childhood years. This may happen at a routine check-up, or because parents are worried about their child's growth or development. The worry may be that a girl is not growing as well as expected, for example, or she may be experiencing persistent infections of the middle ear, both of which might lead to a suspicion of TS. It may also be found that she has other features characteristic of TS, for example an increased carrying angle of the elbows (cubitus valgus), or spoon-shaped or hyperconvex nails.



Figure 1.3: A characteristic feature of TS is a broad chest with widely spaced nipples.

Table 1.2: Karyotype testing

This can be done either during pregnancy (by amniocentesis or CVS) or at any time after birth. It involves obtaining a sample of cells, either from a blood sample or during pregnancy by amniocentesis or CVS, and analysing the chromosomes in the cells. The chromosomes are studied under a microscope by a cytogeneticist specially trained to detect abnormalities in the pattern of chromosomes by the specific banding pattern after staining. This allows him/her to see if any chromosomes are missing or if small parts of the chromosome are abnormal. After birth, a karyotype is usually obtained from a sample of blood. Occasionally, if there is strong clinical suspicion of Turner syndrome, despite a normal blood karyotype, a second tissue sample is tested. This can be done using a scraping of cells from the inside of the cheek (a buccal smear) or a tiny sample of skin cells (a skin biopsy).

Some girls with TS are not diagnosed until they are teenagers. These girls are usually taken to a doctor because they are not showing any signs of puberty at the expected time: their breasts do not develop and their periods do not start (see Table 1.3). Teenage girls with TS may be shorter than their peers or have other features of TS which had, up until that time, not been noticed.

Although girls with TS may have some of the characteristic features set out in Table 1.3, rarely do they have all of them. However, short stature and infertility are nearly always present. In all cases, if TS is suspected at any stage because some or all of the characteristic features are present, the diagnosis is confirmed by chromosome analysis (karyotype testing) from a blood sample. A karyotype test can be done at any age.



Table 1.3: Characteristic features leading to a suspected diagnosis of Turner syndrome (not all the features listed may occur)

Possible features in newborn babies

- Swelling (oedema) of the hands and feet
- Folds in the back of the neck (webbed neck)
- Low birthweight
- Small or absent nails
- Low hairline
- Low set ears
- Small jaw (micrognathia)
- Widely spaced nipples
- Anomalies in the left side of the heart, especially narrowing or coarctation of the aorta or an enlarged left ventricle (left ventricular hyperplasia)
- Feeding difficulties

Possible features in childhood

- Any of the signs seen in the newborn listed above although if the features are not present at birth the child will not develop them later
- Short stature (with a declining rate of growth)
- Increased carrying angle of the elbows (cubitus valgus)
- Spoon-shaped or small hyperconvex uplifted nails
- Multiple pigmented naevi (moles)
- Characteristic facial appearance (drooping eyelids)
- High arched palate
- Short fourth toe and short fingers
- Chronic problems with middle ear infections (otitis media)
- Short sightedness (myopia) or other eye problems
- Raised levels of the hormone follicle stimulating hormone (FSH)
- High blood pressure
- Kidney and urinary tract problems
- Thyroid problems
- Behavioural problems, hyperactivity, immature behaviour
- Specific learning difficulties and problems with spatial awareness
- Sleeping problems

Possible features in adolescence

- Unexplained short stature
- Breast development fails to occur
- Menstrual periods fail to occur
- Thyroid problems
- High blood pressure
- Hearing problems

Having TS affects many parts of the body and can have a profound effect on quality of life. Each girl with TS should receive treatment individualised to her specific needs, preferably at a specialist TS clinic, or from a doctor who has a special interest in TS.

The genetics of Turner syndrome

A person's genetic make up is inherited from both parents in the form of chromosomes. Chromosomes contain genes that determine how we grow



and develop. Normally, each cell in the body contains 46 chromosomes. These are arranged in 23 pairs. One half of each pair is inherited from each parent. One of these pairs of chromosomes, the sex chromosomes, determines the sex of a baby. A boy is born with an X and a Y chromosome (represented as a 46,XY karyotype) and a girl has two X chromosomes (a 46,XX karyotype).

Occasionally, however, a girl is born with only one X chromosome instead of the usual two (represented as 45,X or previously as 45,X0, where 0 indicates the missing X chromosome) (Figure 1.4).

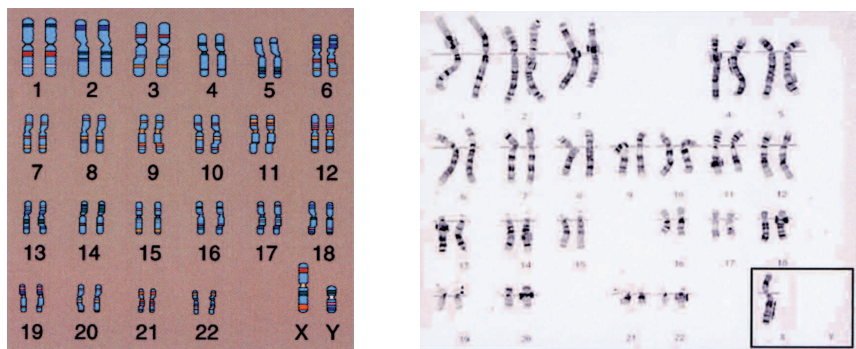


Figure 1.4: In Turner syndrome (right) there is usually only one X chromosome instead of two, making 45 chromosomes in total rather than 46 (left).

This is known as Turner syndrome after the American endocrinologist Dr Henry Turner who first described the condition in 1938. The missing X chromosome, from either the mother's egg or the father's sperm, is lost at some time during cell division, in the course of egg or sperm production, or early on in the development of the embryo. It is estimated that fewer than 1% of female embryos that are missing an X chromosome survive to the end of pregnancy [6].

The 45,X karyotype is known as *classic TS* (or *non-mosaic TS*, see below) and every cell in the girl's body lacks the second X chromosome (monosomy). However, other alterations in the X chromosomes can also lead to TS. Some girls with this karyotype may have two X chromosomes but one of those X chromosomes may have certain abnormalities (for example a partial deletion).

In other cases a girl may be missing the X chromosome from only some cells in the body (represented as 46,XX/45,X), or there may be abnormalities in the second of the X chromosomes in only some cells in the body. In both cases, this means that only some body tissues are affected. This karyotype is known as the *mosaic* form of TS. Girls with TS



in mosaic form often have less obvious physical characteristics. They may have functioning ovaries which means they may undergo spontaneous puberty. And they may therefore be fertile for a short time during their life [3]. The special problems encountered by girls with TS in mosaic form who are able to conceive are discussed in detail in Chapter 3.

Some women have a mosaic karyotype that includes a small ring X chromosome which is thought to occur as a result of breakages in the X chromosome. This ring X karyotype is generally associated with autoimmune disorders and deafness but not with congenital abnormalities [4]. However, some girls with small ring X chromosomes have severe learning difficulties and dysmorphism, while those with larger ring X chromosomes are thought to have a higher incidence of bicuspid aortic valves.

The cause of the missing or abnormal X chromosome in TS is not known. No risk factors (for example, raised maternal age, diet during pregnancy, etc) have been identified as being associated with an increased risk of having a baby with TS [3]. It has been estimated that only about 1% of affected foetuses survive to birth and that as many as 10% of spontaneous miscarriages have the chromosomal abnormality characteristic of TS [1,2].

Genetic counselling

When the parents of a baby with TS are first told the diagnosis, whether during pregnancy, at birth, or later in childhood, they will have countless questions. 'Why me?', 'Did I do something wrong?', 'Could I have prevented this happening?', 'If I have other children, will they also have TS?'. These questions should ideally be addressed by a specialist in genetic counselling. Genetic counselling is not always offered, but the general practitioner, obstetrician or paediatrician can refer patients to a local genetics department. Each region of the country has at least one genetics centre.

Parents who discover during pregnancy that they are expecting a baby with TS are often frightened and confused. It is important that they are given clear, comprehensive and correct information about TS. Ideally they should have a frank discussion of the problems associated with the occurrence of TS within the family, as well as the likelihood of another baby being affected (it is rare for one family to have more than one child with TS). Counselling will also cover the various physical signs the baby girl may have. It is particularly important that the parents are told that their child is very likely to be of short stature and that her ovaries will probably not be functioning. The information given to parents must be sufficiently clear and comprehensive, to enable them to make



an informed choice as to the course of action most appropriate in their situation. It is sadly the case that some pregnancies where TS is diagnosed *in utero* are terminated because parents do not receive accurate information on TS or an adequate explanation of living with TS. Similarly, some parents receiving a diagnosis of mosaic TS *in utero* may be led to believe that their child may not be badly affected and then be upset when their baby is born with more physical signs of TS than they were expecting.

Genetic counselling is also important for the parents and their daughter if she is diagnosed with TS either in childhood or adolescence. For a family facing the future with a child affected by TS, counselling should address their questions and fears and help them cope with the situation. Above all, counselling should emphasise that girls with TS can and often do lead healthy, happy lives.

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Personal Experience of TS

For the love of a child – Michelle on *in utero* diagnosis

As I opened the envelope the only thing I thought was strange was that the letter was on expensive yellow paper instead of the usual white paper with the blue NHS symbol on the top. Little did I know that the next three words I saw would have such an effect on me and probably change all our lives forever. The words 'your AFP test' filled me with fear. I was afraid to read on but I had to. My test result was abnormal. I saw the words 'Down's syndrome and spina bifida.' I was just stuck to the spot. I had a 1 in 13 chance of having a baby with an abnormality.

The following Tuesday Tom and I went to the clinic for a detailed scan. We were seen by a doctor, a specialist midwife, another midwife and a lovely lady whose job title I did not see. All these people in one room to see one baby. All I could think of was that things must be very bad.

As I looked at the screen, I saw a little baby that looked like any other, but glancing at the people who were doing the scan, I could see they had such concentration on their faces and no expression in their eyes. Finally, the doctor got up, tore a photograph off the scanner and left the room. We didn't know what to make of this. It could be something, it might be nothing, but when the door re-opened with the doctor and a consultant, I started to imagine all sorts of terrible things.

Finally, it was over. All I remember that Tom and I said to each other at this stage was 'This is bad isn't it' but, before we could answer each other, the consultant and specialist midwife came into the room. They told us that because the baby wasn't being very co-operative we would have to come back after lunch for another scan.

When we arrived back at the hospital we were called back into the scan room virtually straight away. This time there was the consultant and the specialist midwife. We were expecting yet another couple of hours of scanning like before, but were both surprised when, after about 20



minutes, the consultant stopped and said, 'I have seen a lot of swelling on your baby's head, something of cystic appearance on baby's neck and also a lot of swelling to baby's abdomen and, in my opinion, this is consistent with a chromosomal abnormality.' At the time, this was like being spoken to in a foreign language. We did not understand what all of this meant.

Back in the consulting room the consultant told us that what we were about to hear would be upsetting and we might not be able to take it all in. We should ask as many questions as possible. Then came the bombshell. The consultant told us that in her opinion, the chromosome abnormality was Chromosome 21 - this means Down's syndrome. I was inconsolable and Tom was in a state of shock. That's when we asked the questions. How? Why? When? Why us? What did we do wrong? Is this our fault?

When we calmed down a bit and it started to make a bit more sense, we were told our options. We could terminate the pregnancy at that stage or, to make doubly sure, we could have an amniocentesis test, although that had a 2% miscarriage rate. We decided to have the amniocentesis and were given an appointment for the following morning.

I will never forget the next day. We were greeted with hugs, kisses and genuine concern from the medical staff. We realised that the care we were receiving was excellent and the people we were dealing with were not just people in white coats and blue uniforms - they genuinely felt for us and what we were going through. Everyone was so nice. They showed their caring, compassionate sides, which made the procedure easier. After the sample was taken we were told that the test result could take up to three weeks to be confirmed but, because of our situation, they would ask for a FISH test to be carried out and this result should be back a lot quicker.

I was sitting at home the following afternoon when the phone rang. I remember saying, 'Well, you can tell me the results now,' and the specialist midwife said, 'Michelle, it is not Down's syndrome, it is Turner



syndrome,' and when she explained what Turner syndrome, or TS, was I felt like the miracle we had asked for the previous day had been granted.

We arranged to see the specialist midwife and my own consultant who had looked after me during my previous two pregnancies the next day, so all the facts about the syndrome could be explained to us in full. They began by telling us our baby was a little girl and that TS can only ever occur in little girls and is generally characterised by short stature and non-functioning ovaries. This in turn leads to absence of puberty and infertility. They told us that the shortness in stature can be improved through growth hormones and that puberty can be artificially started by giving girls oestrogen when they reach an appropriate age.

When the child was born she would probably have puffy hands and feet which, over time, would rectify themselves. There could also be feeding difficulties as some of the children can have a high arched palate but, with the aid of special teats, this can be overcome. We were told that a more serious part of TS is a coarctation of the aorta - that is narrowing of the main artery in the heart - and unfortunately, our child did have this condition. But hopefully, through surgery, this could be put right.

We also learned about the other tell-tale signs of TS - webbing of the neck, widely spaced nipples, and sleeping difficulties - and that intellectual abilities span the normal range. However a child with TS can be very shy and lack confidence but, with the appropriate support of her family and friends, this need not be a problem.

I have asked myself on many occasions, whether I would have preferred not to have known about the problem until much later. I can honestly say that, even though we have suffered a lot of pain and suffering, I am grateful that I know now. Not only do I know what to expect and look out for, but the care and the monitoring of the baby and me has been remarkable.



I made it my mission to find out everything I possibly could on TS and by talking to support groups and parents who have gone through it before me, the information I gained gave me enormous strength and the will to carry on. There is nothing better than talking to people who have first-hand experience, even if they are strangers. Those strangers turned my life around when I didn't know which way to turn.

During the time we were being given all the relevant information about our child's condition we were also given an appointment for genetic counselling to find out why and how this condition occurred at the time of conception. I was extremely pleased that I took up this offer as the attention to detail and the explanations given were excellent and it was at this point that a weight seemed to be lifted from my shoulders. For the first time in months, I seemed to be able to put the pieces of the puzzle together.

It was reassuring to be told that it was neither Tom's fault nor mine that TS had occurred in our child. Just having that knowledge gave us the strength to carry on and focus on more important things. I left the appointment feeling a lot more knowledgeable and confident about the future. All I can say to anybody who is considering genetic counselling but is unsure about whether to go or not is, 'Go, get the answers to your questions and maybe you will find peace of mind'.

Our daughter Molly-Jo is now eight weeks old. She was born with no physical signs of TS, apart from swelling of both feet. When I first saw her, the relief was indescribable. The feelings I have now, apart from obvious relief, are sometimes anger and isolation, but then they change to gratitude, luckiness and happiness.

Sometimes I look back and wonder if during the pregnancy we were sometimes given too much information. Our minds often felt just so full of problems. My plea would be for this information to be spread over the months before the child is born, so that parents have time to digest it instead of having to cope with everything in one go. Once the



baby is born, however, my view is that parents are not given enough information. After the child is born, you need to be reassured that everything is all right, and whether your child has problems associated with the syndrome or not. I found myself telling the midwife what she should be looking for. It's quite frightening to realise that you know more about your child's condition than the midwife does.

With all the care and attention Tom and I were given during my pregnancy, it would be impossible to think of any occasion when we were treated unsympathetically, but I do feel that sometimes, because the woman is the person who is carrying the child, the focus is rather too much on her. Often it may actually be harder for the father. Mother and father should be involved in everything because the father is very much a part of the equation.

So to all the people who are just starting out along this very hard road, please believe me when I say that, with all the help that is available and the love and support that is given, you will come through the other side. And whatever you choose to do and whatever decision you make, always remember, you made those choices for the love of a child.



Specialist paediatric care in Turner syndrome

A multidisciplinary approach

As we have seen in Chapter 1, a girl with TS may be affected by some of the characteristic features of the disorder. This means that she may be under the care of one or several specialists, for example an ear, nose and throat (ENT) specialist or a cardiology specialist, and thus be receiving a variety of treatments. Every girl should be treated according to her individual needs and ideally a multidisciplinary approach should be taken to treatment, allowing the girl – and her family – to have the benefit of specialist care and advice regarding all the symptoms she may have.

Due to the syndrome, some underlying problems a girl has may not be fully appreciated. However, it is important that all potential difficulties are addressed so that the girl with TS can have the best possible quality of life and reach her full potential. An example is the child with small stature and heart problems but who also has recurrent ear problems. Alongside her other problems, the recurrent ear infections may have been neglected, but it is important that she also receives specialist care for them.

This chapter summarises the specialist areas of treatment likely to be encountered by a girl with TS and her parents. TS is a lifelong condition, however, and the particular needs of the adult TS patient are discussed in detail in Chapter 3.


Endocrine problems

Those diagnosed with TS prenatally or at birth should be under the care of a paediatrician or a paediatric endocrinologist (a specialist in hormonal problems) immediately after birth.

Growth problems and short stature

As discussed in Chapter 1, short stature is one of the features almost invariably present in girls with TS and is usually the key to the diagnosis of TS after infancy. However, girls with TS usually grow at a normal rate until they are about 3 or 4 years old. Only then does their growth begin to slow down (usually between the ages of 3 and 12 years) [1]. Another feature is the failure of the growth spurt that girls experience at puberty.

Recent research has shown that growth rate can also be reduced in the first 3 years of life and therefore a diagnosis of TS should be considered in any girl who fails to thrive even at a very young age [2]. Regular measurement of height and plotting against the charts in the 'Red Book'



(Personal Child Health Record) is important if there is any suspicion that a girl is not growing as fast as her peers. These growth charts use the parents' height to calculate a projected adult height for the child. Examples of the charts against which children's rate of growth is measured can be obtained from the Turner Syndrome Support Society.

For the majority of girls with TS it is possible to increase their final height potential with growth hormone (GH) treatment. This is something that the paediatric endocrinologist will discuss fully with the girl's parents. Additionally, some specialists may recommend using the anabolic steroid oxandrolone to promote growth. Recommendations for the management of short stature using GH are given in Table 2.1 [1].

Table 2.1: Growth hormone therapy in Turner syndrome [1]

For growth hormone treatment to have maximum effect TS should be diagnosed and treatment started as early as possible

For girls below 9 years of age, therapy can be started with growth hormone alone

Recommended starting dose: 0.05 mg/kg/day

Growth should be monitored at intervals of 3-6 months

Dose is calculated on the basis of a girl's weight and should be individualised if response to growth hormone is inadequate

For girls aged 9-12 years (or in girls aged over 8 years who are already far below the normal growth curve) the anabolic steroid oxandrolone may be considered in addition to growth hormone

Anabolic steroids should never be used alone to promote growth

The dose of oxandrolone now used is low and does not cause the side effects that were seen in the past (virilisation – the appearance of masculine features)

Therapy with growth hormone (with or without oxandrolone) should be continued until final height has been attained or until the epiphyses of the bones have fused and the patient's height has increased by less than 2 cm over the previous year

To induce puberty, oestrogen should be given. However, because it limits growth of the bones, it should be timed so as to minimise any negative effects on growth and attainment of adult height while inducing puberty at an approximately normal age

How successful this treatment is depends on a number of factors, such as the age at which GH treatment is started, how the girl responds to the treatment, and how well the parents and the child herself comply with treatment regimen. Compliance with treatment (now sometimes known as concordance, see box on page 25) is important in GH therapy because the treatment has to be given daily for a number of years, without any instantly visible rewards [3].

Many interrelated factors affect compliance, including patient and parental expectations, and understanding of the treatment and the nature of the problem being treated. Training in administering GH treatment and patient and parent involvement in choosing the most suitable device for administration are also important as offering choice has been shown to improve compliance with treatment. The various devices available for administration of GH (needle-free devices, pens, auto-

injection devices) are set out in Table 2.2 and examples of these devices are shown in Figure 2.1 [4].




Figure 2.1: Some of the devices used for administering GH treatment.

Table 2.2: Some growth hormone delivery devices offered by various manufacturers

Company/device	Features of device
Ferring/needle free device	Multi-dose, device delivers GH in less than 1 second without a needle. Requires daily loading, dose delivery by pressing a button.
Pfizer/pen	Multi-dose with dual-compartment cartridge, digital display of dose, dose injected after pressing a button, manual needle insertion.
Pfizer/syringe	Fixed dose single use dual-compartment injection device, manual insertion, range of doses available, room-temperature storage, preservative-free
Novo Nordisk/pen	Multi-dose, liquid rHGH, optional automatic needle insertion, dose delivery by pressing a button, room-temperature storage.
Serono/automatic injection	Multi-dose, hidden needle, automatic needle insertion and injector device, dose delivery after pressing single button.
Lilly/pen	Multi-dose, digital display of clicks, dose injected after pressing a button, manual needle insertion, optional needle cover.
Ipsen/pen	Multi-dose, with liquid rHGH , digital display of dose, dose injected after pressing a button, manual needle insertion.
Sandoz/pen	Multi dose, with Liquid rHGH, dose delivery by pressing a button, manual needle insertion.
Ongoing research and development in this area means that the range of devices above will always be changing. Adapted from Burke, 2001 [4], with permission. Our thanks to Angela Casey for her help updating the photo and table.	





The GH used in the treatment of short stature in TS is biosynthetic and identical to human growth hormone derived from the pituitary gland. It therefore has the same effects as the natural hormone. GH treatment, using for example Zomacton® 4mg Injection (somatropin, Ferring Pharmaceuticals Ltd) is commonly given at a dose of 0.33 mg/kg/bodyweight (approximating to 9.9 mg/m²/body surface area) per week given by seven nightly subcutaneous or transcutaneous injections. Girls receiving the GH treatment and their parents need to be carefully instructed in storing the GH, reconstituting the GH powder using the solvent provided and also in administering the injection itself.

Several manufacturers of GH offer a comprehensive package of additional support to address all potential difficulties that may arise with GH treatment. These include training and compliance visits from a trained nurse, coordination of repeat prescriptions, home delivery of supplies, follow-up monitoring and progress reports, and free phone contact with 24-hour nurse support [4].

GH treatment started early enough (see Table 2.1) and administered regularly has been shown in clinical studies to normalise growth during childhood for groups of girls with TS and adult heights near or within normal ranges can be achieved for most girls with TS [5]. The age at which GH treatment is given is critical to the ultimate adult height in TS, with a longer period of GH treatment prior to pubertal induction (see below) thought to be beneficial.

Adolescence and puberty

The ovaries of most girls with TS do not produce adequate amounts of sex hormones to induce puberty and the development of secondary sexual characteristics such as breast development, so oestrogen therapy is required at an appropriate age. The dose of oestrogen given is increased slowly and gradually over a number of years to mimic natural development and, once it has reached a sufficient level, the hormone progesterone is also introduced to induce withdrawal bleeding.

The addition of oestrogen throughout adulthood is also important to maintain the health of the heart and for the proper mineralisation of the bones to prevent osteoporosis in adult life. The topic of osteoporosis in TS is discussed in more detail in Chapter 3.

The lack of ovarian function in girls and women with TS effectively means that their hormonal situation is equivalent to the postmenopause. Because they will be receiving lifelong oestrogen therapy it is important that, like postmenopausal women, girls and women with TS are treated with natural human oestrogens like 17-beta oestradiol or similar



hormones (as used in most hormone replacement therapy for the treatment of menopausal symptoms). In terms of effects on the heart, circulation, and liver, natural oestrogens are preferable to the synthetic ethinyloestradiol found in the contraceptive pill.

In a small number of girls with TS, puberty may occur spontaneously and they may have menstrual periods. A handful have even been able to conceive, however this is very rare. Certain features of puberty are not controlled by the ovaries and it is important that parents realise that, even if their daughter develops underarm or pubic hair, this does not mean that she has functioning ovaries. Spontaneous puberty also means that the bones will fuse and stop growing and any GH treatment will cease to be effective, although there is no current evidence that these girls are any shorter as adults than those whose puberty is induced with oestrogen treatment.

It is also important to stress that, although most girls and women with TS have non-functioning ovaries, they do have a normal uterus and vagina and are able to have an entirely normal sex life. The topic of sexual health in both girls and women with TS is discussed in detail in Chapter 4. The special problems encountered by girls and women with TS with functioning ovaries who are able to conceive and the particular need for counselling are also discussed in detail in Chapter 3

Thyroid function

Between 10 and 30% of girls with TS will have an underactive thyroid gland (known as primary hypothyroidism) [6]. However, hypothyroidism often has no obvious clinical symptoms so the endocrinologist should measure levels of thyroid stimulating hormone (TSH) at the time TS is diagnosed and then every year to check that the thyroid gland is functioning properly.

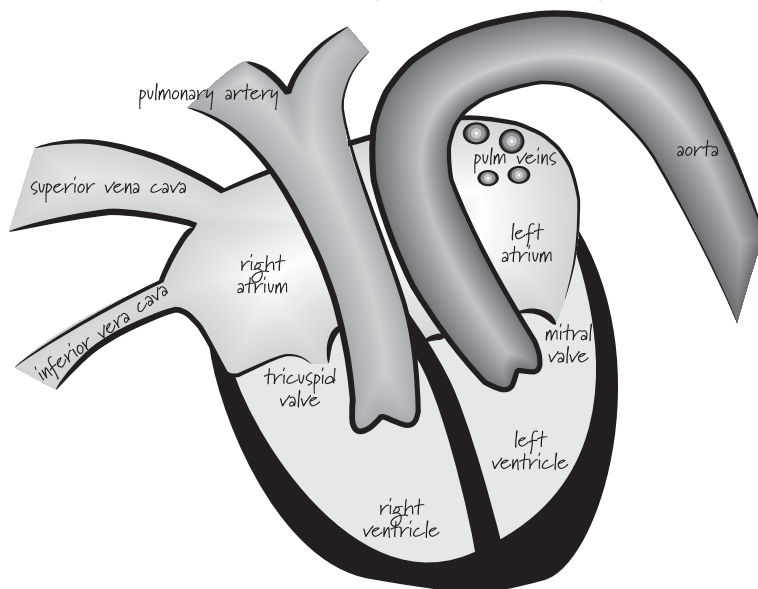
Heart problems

Babies with TS born with a heart murmur or narrowing (coarctation) of the aorta will need expert assessment by a cardiologist and sometimes require surgery to correct the problem.

The most common cardiac abnormalities in TS are congenital heart defects, also known as cardiovascular malformations. These occur in approximately three-quarters of fetuses affected by TS and 30% of babies that survive to term [7-10]. Figure 2.2 shows the anatomy of the heart and lists the various features. Of these cardiovascular malformations, defects involving obstruction in the left side of the heart, especially bicuspid aortic valve, account for 30-50% [11]. Other less common left-sided problems are aortic stenosis (or narrowing),

coarctation of the aorta (narrowing of the aorta just beyond the point where the arteries branch off for the head and arms) and anomalies of the mitral valve.

the heart is like a house



Muscle walls	Myocardium
Two upstairs rooms	Right atrium and left atrium
Dividing wall upstairs	Atrial septum
Two downstairs rooms	Right ventricle and left ventricle
Dividing wall downstairs	Ventricular septum
Two doors between upstairs/downstairs	Tricuspid valve and mitral valve
Two pipes entering from the body	Superior vena cava and inferior vena cava
Four smaller pipes entering from the lungs	Pulmonary veins
Large pipe exiting heart going to the lungs	Pulmonary artery
Large pipe exiting heart going to the body	Aorta

Figure 2.2: The analogy between the heart and a house, where the various anatomical structures are equivalent to walls, rooms, doors, and even plumbing, helps us to understand how the heart works and, more importantly, what can go wrong. Reproduced with permission from Dr Angela Lin who created this image of the heart. She is a medical geneticist with training in paediatric cardiology and an interest in TS. Dr Lin points out that this diagram is not drawn to scale, nor is it anatomically complete, but aims to convey the basic structure of the heart.

Rarer cardiovascular defects are aortic dilatation and dissection – a thinning and ballooning of the wall of the aorta, which can ultimately



rupture, causing death [7-10]. In very rare cases, girls with TS can have aortic dissection but have no underlying heart problem that would bring them to cardiac care. For this reason, it is recommended that all girls with TS should be regularly monitored for dilatation of the aorta.

In addition, the blood pressure should be monitored every year in all patients with TS. Patients with a structural heart defect should also be advised of the need to take antibiotics before any dental procedure to prevent bacterial endocarditis (inflammation of the inner lining of the heart and valves due to infection).

If the child diagnosed with TS is not found to have any heart defect on their initial cardiac evaluation, the cardiologist will recommend a repeat cardiovascular examination and echocardiogram some time in adolescence (12-15 years of age), which should pay particular attention to the aortic root. If at that stage any enlargement of the aortic root is detected, further echocardiograms are required to establish the severity of the problem and blood pressure should be monitored. In those with significant aortic root dilatation, treatment with beta-blocking drugs may be appropriate.

High blood pressure (hypertension) is quite common in adolescents (and adults) with TS, even those without heart or kidney defects, and the blood pressure should be checked regularly and hypertension treated where necessary.

Ear, nose and throat problems

Girls with TS are more prone than others to middle ear infections (otitis media). Recurrent infections can lead to deafness if not promptly treated [12]. For this reason a consultation with an ear, nose and throat (ENT) specialist is advisable.

In girls with TS, middle ear infections occur particularly between the ages of 1 and 6, and most often around the age of 3 years. The cause of these repeated infections is not known, but slowed growth of the temporal bone in the skull may be important [1]. There may also be problems with mastoiditis – a potentially dangerous inflammation of the honeycomb-like air cells behind the lower part of the ear – when infection spreads from the inner ear. Middle ear infections may also lead to the formation of cholesteatoma – a mass of non-cancerous tumour-like cells shed by the outer layer of an infected eardrum that can invade the middle ear through a perforation in the drum and damage the inner ear.

Treatment of these repeated middle ear infections is therefore vital, and the specialist may recommend the insertion of grommets to help drain

the middle ear. In all cases careful follow-up is required. For any girl of short stature with repeated ear problems, the ENT specialist should be suspicious of a diagnosis of TS and refer the patient to a paediatric endocrinologist.

Hearing tests are also important. Girls as young as 6 years of age can have what is known as sensorineural hearing loss [1]. This is an inability to hear high frequency sounds and is caused by defects in the inner ear mechanism. The condition is progressive and commonly leads to hearing problems in later life for those with TS.

Kidney problems

Some girls with TS will have congenital malformations of the urinary system, for example a horseshoe kidney (Figure 2.3). Although most of these inborn abnormalities do not have any clinical significance, some may result in an increased risk of high blood pressure, urinary tract infections (UTIs), or hydronephrosis – ballooning of the urine collecting system in the kidney resulting from obstructed flow of urine at any point below the kidney [13]. Girls with TS should therefore have a renal ultrasound scan at the time TS is diagnosed and, if any abnormality is detected, appropriate treatment will be given by the specialist. Patients with any malformations should have regular ultrasound and urine tests.

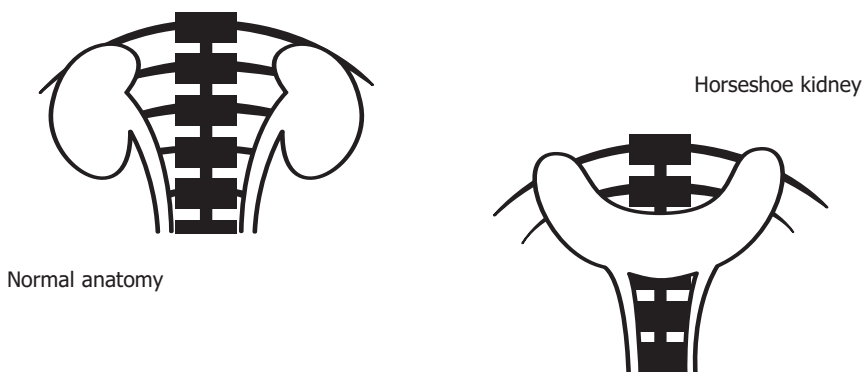


Figure 2.3: A horseshoe kidney is sometimes seen in girls with Turner syndrome.

Feeding difficulties

If they have a small jaw, or high arched palate, babies and young girls with TS may well have problems in feeding and/or talking [14]. In such cases, referral to a dietician and/or speech therapist may be recommended. They may also have various dental problems, particularly overcrowding and malocclusion (inability to bite efficiently), and therefore difficulties in eating. This tends to be less of a problem for girls who are diagnosed and treated with GH early. If a girl with TS has not



already had reason to visit an orthodontist, an orthodontic examination is recommended at around 8-10 years of age to prevent any potential problems.

Weight control

Girls with TS tend to put on weight, and their characteristic broad chest, stocky build and short stature, particularly relatively short legs, may exaggerate the appearance of this. Weight should be evaluated regularly and advice given on avoiding obesity [1]. Girls with TS should be encouraged to eat a healthy diet and take regular exercise, especially weight-bearing exercise, for example running or cycling.

Occasionally, girls with TS may suffer from the eating disorders anorexia nervosa and bulimia nervosa. Parents of daughters should always be aware of the potential for such problems to occur, but girls with TS may have low self-esteem and be particularly conscious of their self-image, putting them at higher risk of developing these disorders. The issues surrounding body image and self-esteem are discussed in more detail in Chapter 4.

Eye problems

Girls with TS may experience a number of eye problems. The most common are a strabismus (squint), amblyopia (failure of the retina to form sharp images) and ptosis (drooping of the eyelids). Regular eyesight tests and an ophthalmological evaluation should be part of the paediatrician's regular examination and, where necessary, the patient should be referred to the ophthalmologist for specialist care.

Skeletal problems

Girls with TS can have an increased risk of congenital dislocation of the hip. Around 10% of girls with TS develop scoliosis (curvature of the spine), most commonly during adolescence. The paediatrician will evaluate the patient for orthopaedic problems and refer to the orthopaedic specialist where necessary.

Other problems

Lymphoedema

Although swelling (lymphoedema) of the hands and feet is most common in babies with TS, it does not always go away once the child starts walking. It is thought to resolve in only a quarter of cases [15]. Lymphoedema can also be associated with the start of GH or oestrogen therapy.

Lymphoedema is a chronic condition characterised by swelling of one or



Figure 2.4: The presence of lymphoedema at birth is a characteristic sign of TS. It can persist into adult life.

more limbs and is caused by failure of the lymphatic vessels to drain fluid from the tissue spaces (lymph). The primary lymphoedema seen in TS is caused by a developmental defect of the lymphatic vessels during foetal growth, reducing their ability to function [16], and affects mainly the hands and/or feet (Figure 2.4). Secondary lymphoedema is much more common and can be caused by tumours, cancer treatment, or trauma.

If left untreated, the swelling may increase and changes in the skin may develop, for example it may become very dry and wart-like lesions develop. These are in fact lymphatic vessels that have become so swollen they bulge under the skin (papillomata). Sometimes small breaks in the skin cause lymph fluid to leak on to the surface of the skin (lymphorrhoea). There is also a very small risk of developing lymphangiosarcoma on the affected limb(s) if the swelling remains untreated.

Because the fluid causing the swelling has high concentrations of protein it is an ideal medium for bacterial growth and sufferers therefore have an increased risk of developing infection (cellulitis), the tell-tale sign of which is inflammation around the toe nails.

Girls with TS are particularly at risk from ingrowing toe nails as they often have increased transverse curvature of the nails. This makes the nails prone to ingrow if they are badly cut, or cut too short. If footwear is too short, too tight (which may be unavoidable due to variable swelling) or pointed, this will result in toe nail problems. Also, girls tend to be reluctant to complain of pain at the affected site so the problem is only brought to the attention of the parents at a late stage. Previous infections make the toe nail prone to recurrent infections.



Patients may have difficulty with clothing and shoes because of the size of the swollen limb(s) and this may in turn cause some psychological distress for both the child and the parents [17]. Particular care should therefore be paid to footwear, for example always having the feet measured before buying shoes, avoiding slip-on shoes or sling-backs or shoes with high heels. The same applies to restricting sleep suits and babygros for babies. Footwear should only be fitted by a qualified shoe fitter, e.g. Clarks and Start Rite, with particular attention being paid to the toe box area of the shoe. Is it deep enough, wide enough, 'foot shaped', and is the material of the shoe upper soft and flexible without seams or stitching? Where the problem is severe, an excellent choice of stock footwear is available through a podiatrist or orthotist.

Referral to a lymphoedema specialist is essential to provide appropriate treatment and instruction in self-care, and to prevent any possible secondary effects. Treatment consists mainly of the following:

- Specialist compression hosiery which should be worn daily and removed at night
- Instruction on simple lymphatic massage to assist drainage from the swollen area
- Encouragement to use the limb(s) as normally as possible
- Advice on skin care to prevent infection.

Skin care advice is aimed at avoiding any cuts or breaks to the skin on the affected limb(s), cleansing and covering any broken areas as soon as possible, and treating any infection immediately. If an ingrowing toe nail is suspected, immediate podiatric care should be sought. The toe should not be wrapped with an airstrip type dressing or tape as this will compound the problem. Tubular dressing with sterile gauze is best. Skin should be moisturised daily to prevent drying and keep it supple, and care should be taken when cutting toe and finger nails. If the nails are a particular problem their care should be handed over to the podiatrist. Hygiene is also very important in prevention of infection especially between the toes. The feet should be washed daily and dried gently between the toes before applying a small amount of talcum powder as a lubricant. Do NOT use creams between the toes unless specifically told to do so.

Occasionally, if the swollen limb is too large to fit hosiery, or skin changes have developed, more intensive therapy is required. This consists of a 2-3 week course of daily bandaging, a more specialised form of massage (manual lymphatic drainage) and sometimes the use of an intermittent pneumatic pump. This intensive treatment reduces the swelling and reverses the skin changes, allowing application of compression hosiery.



The majority of patients with TS who have lymphoedema do not currently receive appropriate treatment [15]. However, there are specialists throughout the UK who can effectively treat this condition and there is therefore no need for people with TS to suffer lymphoedema without treatment.

Plastic surgery

Surgery is available to girls who have very drooping eyelids and webbed necks. However, the risk of excessive scarring (known as keloid scarring) is high in TS. If any surgery is considered the benefits of surgery should be carefully weighed against the risk of unsightly scarring. This can apply even to simple procedures such as ear piercing [1]. When consulting a plastic surgeon for any procedure, it is wise to ask if they have experience of treating girls with TS.

Prevention is better than cure

It is important to stress that not every girl with TS will have all the problems discussed in this chapter. However, prevention is always better than cure and it is wise to be informed about possible problems in order to prevent them developing.

Compliance *versus* concordance

Historically, the relationship between doctor and patient has been one of 'doctor knows best'. It is now increasingly recognised, however, that a partnership between doctor and patient is important. The concept is moving away from 'compliance' with treatment (implying obedience) to one of 'concordance', which stresses that treatment is an agreement reached after negotiation between patient and doctor (or other healthcare professional) that respects the beliefs and wishes of the patient.

Source: Stitches, The Journal of Medical Humour





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A father's view from Mal

It is September 2000 and the location is a hospital in Cyprus where I am a serviceman and live with my wife Tracey, our son Daniel and our daughter Jenna. Jenna had been diagnosed at the age of two with a high-capacity bladder and horseshoe shaped kidney. We were told that this was not a major problem but that she should have a scan every year to ensure that everything was okay. Jenna was coming up to her ninth birthday and every year her scans had revealed no problems. The consultant paediatrician was happy with the latest results and felt that future scans were unnecessary. We were thrilled. But before he signed her off he asked if he could carry out one last test as he considered Jenna to be small for her age. My wife and I laughed at this as I am only five feet six inches tall and my wife is only five feet four, and both Jenna's Grandmothers are exactly five feet tall. We knew that Jenna would never be the tallest girl in the world but we agreed to the blood test. At the time the consultant did not inform us what he wanted to test for, and we didn't feel compelled to ask.

A week later we were called in to discuss the result with the consultant. Tracey and I were so sure that everything was okay that I agreed to go on my own as she was on a training course. I arrived at the hospital and knew instantly from the consultant's face that something was wrong. He asked me to sit down and naturally I became even more uneasy and concerned. He told me that Jenna had Turner syndrome, or TS, and went through the diagnosis in what seemed like great detail at the time – only later did I realise that he had only scratched the surface.

I felt like I had been kicked in the chest, and everything became a bit of a blur. I had been able to take in a lot of what the consultant had told me, especially the major characteristics of TS relating to short stature and non-functioning ovaries. I asked him what chances Jenna had of conceiving naturally later in life and he gave an assessment of



50/50. Initially this gave me some hope – I knew that many women find it difficult to conceive and Jenna would still have a chance of having children naturally. But this hope quickly faded. The consultant informed me that Jenna has an extremely rare TS karyotype and that she would probably need to have her ovaries surgically removed otherwise there was a strong possibility of ovarian cancer from the second decade of her life.

I left the hospital feeling numb and was already asking myself questions. How would I tell all of this to my wife? How would she react? Why us? Why Jenna? What do we tell Jenna? Will she be able to cope? What do we say to her brother? How will he react? Whose fault is it? Is it anyone's fault? Could it have been prevented? Jenna was nearly nine years old and I had been informed that cancer could occur from around the age of ten, so would she get it? Did she have it already? What will her grandparents say? Will they treat Jenna differently? Will we treat Jenna differently? The list of questions went on and on. My head was in a spin and I wondered whether or not we would be able to deal with it.

When I sat down to tell Tracey what had happened I tried to be calm and rational but my emotions were running high. I knew what I wanted to say but found it hard to get everything out. The lump in my throat was getting bigger and then it happened – I cried, something that men aren't supposed to do. I cried like a baby and so did Tracey. We hugged and held each other, feeling sorry for Jenna, feeling sorry for ourselves, not saying much at all. It was all so very hard to take in. At the time, the worst thing for us to accept was that our daughter (who at the tender age of eight was already a kind, caring and responsible child) possessed so many maternal characteristics but would not be able to have children of her own naturally.

Then it struck us. Jenna was still the same gorgeous little girl she had always been but she had a condition that needed careful monitoring and specialist treatment. Change in some areas of her life would be inevitable but we were convinced that we would manage these together,



as a family. From the very first day we knew how important it was that when she reached the appropriate age Jenna should know about her condition and how it would affect her.

The next step was to find out as much information as possible about TS. Although they were sympathetic, the medical officers on the island knew less than us about TS but promised to find out more information. All they managed to do was send us a few photocopied paragraphs from medical journals giving an extremely brief description of TS. I was due to return to the UK on posting the following July and had been informed that Jenna would be able to get all the treatment she required then. I made it clear that I was not happy with this and managed to persuade the Service authorities to fly us all back to see a consultant paediatric endocrinologist in England within a couple of months of receiving the diagnosis.

In the meantime, prior to attending the specialist appointment, Tracey and I used the internet to become better informed on all matters relating to TS. To our delight we came across the Turner Syndrome Support Society (TSSS) web page and immediately contacted them for help and assistance. It was fantastic to make contact with people who knew instinctively what we were going through. It was arranged that I should meet Steve (who manages the TSSS web page) and his wife Susie (who had been diagnosed with TS as an adult) and suddenly we didn't feel alone anymore. From them I learnt so much about the practicalities of TS and of the role of the TSSS in supporting girls, women and their families with the management of this condition.

Susie especially was able to convey to me what it was like to grow up with TS. She stressed that it would be different for Jenna as her condition had been diagnosed early enough for her to receive growth hormone and other treatments that would improve her final height and general well being. I received so much information and positive feedback from both Steve and Susie that I felt much better prepared to help our little girl with whatever she would have to face.





Later that month we flew back to the UK to see the consultant endocrinologist. The meeting went as well as could be expected but in reality we had already obtained so much information ourselves that we didn't learn anything new. It was confirmed however that Jenna would definitely need to have her ovaries removed. We had mixed feelings about this but ultimately knew that there wasn't really a decision to be made - the possibility of an ovarian tumour in later life meant that it was crucial for surgery to be carried out. We decided to wait until the surgery appointment was confirmed before broaching the subject with Jenna.

I managed to persuade the military authorities to start Gtt treatment for Jenna while we were still in Cyprus. I also persuaded them to post me back to the UK earlier than originally planned. I felt that in doing so we would be closer to specialist facilities that would ultimately help with the management of Jenna's condition.

We were so proud of Jenna - from the outset she had been so courageous in coming to terms with TS and the fact that she required daily Gtt injections. We were so worried as to how she would react, yet in the end our fears were groundless and the injections quickly became part of her daily routine. We were also proud of her brother Daniel. Although only a year older than Jenna, and not quite fully understanding the implications of TS, he too took everything in his stride and was totally supportive to her. He said that he understood that Jenna would face difficulties as she grew up but all he wanted was 'for her to have the best possible life'. It was at this point that I realised just how strong both our children are. Whenever we discuss a new aspect of Jenna's condition we always do so initially with Jenna alone. Jenna insists however, on explaining everything to Daniel afterwards as she feels that it is important for him to know and to be involved.

Just before Christmas Jenna was admitted into hospital for the surgical removal of her ovaries. Tracey and I felt that it was important prior to the operation to inform Jenna of exactly what was going to happen to



her and the implications. We have not told her that she cannot ever have children but that, just like needing help to grow, she will need help to have children if that is her wish. Whether this will be through fostering, adoption, IVF, or any other future technique that has still to be developed will be addressed later in her life. However, even at this early stage in Jenna's development, she knows that there will be choices to be made but also that her family will all be there to love, help, support and advise her through the difficult times that will inevitably arise as she grows up.

When we met her surgeon after the operation he informed us that surgery had been a wise choice as there were clear indications that all would not have been well if her ovaries had not been removed. This was a huge relief to us – the prospect of what could have happened to her did not bear thinking about. We were so grateful that Jenna's condition had been diagnosed by her Cypriot doctor early enough. None of the UK specialists she had seen since the age of two had been able to arrive at the diagnosis and this is a major concern. Just how many girls are slipping through the net and not being diagnosed early enough?

A lot has happened since Jenna's condition was first brought to our attention. Initially it was as if our world was coming to an end and we wondered whether or not we would be able to manage. Now, even



though Jenna has only been on growth hormone for just over 18 months, we have already seen a marked improvement in her growth. She has always been the smallest in her class and had quite enjoyed her nickname of 'titch'. But with the help of Gilt treatment Jenna is catching up with her peers (she is no longer the smallest in her class) and is going from strength to strength.

Like most fathers, I will do anything for my children and I am certain that with the continued love, support and encouragement of all her family



and friends Jenna will continue to have a wonderful life. Finding out that our daughter has TS initially seemed like the end of the world to us - in reality it was just the start of a new one.



Specialist adult care in Turner syndrome

A multidisciplinary approach to adult care

Girls with TS generally receive care from a wide range of medical specialists during childhood but once puberty has been induced and final height attained, the tendency has been for them to be discharged either from specialist TS clinics or the care of the paediatrician. The consequence has been that, until recently, adult women with TS had limited access to dedicated specialist care.

The healthcare profession has been slow to realise the problems that women with TS face, particularly the risk of cardiovascular complications and the long-term consequences of oestrogen deficiency. Women with TS are in fact susceptible to a number of specific medical problems as well as cardiovascular disease. These can include osteoporosis, thyroid, gastrointestinal and kidney disorders, hearing problems and diabetes.

Quality of life in women with TS may be improved with access to better care as adults. Women with TS require long-term follow-up and early treatment for any problems will ensure they have the best possible quality of life and that their life expectancy is as good as for women without TS. In many cases, relatively simple preventive measures will address these issues [1].

Coordination between paediatric and adult specialists is therefore essential for the smooth handover of care between childhood/adolescence and adulthood so that lessons learned in each field are shared. Ideally, adult women with TS should be under the care of a specialist TS clinic with a multidisciplinary team equipped to manage the specific medical problems associated with the syndrome. This should provide a regular review of all aspects of the woman's health and wellbeing. There are a number of established specialist TS clinics throughout the country and a complete list can be obtained from the TSSS. Table 3.1 shows a typical adult management programme [2].

Diagnosis of Turner syndrome in adulthood

As we have seen, TS is generally diagnosed either prenatally, at birth, in childhood or at the time of puberty. In some cases, however, the diagnosis of TS is not made until a woman reaches adulthood. This can happen if as a girl she entered puberty spontaneously but later has problems with amenorrhoea (lack of menstrual periods) or infertility [2]. As we have seen in Chapter 2, if a diagnosis of TS is suspected, chromosome analysis (karyotype testing) is used as confirmation.



Table 3.1: Suggested follow-up for an adult with Turner syndrome [2]

First visit

Karyotype testing
Renal and pelvic ultrasound
Thyroid autoantibody testing

Annual follow-up

Physical examination (height and weight used to calculate body mass index, blood pressure, cardiovascular assessment, etc)
Thyroid function
Lipid analysis
Blood glucose
Liver function
Renal function

Every 3-5 years

Echocardiography
Bone densitometry
Audiogram

Characteristic problems for women with Turner syndrome

We have seen in Chapters 1 and 2 that children with TS have a wide variety of characteristic clinical features. Similarly, women with TS face a unique set of problems.

Short stature

Women with TS who have not had access to GH treatment in childhood are almost invariably of short stature, with an average final adult height of between 143 and 147 cm. The particular issues surrounding the concept of body image for women with TS are discussed in more detail in Chapter 4.

Skeletal abnormalities

Many of the characteristic physical signs seen in women with TS are the result of structural bone defects [3]. Even after GH treatment to increase final height, women with TS may have disproportionately short legs and perhaps underdevelopment (hypoplasia) of the cervical vertebrae in the neck, which causes a short neck. There can also be associated scoliosis (curvature) of the spine.

Many women with TS have a wide carrying angle of the elbows (cubitus valgus), as a result of a developmental defect of the bones in the arms [2]. Similar problems may occur in the knees and some women with TS have short fingers and toes (Figure 3.1). It is mostly the case that these skeletal differences in TS cause few problems. The facial characteristic in TS are also mainly due to skeletal abnormalities (small jaw, a downward droop of the outer corner of the eyes, and epicanthic folds, a high arched palate, and low-set ears).

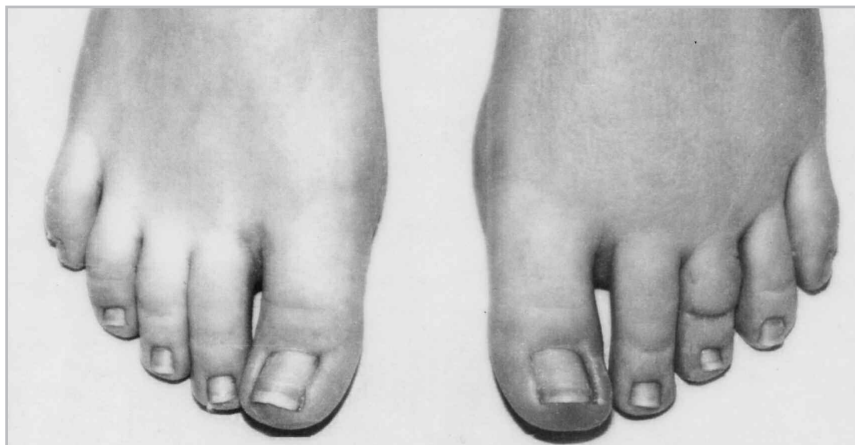


Figure 3.1: Girls and women with TS often have characteristically short toes and hyperconvex nails.

Osteoporosis

Bone mass increases steadily during childhood and adolescence and reaches a peak in the third or fourth decade of life, after which it levels out. The term osteopenia is used for decreased bone mineral density and early bone loss. The term osteoporosis is used to describe a more advanced stage of bone loss. If osteopenia is not treated it may result in the much more serious osteoporosis where the bones are unable to perform their supportive function.


In women with TS, peak bone mineral density is reduced by 25% [4,5]. Even though some of the diminished bone mass can be accounted for by short stature, women with TS have a higher fracture rate than women without TS [6]. It has been estimated that women with TS are 10 times more likely to develop osteoporosis [7]. Treatment with GH during childhood and oestrogen therapy improve bone mass in women with TS, although it still tends to be below normal [8].

In women with TS, oestrogen replacement therapy should therefore be optimised and measurements of bone density undertaken. Lifestyle advice regarding exercise and adequate calcium intake is important in order to keep bones as strong as possible and prevent osteoporosis. The significance of hormone replacement therapy in women with TS is discussed in more detail on page 37.

Lymphoedema

Swelling of the hands and feet is a key sign of TS in a newborn baby (see Chapter 2). Frequently this resolves as the baby grows but it can recur, particularly after oestrogen therapy is started. In most women, any





oedema that occurs can be controlled using support stockings and/or diuretic drugs. Lymphatic massage can also help. If lymphoedema is a serious problem then a consultation with a lymphoedema nurse specialist is often beneficial.

Gynaecological issues in women with Turner syndrome

In babies born with TS, the ovaries fail in the first few months or years of life (equivalent to a premature menopause) and they are generally too small to be identified by ultrasound in adult women (streak ovaries). Because of this early ovarian failure and subsequent oestrogen deficiency, most girls with TS do not enter puberty spontaneously, and breast development is minimal or fails to occur at all [2]. Spontaneous puberty does sometimes occur, particularly in girls with TS in the mosaic form. However, very few girls with TS maintain ovarian function long enough to be fertile in adulthood and only a very small number of women with TS become pregnant naturally [9]. Women who have not received oestrogen therapy will also have a small undeveloped uterus.

Fertility and pregnancy

Genetic counselling is strongly advised before any woman with TS who has functioning ovaries embarks on pregnancy because of a number of possible problems. The risk of miscarriage and stillbirth is high in pregnancies in women with TS and reports suggest that almost half of those babies surviving to term have some form of chromosomal abnormality (particularly Down's syndrome or TS) or congenital malformation (especially heart defects and neural tube defects such as spina bifida) [2,9]. Even though this historic data may exaggerate this risk, women with TS who become pregnant should be offered prenatal diagnostic testing (see Chapter 1).

It is also important that women who do have ovarian function are advised that there is a high risk of early ovarian failure and that this should be taken into consideration when planning pregnancy. The complex issues involved in the counselling of women with TS regarding fertility and pregnancy are discussed in more detail in Chapter 4.

***In vitro* fertilisation**

The majority of women with TS will be infertile and for them egg donation via *in vitro* fertilisation (IVF) is a possible option [10]. It is recommended that two embryos are implanted at most. The risk of miscarriage is high in women with TS, with only around half of pregnancies carried to term [11]. Pregnant women with TS may also be at increased risk of cardiovascular complications, particularly aortic root dissection [10]. All women should therefore have a full cardiological



assessment before becoming pregnant. Because hypertension is more common in women with TS this should be monitored and actively treated. However, pre-eclampsia does not seem to occur more often in women with TS than in those without [2]. Ideally, women with TS should therefore be cared for in a specialist obstetric centre. Caesarean section is generally recommended for women with TS because they tend to have a small pelvis.


Currently, although still at an early stage, much research is being undertaken into the removal of functioning tissue for cryopreservation with the aim of reimplantation later on when women wish to become pregnant. The technique is considered an option mainly in women about to undergo cancer chemotherapy [12]. If this technique is to be considered in women with TS, the need for counselling as to the uncertainty of the outcome, as well as the high risk of foetal abnormalities associated with spontaneous pregnancies in women with TS, will be absolutely essential. For women with TS, pregnancy via egg donation is likely to be more appropriate (see Chapter 4).

Hormone replacement therapy

Long-term oestrogen replacement therapy in women with TS is important for gynaecological health and in the prevention of osteoporosis. It has been thought to have a role in protecting against atherosclerosis (a degenerative disease of the arteries where fatty deposits impede normal bloodflow) [2]. Women should receive combined cyclical therapy with oestrogen and progesterone, with the dose and route of administration individualised according to the woman's needs and preferences. The risk of breast cancer in women with TS is not thought to be higher than in the general population and oestrogen therapy is not thought to increase that risk [7,13].

Cardiovascular monitoring

As well as the congenital left-sided heart abnormalities seen in babies and children with TS (see Chapter 2), adult women with TS have a number of other cardiac risk factors, particularly an increased risk of aortic dissection (where the wall of the aorta splits allowing blood to pass into the aorta at pressure leading to possible rupture) and ischaemic heart disease (inadequate flow of blood in the heart). Aortic dissection can be the cause of sudden death in women with TS but if partial dissection is diagnosed then it can be treated by medication and monitoring and, in some cases, surgery. The risk of hypertension has also been shown to increase three-fold in women with TS [7]. Levels of lipids are also raised (hyperlipidaemia). Women with TS should therefore have annual cardiovascular evaluation, with blood pressure measurements at least annually and any hypertension aggressively treated with beta-blocking



drugs and/or diuretics [2]. Women found to have defects such as aortic root dilatation should have an annual echocardiogram and those women whose echocardiograms are normal should be reassessed every 5 years [2].

Although many of the cardiovascular conditions discussed above may sound alarming, it is important to understand the potential problems that girls and women with TS may have. Regular health monitoring throughout life is vital in TS so that these conditions can be detected and treated.

Diabetes

Type 2 diabetes is more common in women with TS than in the general population and also tends to develop at a younger age in women with TS [7]. Blood glucose and lipid profile should therefore be assessed annually and women who are overweight should be encouraged to lose weight. The advice of a dietician can be useful here.

Thyroid problems

The incidence of thyroid disease increases with age in women with TS [14] but the reasons for this are not known [2]. GH therapy may increase the risk.

Renal disorders

The structural abnormalities in the kidney sometimes seen in TS mean that women may be at risk of hypertension, as well as urinary tract infections (UTIs), which should be treated vigorously. Monitoring for any progressive impairment of kidney function is also important in women with TS.

Gastrointestinal disease

Women with TS have an increased risk of developing inflammatory bowel disease (IBD) (ulcerative colitis and Crohn's disease), although the reasons for this are unclear [7]. Gastrointestinal symptoms often develop at a young age – generally around the age of 16. IBD can be severe in women with TS. Any woman with TS who has unexplained diarrhoea or rectal bleeding should be referred to a gastroenterologist to rule out a diagnosis of ulcerative colitis or Crohn's disease.

Women with TS often have abnormal liver function tests and may also have an increased risk of developing chronic liver disease, perhaps as much as five times that of the general population [7]. Again, the causes of this are unclear but it does not appear to be related to excess intake of alcohol or infectious hepatitis. It could be related to use of the oral

contraceptive pill for oestrogen therapy [2].

Malignancy

The incidence of breast, ovarian and endometrial cancers in women with TS does not seem to differ from that in women without TS [7,13]. The risk of developing breast or endometrial cancer in particular may increase with the increasing use of hormone replacement therapy (HRT) in women with TS but only if oestrogen is taken unopposed, i.e. without added progesterone. Girls and women with TS generally take progesterone to induce their menstrual periods and the benefits of HRT to women with TS are enormous, far outweighing any risk. It is important however that women with TS receive natural oestrogens combined with progesterone [2].

Some women with a rare mosaic TS karyotype with some Y chromosome fragments (45,X/46,XY) may develop gonadoblastoma – a type of tumour of the ovaries that is often malignant. Gonadoblastomas can produce the male hormone androgen which will cause virilisation. For these reasons, it is recommended that non-functioning ovaries in women with this particular mosaic form of TS are surgically removed at around the age of 10 years as the risk of developing gonadoblastoma increases with age [2].

Some women with TS have also been found to be at increased risk of colon cancer [7]. Although women with TS are at increased risk of IBD, which itself increases the risk of colon cancer, bowel disease did not precede the development of colon cancer in any of the reported cases. The reason why the risk is increased is therefore unclear. It has been suggested that long-standing oestrogen deficiency may play a role [2].

Ear, nose and throat

Sound passes through the outer ear and the middle ear to the nerves of the inner ear. Obstruction to the passage of sound, usually due to infection or catarrh (glue ear) in the middle ear causes a degree of hearing loss (conductive hearing loss). This is very common in all children, but there is an increased incidence in girls with TS. This problem occurs less frequently in adult life.

From the teenage years onwards there is an increased risk of developing hearing difficulty due to deterioration of the nerve function in the inner ear (sensorineural hearing loss) [15]. This develops gradually and may not be noticed for some time. It is important that regular hearing assessment initiated in the childhood years continues through the teenage years and during adult life.



Eye problems

As many as two-thirds of women with TS have ophthalmic problems [16]. Strabismus (squint) and ptosis (drooping upper eyelids) are the most common [2]. Other problems seen in women with TS are amblyopia (failure of the retina to form sharp images) and reduced colour vision.

Skin disorders

Multiple pigmented naevi (moles) are very common in women with TS (Figure 3.2) and GH therapy may cause an increase in their occurrence [17]. The risk of these becoming malignant is not increased in TS, however [17], and naevi need only be surgically removed where they are likely to be irritated by clothing or if any malignancy is suspected.

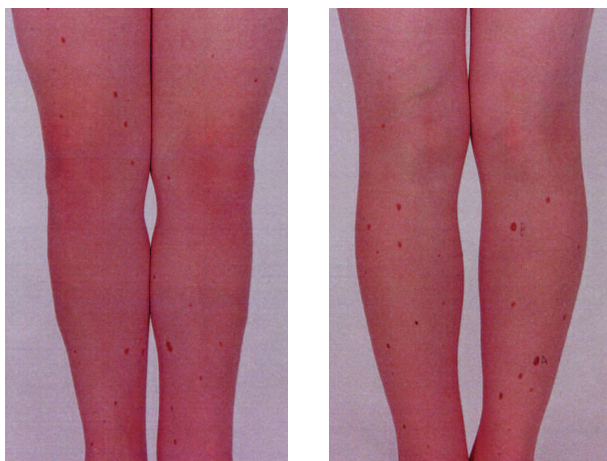


Figure 3.2: Multiple pigmented naevi (moles) are common in women with TS.

Skin conditions such as psoriasis, alopecia (hair loss) and vitiligo (abnormal skin pigmentation) also occur slightly more frequently in women with TS [3]. Long-standing oestrogen deficiency may result in fine facial wrinkling in women with TS who do not receive oestrogen therapy and, as discussed in Chapter 2, women with TS have an increased risk of keloid scarring (overgrown and thickened scars) after surgery.

Psychosocial issues

Intellectual function in girls and women with TS is generally absolutely normal and they have intelligence levels similar to those without TS. The only exception is a group who have a mosaic karyotype that includes a small ring X chromosome that appears to be associated with severe intellectual impairment [18].



In some cases, girls and women with TS have difficulty with certain non-verbal communication skills and can find it hard to interpret other people's facial expressions and body language, which may lead to misunderstandings and difficulty in maintaining friendships. They can also have poor arithmetic ability, difficulty in constructional tasks, poor sense of direction, and difficulty in learning to drive. Short-term memory and attention may also be reduced and the ability to plan and carry out multi-step tasks may be impaired [2]. These issues are discussed in more detail in Chapter 4.

Despite these potential problems, however, a significant number of women with TS complete a university degree and the majority have no difficulty in finding employment [2]. Many women with TS enter the childcare or healthcare professions such as nursing and nursery nursing.


Sometimes, problems with non-verbal communication mean that women with TS find it difficult to make friends and enter into sexual relationships. This could also be due to poor self-image on account of short stature and delayed sexual development. Women with TS therefore often benefit from the help of a clinical psychologist for counselling about their anxieties relating to short stature, infertility and sexual relationships. These issues are also discussed more fully in Chapter 4.

Prevention is better than cure

It is important to stress that not every woman with TS will have all the problems discussed in this chapter. However, prevention is always better than cure and it is wise to be informed about possible problems in order to prevent them developing.

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Personal experience of TS



A story of two women

My parents had decided to take my twin sister and me to see a paediatrician. We were both fairly short and small at the age of 14, and had not begun to develop or have regular periods. Naturally my parents were a little worried about this, hence the visits to the doctor began, to see what was wrong with us.

My family had moved from New Zealand to Bangkok, Thailand, and it was in Bangkok that my sister and I were diagnosed as having Turner syndrome, or TS. Fortunately we were put in touch with a lovely paediatrician, who ran the tests and began hormone replacement therapy. As we walked out of the hospital to catch the bus home on that memorable day, I told Mum, 'Well, that makes me extra special then, doesn't it.' She hugged me tight and through her tears she said, 'Yes, it certainly does.' It was a moment I cherish even today. My mum, sister and I walked together, lost in thought and wonder at what it was all going to mean.

My parents then began to collect and acquire all the knowledge and information they could about TS and it was only after several months that they realised how special my sister and I really were. To be a twin with TS is apparently a very rare occurrence, it never ceases to amaze me! My sister and I are very close, and as we have grown up, I have so appreciated having someone to talk to and share joys and sorrows with. I also appreciated having someone there to remind me to take my tablets!

Back in New Zealand, where we were born, we had problems early on keeping down food. The doctor in New Zealand advised Mum to feed us on potatoes and gravy, and clearly had no idea about TS. Both my sister and I grew out of the vomiting eventually, but began to suffer with ear infections in our childhood years. We both had many ear infections and my sister had grommets inserted when she was seven. I did not suffer quite so much, so did not have the operation but am now

☺
☺
☺

reaping the consequences! My sister has only one working kidney, while I have a horseshoe kidney which means I need to drink plenty! My sister wears glasses or contacts lenses but I have had the good fortune of not having any eye problems at all.

I'm working now as a registered nurse in a hospital in the UK. I began my nursing training in 1994 in New Zealand, and was informed that I would not have any trouble working as a nurse. I was told the only problem might be my short stature, and hence trouble with lifting. The training was very hard, and I found it quite difficult to do all the necessary assignments but eventually I passed, and graduated in 1997 with a Bachelor of Nursing. My sister also went to university and graduated with a Bachelor of Business Studies in New Zealand, and is now studying to gain a Masters degree in Library Studies. I enjoy my job, and find it challenging at times, but I have my own ways to get around any physical limitations. I do find the mathematical side of nursing difficult, but I get other people to check my calculations.

Our story certainly proves, yet again, that there are no limitations to what women with TS can do. I would like to think that my experiences and life will inspire those women who want to go into nursing. If I can do it, then I believe anyone can, with TS or not! Go for it! If you have something you want to do, there is absolutely nothing to stop you from achieving your dreams and heart's desires.





Living with Turner syndrome

Part 1:

Issues affecting children with Turner syndrome and their parents

Individual needs

We have seen in Chapter 2 that every girl with TS will have individual physical needs and how important it is that all potential difficulties are addressed so that she can have the best possible quality of life. Living with these physical needs can affect the way a girl copes with daily life, at home and at school, in terms of how she sees herself and regarding her relationships with family and friends. Everyone has individual strengths and weaknesses and, in this, girls with TS are no different. They just need support and encouragement to reach their full potential and live healthy and happy lives.


Spatial awareness – what does it mean?

Girls and women with TS sometimes have problems with spatial awareness. However, spatial awareness is also something that can be well developed or underdeveloped in people who do not have TS. People who lack a sense of spatial awareness have difficulty reading maps and following directions, and sometimes find it hard to catch a ball or play games that require hand-eye coordination. The box on page 46 explains the concept of spatial awareness in more detail.

How a lack of spatial awareness affects a person varies considerably. Some girls with TS may find it difficult to keep their room tidy and may react by imposing a very rigid order on their world. Others react to the same problem by rejecting order completely and living in untidy chaotic surroundings. The person may also be rigid and inflexible in their relationships, may invade other people's personal space and fail to see the point of view of others. Relationships with teachers may be particularly difficult.

Short-term memory

Some girls with TS may have problems with short-term memory and concentration and this does not necessarily improve as they get older as it might in other children. In some cases, long-term memory can be exceptional and girls can remember events from years ago but they have difficulty in recalling instructions, for example for taking medication or



how to get somewhere. These problems can persist into adult life, but girls and women with TS are very good at finding strategies to cope.

The concept of spatial awareness

The connection between points in space and time is referred to as spatial relations. Awareness of this relationship (spatial awareness) develops in infancy. During the course of a baby's development he or she learns to relate the world to the body. For example, as a child we learn to judge whether to duck our head to walk under something or how high the packet of biscuits is on the table and whether we can reach it. This understanding of the world by relating sizes and positions of things to ourselves is also true for shape, quantity and how things move. For each of these qualities we build up a system of relationships.

As a child grows and learns to talk, he or she also learns that spatial relations are fundamental to reasoning and understanding the world, and the importance of cause and effect, comparing and contrasting, how to plan and anticipate things.

An understanding of these spatial relationships is important in mathematics, spelling, punctuation, understanding time, drawing, copying and handwriting, as well as reading comprehension. It can also affect things such as tying shoelaces, staying in step with a marching band, getting the right change from a £5 note or simply finding one's way around an unfamiliar town centre.

Pre-school children

Babies and young children with TS often have difficulty feeding. These problems can appear to be behavioural but are in fact due to certain physical problems. For example, a high arched palate can cause babies to gag on food and be unable to keep anything down (see Chapter 2). These problems can be particularly stressful for parents. However, using teats designed for babies with cleft palates can help and the problem does generally improve with time. Patience and perseverance are the key and children with TS should be encouraged to try a wide variety of foods. A speech therapist can often help with feeding problems and the one-to-one attention in speech therapy sessions has the added benefit of helping improve the concentration of the child with TS.

Children with TS are often over active and sometimes immature for their age. They are generally bright and bubbly and want to play all the time. However, they also tend to need very little sleep – something that can also be stressful for parents. One of the most useful things for parents is to meet other parents via a support group.



Portage

Access to portage services can also be helpful (see Useful names and addresses). Portage is a home visiting service for families of children with special needs or a delay in their general development. Together, the portage home visitor and the parents put together a profile of the child's skills in the areas of cognitive ability, language, self-help, and motor and social skills in order to identify target skills and objectives for future teaching. As well as benefiting the child's overall development, regular visits from the portage home visitor are a source of support for the parents too, who can share their anxieties with someone who understands the practicalities of coping with a child with TS. A programme is tailored to the child's individual needs, in conjunction with other specialists such as the speech therapist. The emphasis of portage is to find out and build on what a child can do, rather than what she cannot.

Nursery education

Attending a nursery school can be very beneficial for children with TS and is excellent preparation for primary school. Nursery places should be actively pursued and children should be encouraged to attend nursery school as much as possible.


Benefits for families with a child with TS

The situation regarding benefits for families with a child with TS tends to change regularly as new schemes are introduced. Currently the Disability Living Allowance is available, and families receiving either the Working Families' Tax Credit or the Disabled Person's Tax Credit are entitled to free NHS prescriptions, free dental treatment, free sight tests and vouchers towards the cost of glasses or contact lenses, as well as refund of travel costs to hospital for NHS treatment.

Patient support groups such as the TSSS have produced guidance regarding the complex system of benefits and provide support in making applications.

At school

As well as spatial awareness problems, some of the physical problems of girls with TS (see Chapter 3) may affect how she performs at school. Problems sometimes arise when a teacher has not been given any information as to what he or she might expect from a girl with TS. Very often, simple adjustment to teaching approaches can avoid misunderstandings and help a child to fulfil her academic potential. As ever, it is important to remember that every girl with TS is an individual with particular strengths and weaknesses. They are not all the same and the approach to their education should be tailored to their specific needs.



On the positive side, girls with TS often have lively and bubbly personalities, and are kind and caring towards other children. At school they will be the first to offer help and assistance to other children and to the teacher, although this may sometimes be interpreted as attention seeking. They will always try hard and the right approach will mean a happy and contented pupil.

Keeping to a routine at home and school

Most girls with TS respond well and work best in a structured and well-ordered day. They need to know exactly what they will be doing during the day so it helps if the teacher can give a brief outline at the start of the day or lesson of the topics that will be covered and in what order. Handouts summarising the work to be covered can be very reassuring, as can visual aids such as a cardboard clock set at the time a particular event will happen. The child can then be told that the event will take place when the real clock looks the same as the cardboard clock. Keeping a diary can help, as can taking notes, both of which reinforce what has been said.

Table 4.1: The TSSS has developed 10 'top tips' for teachers of girls with Turner syndrome

- 1 Try to ensure that the child sits facing the teacher for the majority of the time
- 2 Make eye contact with the girl whenever possible
- 3 Use hand-outs rather than the blackboard whenever possible
- 4 Try to keep background noise to a minimum
- 5 When giving an instruction, start with the girl's name and then finish with her name
- 6 Give full and detailed instructions and repeat them if possible
- 7 Encourage the use of visual aids and colour to aid memory
- 8 Set realistic targets and reward with lots of praise
- 9 If a girl is not reaching academic targets then praise other virtues such as helpfulness
- 10 Listen to her concerns and treat her fairly – in this way the girl with TS will reach her full potential and become a valued member of the class

Some girls with TS may find sudden changes of routine difficult, for example if the usual teacher is ill and someone unfamiliar takes the class. The same applies at home. It can help if her clothes are always laid out in the same order. If it is games day at school, pinning a picture of someone playing games on the door the night before, along with laying out the kit, will serve as a prompt when she wakes up.

It is important not to make changes to routine just for the sake of change. The girl with TS needs to do things in her own way in her own routine, even if she does not appear to be doing things in a logical order.



The only exception is if there is a danger to the child. Making a fuss about small things will detract from getting the message across about something important. For example, it does not matter if a girl brushes her teeth in the middle of getting dressed but it does matter if she touches the saucepan of boiling water.

Self-confidence

Many girls (and women – see Part 2 of this chapter) with TS lack self-confidence and have a poor self-image. Although these are problems that can be experienced by girls and women in general, those with TS are often easily hurt or discouraged if anyone is unsympathetic. They may not be keen to try new things, particularly something that they see as too challenging. Gentle encouragement and reassurance is needed to build enough confidence to participate. Girls with TS also need to know that they have followed instructions properly and that what they are doing is correct. Continued reassurance is important.

Learning difficulties

Girls with TS may have some subtle and quite specific learning difficulties. They can be very articulate with a natural flair for English and other languages but mathematics, in particular, may cause problems. They seem to be able to cope with the problem-solving aspect of maths but have great difficulty with abstract concepts and spatial awareness (see page 46). Anything to do with trigonometry is particularly hard because they find it difficult to visualise a three-dimensional object in two-dimensional form.

These problems mean that the child may need time to learn in her own way and at her own pace. Girls with TS can think creatively and sometimes this is in an original way – going off at a tangent – but it does lead to progress.

Teachers may also find that a child may be able to express herself well verbally but finds putting her ideas into writing very hard. In addition, her ability to write may not match her ability at reading and speed and neatness of work can be a problem.

Motor skills

Girls with TS can also have problems with fine motor coordination skills and this makes fiddly tasks hard, if not impossible. Getting changed for PE lessons can be a particular problem if a girl finds it hard to manage buttons and laces. Because their hand-eye coordination is often poor and because of their short stature, games lessons themselves are also a problem for the girl with TS. PE teachers need to be aware of the particular problems and understand that persistent criticism is likely to be

counter-productive. Girls with TS need encouragement and the efforts they make should be acknowledged. They often prefer individual sports to team games and do particularly well in swimming, dancing, gymnastics and field events (Figure 4.1). Also, when it comes to picking teams, the girl with TS is frequently the last one to be chosen and the teacher should be aware of this.



Figure 4.1: Girls with TS tend to prefer individual sports to team games and do particularly well in swimming, dancing, and gymnastics. Pauline, Kylie and Alice all have TS and are medal-winning gymnasts. They are all the same age. Shona (right), age 7, shows you are never too young to start.

Following instructions

Girls with TS may find it hard to follow instructions, particularly if they are implied. It is therefore sometimes necessary to state the obvious. Very often a girl will give the impression of having understood when she has not, so it is helpful if teachers are specific, even asking the girl to repeat back any instructions. It may also be hard for girls with TS to perform tasks that require several steps to be linked, even if they can manage each of the individual steps easily. At home and school it can help if tasks are linked to objects. For example, a letter to be posted can be left by the front door with a drawing of post box as a reminder.

Social skills

It may be that a girl with TS goes through a stage where she seems to be immature for her age when she finds it hard to maintain friendships with girls of her own age. This can lead to a sense of isolation and loneliness, particularly at school break time. Bullying can be a particular problem because girls with TS are easy targets on account of their size and appearance and for this reason it helps if friends and schoolmates are educated about TS (see page 53). Because girls with TS also



sometimes have problems interpreting the behaviour of others, they often find it hard to appreciate what is happening and often pretend that everything is fine when it is not. It can be the case that girls with TS display 'attention seeking behaviour', when they are simply trying to find a way to tell their teacher or parent that they are being bullied. The good news is that, although they can be gullible and often misunderstand teasing and joking, this can improve as they get older as they learn coping strategies and are better able to interpret what is actually meant. By the time they go away to college or university they are generally much better equipped to cope with teasing.

Because they can find it hard to plan multi-step tasks, it is often the case that parents of girls with TS have a far greater hand in planning their daughter's social lives. Girls will be quite happy to play with other girls but their parents need to bring them together. This is something that can persist right through to the early teenage years.

Sex education

Parents need to be aware in advance when sex education will be covered at school – it may be earlier than some parents expect – because it is a particularly sensitive area for girls with TS. Teachers should be aware of the physiological aspects regarding puberty and fertility for girls with TS and parents should have the opportunity to view any video material in advance. The issues surrounding sexual health, contraception and pregnancy are discussed more fully in Part 2 of this chapter.

Getting organised

Self-organisation may not come naturally to girls with TS, although it is a skill that can be taught to some extent. The problem may not become apparent until a girl is older when her school work requires her to plan and manage her workload for herself. It can also be the case, however, that girls with TS feel a need (often bordering on an obsession) to know what will happen and when in order to be able to cope. This can be uncomfortable for those around them and can interfere with their education in which case the help of an educational psychologist may be helpful.

Energy and stamina

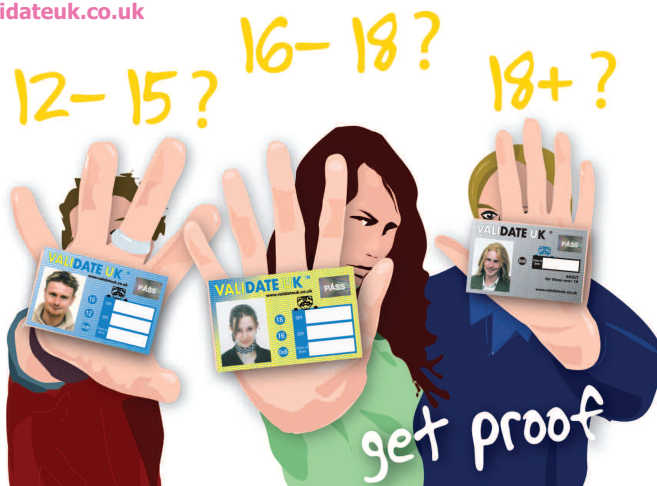
The medication that girls (and women) with TS may be taking – growth hormone, steroids such as oxandrolone or oestrogen therapy – can affect their energy levels. Older girls may have underactive thyroid glands, which can affect their stamina. It is helpful if the teacher is aware of this rather than simply thinking the child is lazy or uncooperative. Younger girls can be overactive, as well as being young for their age, and also strong willed. Parents should also recognise that girls with TS often

have a high pain threshold. This can sometimes mean that what appears to be a minor illness or discomfort could possibly be something more significant.

Perception of age

Girls with TS should not be perceived as vulnerable on account of their small size and consequently patronised or 'babied'. Some younger girls enjoy being 'babied' for a while but the novelty soon wears off. It may be a case of a vicious cycle – if a girl behaves like a baby it is hard not to protect her, and if a girl is patronised then it becomes harder for them to be mature. It is important that girls with TS are always treated as the age they are and never the age they look. They should also wear clothing appropriate to their age, i.e. dress to age not size. Girls may have additional problems because they do not look their age, for example not being able to get in to the cinema because they look younger than they are, and a special identity card such as a Validate UK card, Citizen Card or Prove It Card (18+).

www.validateuk.co.uk



Individuality

The degree to which each of these issues affects a girl with TS and her education varies greatly. Some girls may require only a slight adjustment to the curriculum and teaching to meet their needs. Others may have significant needs which require assistance from the local education authority or other outside agencies. Not all girls will require a statement/record of needs but, to ensure continuity, an Individual Education Programme (IEP) may be helpful both to the girl herself and to her teachers.



Sharing the diagnosis – who to tell and when

For parents of girls with TS, the issue of what to tell their daughters about the syndrome – and when – is important. A girl with TS will realise that she is different from her friends and the point at which parents tell her all about TS and its implications will be a decision for the parents themselves. The experience of adults with TS indicates that it is best to be honest with girls with TS from an early age. Obviously, as we have seen above, the girl's school and teachers need to be fully aware of the situation. For the girl herself, she may find it helpful to tell one or more of her friends about TS and how it affects her, or perhaps to talk about it to a group of her friends, perhaps as part of a school project. Once they know about TS, friends are likely to be very supportive and a great source of strength (see Personal Experiences of TS in Chapter 5).

Psychology of Turner syndrome

Psychological understanding of TS is currently very patchy and more research is needed. Girls with TS and their parents and adult women with TS should perhaps consider what questions they would ask the psychologist about living with the syndrome. In this way psychologists will identify those areas that require further study and prioritise their research.

Further reading

Turner Syndrome Support Society –

How to Help Your Child Survive & Succeed at School: An information booklet for teachers



Part 2:

Life with TS as an adult

What can a woman with TS achieve?

The answer to this question is the same as for any woman – anything she wants, especially if she receives support and encouragement from those close to her. We have seen how, with sympathetic support from teachers, girls with TS can do just as well their peers throughout their school years. Many girls with TS go on to college and university. Leaving home and living away from the family for the first time and making new friends can be a cause of anxiety and apprehension for any girls, even those without TS. However, it can be an especially difficult time for girls with TS because they have been used to the help and support of their parents and family in coping with their particular needs. It should also be remembered that, for any parent, it can be difficult to ‘let go’ of their daughters and allow them to lead independent lives.

Changing to a new general practitioner (GP) may cause some problems, particularly initially if the new GP does not have experience of TS, but it is important that HRT and other treatments are continued and that the girl with TS has support in this, as she will find it difficult not being at home with her parents and the GP she is familiar with.

Self image

In an age when we are surrounded by images of women with ‘perfect’ bodies in the media, girls and women with TS may have a particular problem with their body image. At a superficial level this may be related to lack of physical stature and the fact that some girls and women with TS tend to put on weight easily, or perhaps have a webbed neck or many pigmented moles. Yet a negative body image in TS could well stem from a sense of something being missing – they may have a feeling of not being complete and this can lead to all sorts of insecurities. Low self-esteem affects girls and women with TS in much the same way as it does the normal population. For example it can lead to eating disorders or obsessive behaviour.

However, women with TS should be encouraged to see themselves not as having something missing, but simply to look on TS as an integral part of themselves. In this, they should be encouraged in everything they do to build the confidence to face life with a positive attitude.

Aspects of personality

We have seen in Part 1 of this chapter that girls with TS may have particular features to their personality. They may lack confidence, partly



because of their shorter than average stature and delayed puberty which makes them different from peers. On the other hand, they may be strong willed, especially if their parents have encouraged them to be independent.

Spatial awareness problems mean that women with TS can lack hand-eye coordination (see Part 1 of this chapter for an explanation of spatial awareness), which may mean that they give the appearance of being accident prone or clumsy.

Difficulty in reading facial expressions and misreading body language can lead to misunderstandings in social situations and women with TS may find it difficult to cope with changes in daily routine, which can make them unwilling to tackle anything new. Women with TS are also sometimes unable to see alternative points of view and to appreciate that listening, hearing and understanding what is said is as important as talking. However, this is something that may well apply to many people – not just women with TS!

Career

Many girls with TS go on to college and university when they finish school, and most girls and women have no problem finding a job. Many women with TS lean towards the caring professions such as childcare (nursery nursing) or healthcare professions (nursing and midwifery). There are certainly no limits to what a woman with TS can achieve in terms of a career. However, for those who may have problems with employment help is available from Disability Service Advisers via local Job Centres.

Sex and sexuality

When considering sexuality and sexual relations, the most important aspect to stress is that girls and women with TS are physically completely female. Although most women with TS have non-functioning ovaries and a small uterus they can lead a completely normal sex life. Oestrogen replacement may assist because it helps to maintain lubrication of the vagina.

However, women with TS may be particularly vulnerable when it comes to sexual relationships for a number of reasons. They may have grown up in a very protective atmosphere because of their physical problems related to TS, or they may have difficulties in interpreting the behaviour of others. As we have seen, they can also have low self-esteem and a negative body image. It is also the case that parents sometimes find it difficult to discuss issues relating to sexuality with their daughters and

parents should not be afraid to call on professional help and advice if they need to. It should be stressed however that difficulties with relationships with the opposite sex is something shared by all women. Girls and women with TS need support and encouragement to pursue a broad range of goals in life and not simply focus on relationships.

When they are in a relationship, although they do not generally need to use contraception to prevent pregnancy, it is sensible that girls and women with TS should take precautions to avoid sexually transmitted diseases. Using a condom is as important for the girl or woman with TS as for everybody else.

Having children

For a few women with TS in the mosaic form, their ovaries remain functional long enough for them to become pregnant in the normal way. It is important that any woman with TS who is contemplating a natural pregnancy has access to genetic counselling, preferably prior to getting pregnant. She should be informed as to the risks involved, both to herself and regarding potential problems her baby may have. For example, any underlying health problems she may have (e.g. heart defects) can have increased risks during pregnancy. There is an increased chance of the baby having a chromosome abnormality such as Down's syndrome or TS itself. Antenatal tests for these chromosome abnormalities will be offered to her during the early months of pregnancy.



Figure 4.3: Susie has TS. Here, she is pictured with her husband Steve and their two adopted children Melissa and Geoffrey.

For couples where the woman is unable to become pregnant, adoption is the route that many women with TS and their partners choose to have children. However, some couples choose to undergo IVF (*in vitro* fertilisation). This allows a woman with TS to become pregnant using eggs donated by another woman, which are fertilised by the

husband/partner's sperm. The embryos are then implanted into the womb and the woman herself carries the baby. The egg can be donated anonymously via a specialist fertility clinic or perhaps by a relative or a friend.

The whole process of IVF can be quite stressful, particularly if a number of attempts at pregnancy fail, and the couple need to be committed to the process. They must be prepared for the difficulties they will encounter and for the possibility of failure. IVF treatment with egg donation is not available in most areas through the NHS. However, funding may be available for women with TS as a special case. There may be long waiting lists for donor eggs.

Emotional support

Any relationship requires commitment, honesty, openness, patience, tolerance and understanding and, in this, women with TS are no different from other women. They do sometimes need encouragement and reassurance to achieve their full potential and not use TS as an excuse for not doing something. They also need support and encouragement to attend clinics and ensure that their health is monitored regularly. In this, partners can help by travelling with them to medical appointments – sometimes this can involve long distances – and by getting involved in support group conferences and open days. These can be helpful for women with TS and their families, particularly because it enables them to meet other people with similar problems and they can benefit from sharing their experiences with each other.

Further reading

Turner Syndrome Support Society –

So you are considering a partnership! What does Turner Syndrome mean for us? A general information leaflet for partners.



Going to university – a student's story

When I was informed I had a confirmed place at University to read English Literature and History, I was naturally over the moon, but my jubilation was also tinged with a certain amount of apprehension. I would be living away from home for the first time, with a bunch of other people I didn't even know I'd get on with. I had little knowledge of cooking, and the chances of me being in catered accommodation were slim, as only one of the six main halls of residence provided meals. I would now be having to buy and cook food for myself, do my own washing and ironing, and make new friends, all at the same time as getting to grips with the university way of studying, organisation of work and making sure I knew when all the deadlines were. Even this part was going to be new and strange for me, as I knew from my brother that the approach to studying at university is nothing like how you are taught in school.

I had all these worries going around in my head as my parents drove me down at the beginning of term. As it turned out, a lot of my fears proved unfounded, and I got on reasonably well with my flatmates and there have been no major disagreements! Naturally, a lot has been new and a bit strange for me but, in general, nothing has been as daunting as I feared it might. Coming from a rather small, quiet country village, moving to the city was a big eye-opener for me. I got to go to my first nightclub. I was naturally nervous and unsure as to what it would be like, but it turned out to be great fun – even the bouncer tried to chat me up as we were waiting to go in!

When I first went to University, home seemed like a thousand miles away. In reality, however, it only takes me about an hour-and-a-half to get home on the train should I want to come home for the weekend.




Even this, however, was daunting for me at first, as I had never really travelled on my own before. But after doing it a couple of times, I am now a lot more confident about train journeys, and I even took myself off to Bristol the other weekend to visit my friend who is at university there. I have had to deal with platform changes, trains being terminated – and still managed to get where I want to!

Just like travelling, managing my finances hasn't been as bad as I thought it would be. As long as I make sure I keep track of exactly what I have in my account and what I am spending, I find that I'm not doing too badly. I haven't even got as much of an overdraft as most students seem to have.

One major concern that I did have on going to University was the fact that I had to change doctors. The university insisted that I enrolled with a local GP. I was naturally used to my GP at home and would have liked to remain with him. At first it was a bit strange, and I had to use an A-to-Z map to get to the surgery the first time. Because they did not have my notes initially, I had to explain to them what TS was myself and tell them what medication I was on. Also, I was not given a repeat prescription at first, so had to make an appointment with the doctor every time I needed more medication. Now everything is settled and things seem to be going quite well. My new doctor insists on checking my blood pressure every couple of months because of my medication, which my local GP at home didn't do.

During 'freshers' week I joined a couple of societies, one of which was a great success, the other was not. The lifesaving club I joined was brilliant and since September I have gained several more awards, two of which had to be completed in the sea. I had to travel to a seaside town



to take them in October, when you can imagine how cold the sea was! The support I have received from my instructor has been amazing. She is hopeful that by the end of the year I will have built up enough stamina to attempt my distinction award, which is the only award left for me to do. I hope next year to join some more societies as I have found this has been the best way to get out and meet people with similar interests.

So, after all this socialising how am I actually finding the course? Well, it has been mostly enjoyable, although I fell asleep during the lectures on archaeology! I have had plenty of time to get essays in on time and do any necessary research. This year I only have an average of seven-and-a-half hours each week of actual teaching. If anything, I have found this not enough, as the option is always there to put off doing what you've got to do because you feel you have plenty of time, and that opens up the possibility of forgetting about it until the deadline looms close.

In general I have found university life a challenge, but not one that is beyond me, or anyone else with TS. I have never really found my short stature a problem, and if anything, it is even less so now that I am at university. Here people seem to be much more tolerant and understanding about any height problems than people at school. I certainly found that my height has been less and less commented on as I have got older.

You shouldn't let having TS hold you back. If you think you would like to try university then you shouldn't be afraid to do so. However, you should also not be afraid to ask for help should you need it. I have been lucky so far, and have not needed it, but I do know exactly who to go to should I need any later on, and I know that the help is there.



Personal experiences of TS

Introduction

Throughout this book we have seen how girls and women are affected by the various characteristics of TS. We have also seen the impact TS can have on their lives, and also on the lives of their parents, family and friends. Experience has shown that those with TS and their families often benefit from meeting others with TS. This means they can share some of the problems and the possible solutions and also offer each other support.

Organisations such as the TSSS have a valuable role to play in helping to support those with TS, their families and friends. The TSSS (and similar organisations) provide a source of information and knowledge gained from the personal experiences of those who live daily with TS. The Society facilitates the sharing of this information with a regular newsletter, by holding open days and conferences where the girls and women and their families can meet, and by providing a contact network for those unable to attend. The TSSS is also a valuable source of information for healthcare and other professionals who have patients with TS under their care.

Personal experience stories have been included at the end of each chapter to provide readers with positive examples. This chapter includes more stories that show, first hand, what it is like to face a diagnosis of TS and the different ways people cope. Above all, these stories also show that, with sympathetic help and support, anyone with TS can lead a healthy and happy life.

Diagnosis of TS at birth – a mother's story from Arlene

Kylie, my second daughter, was born on a sunny June afternoon. I was delighted – a beautiful baby girl. Following emergency surgery after an earlier miscarriage, I was not sure if I would ever have another baby, so this was a very special time for the family. When Kylie was born I noticed that her hands and feet were very puffy and swollen and she had no toenails. She was also a very grey colour. I was told this was due to poor circulation in the womb and that she would soon 'pink up'. However, during the first hours of her life the swelling got worse, so a paediatrician was called and he did some tests.

The next day all the tests were complete and my husband and I went to see the consultant. We were given the diagnosis of Turner Syndrome, or TS. We were confused. We had never heard of TS. I can remember every word that was said. To discover that your child has a chromosomal abnormality is traumatic enough, but when it is something you have never heard of before it is even worse. Kylie was transferred to the special baby care unit because of her feeding difficulties. I was in a ward without my baby and my husband was at home. It brings tears to my eyes when I think back to the total isolation and devastation I felt.

Kylie and I were discharged a week later and she had regular check ups with the paediatrician. I began to learn more about TS and I was always asking questions. When Kylie was six weeks old the doctor gave me the address of a support group. After six weeks of worrying, it was such a relief to find that there was such a group. When I read the booklet they sent me a lot of things made more sense. I still had a lot of questions that no one seemed to be able to answer. Why me? Did I do something wrong? Could I have done something to prevent this happening? If I had other children would they have Turner syndrome?



During the first few months of Kylie's life I feel I wasted so much time feeling distressed and that I missed out on a lot of the joy of being a new Mum. The hospital was as helpful as they could be, but there never seemed to be enough time for anyone to talk to me. Because I kept asking questions, it was suggested that I see a genetic counsellor. The genetic counselling was turning point for us. During the appointment we were given a full explanation of how chromosomes work, what is thought to happen in TS and an explanation of the difference between classic and mosaic TS. The Professor who was head of the genetic department gave us the counselling and he

took time to answer all our questions and concerns. He explained that we were at no increased risk of having another baby with TS. He showed us the features that Kylie had that were typical of TS. He drew diagrams and showed us photos - it's amazing how much clearer something becomes when someone takes the time to explain the condition fully. Every point he made was quite clear and he was honest when he did not know. He came across as genuinely interested in our concerns. Because of my personal experience I feel strongly that if a child is diagnosed as having TS, or indeed any genetic condition, the parents should be referred for genetic counselling. It should make all the difference to them as a family.

I was told that the swelling in Kylie's hands and feet would improve as she grew up and it would probably go away by the age of 18 months. As the months went by I bought tights and socks to cover her feet and concentrated on the feeding problems that she had. She was such a happy baby, never sitting still for a minute and her feet never seemed to bother her. When she started walking at about a year old, there was no stopping her, she wanted to walk everywhere. Buying shoes was a nightmare. No shoes were wide enough. Her first shoes were two sizes bigger than she needed to allow for the width and she kept falling over because they were so long. Eventually the hospital gave her little white boots that were very wide, came up past her ankle and laced up the front. Not very pretty, but they did fit.

When her toenails grew they were a funny shape and very difficult to cut. They started to become ingrown and infected. We saw a chiropodist at the hospital who advised me how to cut her nails. He also arranged for a special insole to be made for her shoe, to try and stop her left ankle from turning in. Her left ankle was much worse than the right.

The swelling did not go away at 18 months. I mentioned it to the doctors, all of whom were very understanding but could offer no solution. I learned from talking to other parents that it was a common problem. I asked a physiotherapist for advice. She showed me some massage



techniques that helped a bit. When Kylie started on growth hormone treatment at four-and-a-half she began to look less puffy all over and her feet and ankles improved greatly. For a couple of years the swelling was less of a problem. It was almost completely gone on the right foot, but persisted on the left although that was much better than it had been. Her toenails continued to become infected, but she coped with it.

During the summer of 1996, following a lovely summer holiday, Kylie became very seriously ill very quickly. Eventually the doctors said she had a severe cellulitis (tissue infection) in her left leg. The doctors believe that the infection began in her toe and it had tracked up her leg. She was admitted to hospital where she stayed for three weeks on intravenous antibiotics with her foot elevated. She spent seventeen days in bed not getting up at all. Because elevation was not working, physiotherapists worked on reducing the swelling. It is one of the saddest things I have ever seen. Poor Kylie was like a puppet on a string, and all her muscles had wasted away. She was weak, could not stand or walk on her own. I admit I was absolutely terrified that she had some awful disease. With hindsight, the problem was that her lymphatic system does not work properly, so elevation would not work. What she

needed was to be up and around to get her system to drain the infection.



The swelling has not gone back down in her left ankle. Shoes continue to be a problem. Her ankle looks as though it's injured a lot of the time. The swelling becomes worse in hot climates and on long journeys, especially flights.

Out of sheer desperation I called the specialist lymphoedema clinic at a cancer hospital. We were seen within eight weeks of my GP's referral

and at last I saw a light at the end of the tunnel. The specialist nurse listened carefully to what we said and measured Kylie's feet and legs. The consultant spent time with Kylie explaining that, with support tights and a special massage technique, things should improve. The tights look just like ordinary opaque tights and encourage the fluid to circulate around the legs. The massage instructions are simple and easy to follow. We are at the early stages of trying the massage so not quite sure on how effective it will be, but the tights are great and make a big difference. So the message is that feet are important, elevation doesn't work and girls with oedema must move around as much as possible. Support tights work well and great care should be taken with footwear.


Searching for a diagnosis – Nicola's story

My daughter, Nicola, was born in June 1986 weighing 5lbs 10ozs. From the moment I tried to feed her I knew there was something wrong. For the next six months she was under the care of doctors at the local hospital who felt everything was fine. Early in 1987 she was admitted to a larger hospital for two weeks to undergo extensive tests. Nothing was found. For the following six-and-a-half years she was under the care of a consultant paediatrician at this same hospital. This consultant must have thought I was neurotic. He felt there was nothing wrong with Nicola's growth or feeding, even though she vomited and did not sleep properly for the first 16 months of her life.

Nicola was in and out of hospital with infection after infection. I visited my GP constantly for one thing or another. She was admitted to hospital for grommets (twice) and then T-tubes. Finally I managed to get her some speech therapy and, because I felt her coordination was quite bad, I took her to gymnastic classes. Nicola stayed in the same sized dress between the ages of two and five. I insisted on seeing a growth specialist and, in December 1991, saw a registrar, under a consultant endocrinologist who measured Nicola from top to toe, each arm and leg and everything else and told me there was nothing wrong.

I felt as if I was banging my head against a brick wall. Nobody would





believe me that something was wrong, neither family nor friends and now not even the doctors. I felt so alone. When I went back to the consultant paediatrician he said there was nothing he could do but I insisted on seeing a growth specialist and not a registrar. Reluctantly I was referred again.

In December 1992, pregnant with my third child, I went with Nicola to the growth specialist who did blood tests and looked at Nicola's neck, hands, body and legs. He did not once ask my height or my husband's, whereas all the other consultants we had seen seemed to concentrate on height. I sensed that he knew what was wrong with Nicola but I was too frightened to ask for fear of finding out my child had a life-threatening condition. The specialist said he would see us again in March. Three weeks after my son was born I went for the results and for the first time ever I insisted my husband came with me. Something in my heart told me I should not go alone.

We were told Nicola had Turner syndrome, or TS, which meant nothing to me until he said that 'although she has ovaries, there are no eggs'. I understood exactly what that meant and began to cry. Eventually I plucked up courage to phone the TS support organisation and the person I spoke to described the typical child with TS to me – the feeding and growth problems, always bumping into things, body and facial features and her ability to always make you laugh, etc. I couldn't believe what I was hearing. Everything slotted into place – it was Nicola exactly. All the pieces fitted and the puzzle was completed. I cried for six months and couldn't hold a conversation with anybody. Looking back it probably took me almost three years to function normally again.

When Nicola went on to growth hormone injections in May 1993 I couldn't administer them for three months. My husband always gave the injections, until one day when he could not get home until very late and Nicola pleaded for me to do it because she wanted to go to bed. I had to give her the injection but nearly fainted in the process. It was then that I realised that Nicola herself had no problem having the injections.

A couple of weeks after we were told the diagnosis, and just having a new baby brother, Shaun, Nicola asked if she would ever have children and I told her everything I knew as simply as I could.

Over these last seven years since the diagnosis of TS, I have had no problem getting follow-up care for Nicola. She attends a clinic at a growth centre in London and has been seen by a heart specialist (who says everything is fine), a hearing specialist and an orthodontist with a special interest in TS. Nicola is still receiving growth hormone injections, possibly for another year or so, and takes oestrogen daily. Our GP is now extremely helpful and understanding. Speaking to doctors and specialists has, of course, been very helpful for me to make sure Nicola is getting all the right medical help, but the best help of all comes from other Mums and women with TS themselves who have helped me to understand Nicola's condition.

Nicola is now 13 and is doing well in school. When she started secondary school there were some concerns over her immaturity and it was suggested by several teachers that she should see a psychiatrist. I too felt there were problems so after asking my GP for a referral we saw a psychiatrist for about a year-and-a-half. Discussing Nicola in private and as a family group brought back a lot of my own unresolved feelings, which have now been sorted out, and the outcome of seeing the psychiatrist is that Nicola is fine. She is happy with her condition, her family, friends and school.

I am glad I listened to my heart and not other people when Nicola was a baby. Knowing there was a problem that needed to be resolved kept me going, even though I was up against everyone and felt totally alone. Believing initially that it was simply a problem with her height but then finding out so much more, I went through feelings of 'Why me?' and 'Why Nicola?'. I felt frustrated, devastated, angry and upset but now I am relaxed and contented and very proud of Nicola. She is a hard worker and always keen to help, especially with her younger brother and baby sister, and I love her very much.





Life as a teenager with TS

I'm a 14-year-old girl with Turner syndrome, or TS. I was diagnosed at three days old and have been having growth hormone injections since I was four. As I was fortunate enough to be diagnosed so young, I have always known the effect that TS will have on me. When I was young, I only knew that TS was a growth problem, that I would have to have injections to help my growth and also that I would have to visit my consultant in London every six months.

Then when I was 10 my Mum explained to me the other side effects of TS – that I wouldn't be as well developed as my peers and that, as well as injections, I'd have to take oestrogen tablets. At first I worried that I'd stand out from the crowd and that everyone would gossip about

me. I didn't know how to explain to my friends at primary school. I'd told them that I had a growth disorder and they knew I was having injections, but I didn't know how to tell them about the other side effects, which bothered me much more than the height. I had always felt left out because all the girls in my class had their own best friends and I didn't feel I could talk to any of them about TS because I knew that gossip spread easily in our school. I didn't say anything because I thought they'd think I was strange.

When I was 11, I started secondary school and there everything changed. I made friends who I felt I could really trust and so I told my closest friends that I had TS. They were incredibly supportive. My friend was very willing to listen and I often confided in her when I felt in need of someone to talk to. It really helped to have someone my own age that I felt I could trust.

Last year in English we had to give presentations in front of our form. I chose TS as my topic because I had known the girls in my class for three years and decided that I should talk about something that meant a lot to me. It took a lot of courage but eventually I managed to explain about TS and what it was, so that the girls in my class could understand

why I was smaller, not as sporty or as developed as them.

I felt a real sense of achievement after my presentation because everyone said how brave I was. It really helped, knowing that the girls in my class finally understood why I am the way I am. It's taken a long time but, now everyone knows, I feel so much better because I'm not worried what everyone thinks of me.

I'm still far less developed than the other girls, but that doesn't bother me as much now as it used to. I now know that if I can explain TS to my peers when I'm this age, then it's going to be a lot easier explaining it to anyone in the future.


So to any girl with TS I would say that you can get over the initial loss of self-confidence. You can explain TS to your closest friends and you will feel so much better once you have. I certainly did. If any teenager with TS feels the same way as I did then just remember that you are the way you are. You can't change, and your personality is what counts, not whether you are small or not well developed. If you have personality then that is what will make you go far.

Loving someone special – a partner's story from Les

I met Lisa early in 1992 when I started voluntary work at a day centre for people with learning difficulties. Lisa was a quiet person and although we exchanged pleasantries I didn't take much notice of her until a few months later, when the centre held a fundraising event at a local club. The drinks flowed freely and at the end of the evening a crowd of us went to the local pizza parlour for something to eat. One by one people set off for home and I was left alone with Lisa and, being a gentleman, I asked if I could take her home. To my surprise, she said yes.

We talked for hours about everything – home, the job, life in general. It was easy. I had never opened up like that before and very soon she knew all there was to know about me. I can remember walking home





thinking 'She's the one for me'.

In the next couple of weeks we went out for a drink, then a meal, the cinema, and a shopping trip. We enjoyed each other's company and I quickly fell in love. It was hard keeping it quiet at work and word soon got around. The biggest problem we had to endure came from Lisa's parents. For some reason I did not understand they didn't like me. They could not see past the fact that I was 35 and Lisa was 22. Thirteen years may seem like a big age gap but it has never been an issue in our relationship. When Lisa went on holiday with her sister, I took the opportunity to go and see her Mum and Dad. Sitting round the kitchen table I tried to tell them how much I loved their daughter and I think I finally got through to them.

My first inkling that there was something that Lisa was not telling me came one day while walking on the beach. We'd been together a couple of weeks at that time and she asked how I saw my future. I told her I saw a nice house, a good job and a couple of kids. Lisa went very quiet and into what I now know as one of her sulks. I couldn't work out what I had done.

After six weeks Lisa sat me down and told me that she had Turner syndrome, or TS, and that she could not have children. I don't remember my exact response to this news. I only knew that I loved her and nothing could change that. I would stay with her through anything.

I decided to find out more about TS but the local library only came up with a contact number of a support group. However, ringing this number did put me in touch with a support group and this was a great help. They supplied me with a lot of information on TS by post, on condition that I told Lisa that I had contacted them. When this information arrived it made interesting reading but it only told half the story. It wasn't until I went to my first conference in 1993 and talked to other women with TS and their partners that everything fell into place. Lisa and I moved in together in 1994 and I was amazed at how easy it was to settle

into a new way of life. Naturally the question of marriage came up and we set a date in September 1995. It was a very special day!

Since the day Lisa told me about TS we had discussed the possibility of IVF and after the wedding we started to talk seriously about it. We were lucky that where we lived we could get the treatment on the NHS and put our names on the waiting list for funding. We waited and waited! It took about a year but we got funding for three attempts. Then we were put on the waiting list for a suitable donor and waited and waited again!

Finally, between Christmas and New Year in 1998 we got a call to say we were top of the list. We both had a number of tests done and in April the big day arrived. I was at Lisa's side when she had three embryos implanted. From the very first day, Lisa felt pregnant but sadly by the end of the week she started to bleed and we were devastated when she lost all three embryos. It took us a long time to get over it.

Six months later we went to another conference and for the first time talked openly to a few friends who knew what we had been through. It was there that we decided not to try IVF again. It was a big decision, especially for Lisa, but we agreed that we could not put ourselves through that pain again.

Early in 2000 we contacted an adoption agency and, after what seemed like an eternity of interviews, meetings and courses, we were accepted as adoptive parents. It took some doing trying to convince social workers that I was physically fit enough to bring up a child because a problem with the paperwork implied that I was at death's door because of diabetes. But at last we are able to announce the arrival of a baby, coming in at four months old and weighing 12lb 2oz. The baby has the most beautiful blue eyes and a smile that melts your heart. It has been worth the wait and the ups and downs just to wake up at 2am and see a baby looking up at you smiling away. We now feel complete, Lisa has had a huge smile on her face since the baby arrived.



Many people have supported us through some of the hard times and we would like to thank them all, especially our families and members of support group, who were there for us through the good and bad times. Knowing they were at the end of a phone was a great help to us.

Every relationship comes with its own luggage. Turner syndrome is just part of ours. I heard it said by a woman with TS at last year's conference that the partners of women with TS are special people. It's not true – we're just ordinary guys that were lucky enough to fall in love with someone special. Well I did anyway.



Finding out in later life that you have TS – Angela's story

I was born in 1946 and apparently had some swelling (oedema) of the face, hands and feet. I had an infection when I was three weeks old, which was possibly a urinary tract infection, which I now know is common in TS, and I was one of the early post-war babies to be treated with antibiotics. However, nothing was said about TS. My mother, in particular, had concerns about my lack of growth. I was not growing as fast as my elder sister and two younger brothers. Puberty did not occur for me, and my GP referred me to an endocrinologist. This was in the late 1950s and the chromosome problem of TS had just been discovered. I was admitted to hospital for a week of tests. I was frightened and confused, and nobody told me what was happening. My chromosome test was sent to London and, although the diagnosis of TS was confirmed, nobody told me I had TS. I was seen shortly afterwards by a gynaecologist but nothing further was done and again nothing was said about TS. I did not receive any medication nor was it suggested. Nothing.

After school I trained as a nursery nurse and, following that, as a midwife and became midwifery sister in a midwife-led unit in a rural district. When I was 40 I received an invitation for a cervical smear via my GP. I visited him and said I had never had one before. He checked through my notes and it was then that I learnt I had TS. He realised that this was a shock to me and from then on everyone in the practice was extremely kind and helpful to me especially the female GP.

I felt numb, angry and confused. As I had had no medication I had already begun to develop osteoporosis but it did not prevent me working at that time. Gradually, hormone treatment was introduced which, to my body, felt totally strange. I had of course learnt about the feeling experienced by other women, if not myself, during the course of my work as a midwife. Psychologically I felt that I should at least experience 'periods' and use sanitary pads, although it found it embarrassing when I went through the supermarket check out with them.

Although the osteoporosis improved when I was on hormone replacement therapy I was having mood swings which I found increasingly difficult to cope with. I also developed diabetes, which was exacerbated by stress. My gynaecologist decided to change my hormone replacement to a then relatively new medication, which meant I no longer had periods. I have been taking this ever since and my osteoporosis has been held in check. I also take calcium and a drug called a bisphosphonate.

On medical grounds, because of the osteoporosis and diabetes, I had to take early retirement at the age of 44.

I was extremely lucky to meet a clinical psychologist while I was practising as a midwife to whom I was able to talk. I had a lot of feelings to express, particularly anger. My situation may well have turned out very differently without having a professional to talk to. The hormone replacement therapy has helped me to feel a woman.



Although I have not been able to have children myself, at least as a midwifery sister I was able to bring new life into the world and help women and their partners at such an important time in their lives. It has been a great loss to me not to be able to continue in my profession. I once delivered a baby who was diagnosed with TS soon after birth and years later met up with her and her mother again. This girl had growth hormone treatment and went on to hormone replacement therapy as a teenager. Compared to 30-40 years ago when there was little or no information and no treatment given, this is the big difference now – early diagnosis, correct treatment and full knowledge. These improvements help give me some satisfaction, and the fact that there are now support groups to help parents, girls and women with TS. A vital part of my life now is meeting up with other women who have TS.



Postscript

Over the years I have been involved with the Turner Syndrome Support Society I have met many wonderful people with TS and their families and made many friends; for them and for those yet to be diagnosed I hope that TS will become much better understood by all who read this book.

Lynne Morris

Adviser, TSSS



Turner Syndrome Support Society Committee Members



Glossary

Alphafetoprotein (AFP) - protein synthesised in the foetal liver and intestine found in the foetal blood and uterine fluid. Raised levels indicate a possible chromosome abnormality

Alopecia - hair loss

Amblyopia - failure of the retina to form sharp images

Amenorrhoea - lack of monthly menstrual periods

Amniocentesis - taking a sample of the fluid that surrounds a baby in the womb so that the genetic material of the baby can be analysed

Aneurysm - balloon-like swelling in a blood vessel

Anorexia nervosa - serious eating disorder where the sufferer drastically cuts food intake because they believe they are too fat when they are really very thin

Aorta - the main blood vessel into which the heart pumps blood

Aortic dilation - thinning of the wall of the aorta

Aortic dissection - splitting of the wall of the aorta, allowing blood to enter under pressure and causing a ballooning of the aorta, which may ultimately rupture

Atherosclerosis - a degenerative disease of the arteries where fatty deposits impede normal bloodflow

Bacterial endocarditis - inflammation of the inner lining of the heart and valves due to infection

Bicuspid aortic valve - where the aortic valve has two cusps, or projections

Bulimia nervosa - serious eating disorder where the sufferer regularly binges on food and then deliberately induces vomiting

Cardiologist - doctor specialising in conditions affecting the heart

Cellulitis - spreading inflammation of the skin and subcutaneous tissues caused by infection

Chiropodist - specialist in the care of feet (see also Podiatrist)

Cholesteatoma - a mass of tumour-like cells shed by the outer layer of an infected eardrum that can invade the middle ear through a perforation in the drum and damage the inner ear

Chorionic villus sampling - taking a sample of the membrane that surrounds the baby in the womb so that the genetic material of the baby can be analysed

Chromosome - part of the genetic material present in all human cells inherited from the parents

Coarctation of the aorta - narrowing of the aorta just beyond the point where the arteries branch off for the head and arms

Cubitus valgus - increased carrying angle of the arms because elbows cannot be fully straightened

Cystic hygroma - fluid-filled swelling around the nape of the neck of the foetus in TS

Diabetes (mellitus) - disease in which the supply of insulin is insufficient for the body's needs

DNA - deoxyribonucleic acid which makes up the genetic material of the body

Dysmorphism - severe abnormality of development caused by influences at an early stage of foetal growth

Endocrinologist - specialist in glands/hormones

ENT specialist - doctor specialising in conditions affecting the ear nose and throat

Epicanthic fold - vertical fold of skin from the upper eyelids that covers the inner corner of the eye (sometimes called epicanthal fold)

FSH - follicle stimulating hormone. Hormone produced by the pituitary gland which stimulates the ovaries to produce eggs

Geneticist - specialist concerned with the structure, location, abnormalities and effects of the genes and how they cause disease

Genetic counselling - advice given to parents and prospective parents about the likelihood of a child suffering from a genetic disorder and the consequences of that disorder

Grommets - tiny plastic tubes inserted through the eardrum to help drain the middle ear

Growth hormone (GH) - the hormone that stimulates growth

Gynaecologist - specialist in diseases specific to women, particularly regarding fertility, menstrual disorders, contraception and childbearing

Horseshoe kidney - congenital abnormality where the two kidneys are joined together, either at the upper or lower end, to form a single organ

Hydronephrosis - ballooning of the urine collecting system in the kidney resulting from obstructed outflow of urine

Hypercholesterolaemia - abnormally high levels of cholesterol in the blood

Hyperconvex - abnormally rounded in shape

Hyperlipidaemia - abnormally high levels of lipids (fats), including cholesterol, in the blood

Hypertension - raised blood pressure

Hypothyroidism - underactivity of the thyroid gland

Hyperthyroidism - overactivity of the thyroid gland

IBD - inflammatory bowel disease

IEP - individual education plan

Insulin - hormone that causes cells in the muscles and liver to remove sugar from the blood

Ischaemic heart disease - inadequate flow of blood in the muscles of the heart

Karyotype - the individual chromosome complement of a person

Keloid scarring - overgrown, thickened raised scarring, which sometimes looks inflamed and can be unsightly

Left ventricular hyperplasia (or hypertrophy) - enlargement of the left ventricle of the heart

Lymphoedema - persistent swelling of tissues caused by absence of drainage from the lymphatic channels

Mastoiditis - a potentially dangerous inflammation of the honeycomb-like air cells behind the lower part of the ear

Menopause - the time of life when a woman's ovaries stop producing eggs and she ceases to have periods (monthly bleeds)

Menstruation/menarche - monthly bleeding (periods) when parts of the lining of the womb are discharged if a woman is not pregnant

Micrognathia - a small jawbone

Mixed gonadal dysgenesis - condition in which the female sex organs (ovaries) do not develop properly and may contain some tissue normally found in the male sex organs (testicles)

Mosaicism - occurs when cells do not all contain identical chromosomes

Myopia - short-sightedness

Nuchal folds - folds of skin in the nape of the neck

Nuchal translucency - abnormal translucency around the neck in the foetus in TS

Obstetrician - doctor who specialises in pregnancy and childbirth

Oedema - excessive accumulation of fluid causing swelling, in the case of TS in the hands and feet

Oestrogen - hormone involved in female sexual development and functioning of the female reproductive system

Ophthalmologist - specialist in disorders of the eye

Orthodontist - specialist in correcting the position of the teeth

Orthotist - specialist in using orthopaedic supports to align, prevent or correct deformities or improve function, particularly of the feet

Osteopenia - early bone loss and decreased bone density, which if not treated progresses to osteoporosis

Osteoporosis - thinning of the bones. Occurs when the natural process of bone turnover becomes imbalanced so that more bone is resorbed than is formed

Otitis media - infection of the middle ear

Ovaries - part of the female reproductive system where eggs develop

Oxandrolone - anabolic steroid, sometimes used in growth promotion

Paediatrician - specialist in the treatment of children

Palate - roof of the mouth

PCHR - Personal Child Health Record

Pigmented naevi - moles

Podiatrist - specialist in the care of the foot (see also Chiropodist)

Progesterone - hormone which acts together with oestrogen during the menstrual cycle and prepares the lining of the womb to receive a fertilised egg

Psoriasis - a common skin condition with scaly thickened patches of reddened skin

Psychiatrist - specialist concerned with the management of mental illness

Psychologist - specialist in the scientific study of behaviour, and concerned with how memory, learning, personality, perceptions and emotions relate to behaviour

Pterygium colli - extra folds of skin which stretch from the neck to the shoulders (web neck)

Ptosis - drooping eyelid(s)

Puberty - time when the body begins to develop sexually, e.g. development of the breasts

Scoliosis - deformity of the spine

SEN - special educational needs

Sex chromosome - part of the genetic material that determines whether a person is male or female. Males have one X and one Y chromosome, females have two X chromosomes

Spontaneous puberty - when puberty occurs naturally

Strabismus - squint

Subcutaneous - applied under the skin

Syndrome - used to describe a disorder when a combination of symptoms occur together to form a distinct clinical entity

Thyroid gland - gland situated in the neck across the upper part of the windpipe that secretes hormones to control the metabolism

Thyroid hormone - hormone that helps to control metabolism – the chemical processes that occur in the body

Transcutaneous - entering through the skin

TSH - thyroid stimulating hormone

Ultrasound - method of imaging the body, based on the reflectivity of sound, which uses very high frequency (ultrasonic) sound wavelengths

Uterus - the womb

Vitiligo - skin disorder where patches of skin, especially on the face and backs of the hands, lack any pigmentation

Note: The explanations given in this Glossary are intended as a guide to understanding and should not be regarded as definitive definitions

Useful names and addresses

Turner Syndrome Support Society (UK)

12 Simpson Court
11 South Ave
Clydebank Business Park
Clydebank G81 2NR
Helpline: 0300 111 7520
Tel: 0141 952 8006
Fax: 0141 952 8025
Email: turner.syndrome@tss.org.uk
Website: www.tss.org.uk

Contact-a-Family

209-211 City Road
London EC1V 1JN
Tel: 020 7608 8700
Fax: 020 7608 8701
Email: info@cafamily.org.uk
Website: www.cafamily.org.uk
Helpline: 0808 808 3555
Text phone: 0808 808 3556
Free phone for parents & families (Mon-Fri 10am-4pm & Mon 5.30-7.30pm)

Contact-a-Family Scotland

Craigmillar Social Enterprise & Arts Centre
11/9 Harewood Road
Edinburgh EH16 4NT
Tel: 0131 659 2930
Email: scotland.office@cafamily.org.uk
Website: www.cafamily.org.uk

Genetic Interest Group

Unit 4D
Leroy House
436 Essex Road
London
N1 3QP
Tel: 020 7704 3141
Fax: 020 7359 1447
Email: mail@gig.org.uk
Website: www.gig.org.uk

Antenatal Results & Choices (ARC)

73 Charlotte Street
London W1T 4PN
Tel: 020 7631 0280 (Admin)
Helpline: 020 7631 0285
Fax: 020 7631 0280
Email: info@arc-uk.org
Website: www.arc-org.uk

National Autistic Society

393 City Road
London EC1V 1NG
Tel: 020 7833 2299
Fax: 020 7833 9666
Helpline: 0845 070 4004
Email: nas@nas.org.uk
Website: www.autism.org.uk

Diabetes UK

Central Office
10 Parkway
London NW1 7AA
Tel: 020 7424 1000
Fax: 020 7424 1001
Website: www.diabetes.org.uk

Dyspraxia Foundation

8 West Alley
Hitchin
Hertfordshire SG5 1EG
Tel: 01462 454986
Fax: 01462 455052
Website: www.dyspraxiafoundation.org.uk

National Portage Association

Website: www.portage.org.uk

Pituitary Foundation

PO Box 1944

Bristol BS99 2UB

Administration Line: 0845 450 0376

Support and Information Helpline: 0845 450 0375

Endocrine Nurse Helpline: 0845 450 0377

(available Monday evenings from 5:30pm to 9:30pm
and Thursday mornings from 9:00am to 1:00pm)

Fax: 0117 933 0910

Email: helpline@pituitary.org.uk

Website: www.pituitary.org.uk

Independent Panel for Special Educational Advice (IPSEA)

6 Carlow Mews

Woodbridge

Suffolk IP12 1EA

Advice line: 0800 0184016

Scotland: 0131 454 0096

Northern Ireland: 02890 704606

Website: www.ipsea.org.uk

Benefits Enquiry Line

for people with disabilities

0800 882200

British Thyroid Foundation

PO Box 97

Clifford

Wetherby

West Yorkshire LS23 6XD

Tel: 0113 392 4600

Website: www.btf-thyroid.org

Premature Menopause Support Group (Daisy Network)

The Daisy Network

PO Box 183

Rosendale

BB4 6WZ

Website: www.daisynetwork.org.uk

BEAT

103 Prince of Wales Road
Norwich NR1 1DW
Tel: 0870 770 3256 (Admin)
Adult helpline: 0845 634 1414
Youthline: 0845 634 7650
Email: info@b-eat.co.uk
Website: www.b-eat.co.uk

Sexual Health Direct

50 Featherstone Street
London EC1Y 8QU
Helpline: 0845 122 8690
Website: www.fpa.org.uk

Child Growth Foundation

Registered Office

2 Mayfield Avenue
Chiswick
London W4 1PW
Tel: 020 8995 0257
Email: info@childgrowthfoundation.org
Website: www.heightmatters.org.uk

Membership Enquiries

21 Malvern Drive
Sutton Coldfield B76 1PZ
Tel: 0121 313 3525
Email: jenny.child@childgrowthfoundation.org

British Agencies for Adoption and Fostering (BAAF)

information on adoption and fostering

Saffron House
6-10 Kirby Street
London EC1N 8TS
Tel: 020 7421 2600
Fax: 020 7421 2601
Email: mail@baaf.org.uk
Website: www.baaf.org.uk

Adoption UK

information for adoptive families, local support and self-help groups

46 The Green
South Bar St
Banbury
Oxon OX16 9AB
Tel: 01295 752240
Fax: 01295 752241
Website: www.adoptionuk.org

Infertility Network UK

infertility education and counselling

Charter House
43 St Leonard's Road
Bexhill-on-Sea
East Sussex TN40 1JA
Tel: 01424 732361
Fax: 01424 731858
Website: www.infertilitynetwork.uk.com

The National Osteoporosis Society

Camerton
Bath BA2 0PJ
Tel: 01761 471771
Fax: 01761 471104
Helpline: 0845 4500 230
Website: www.nos.org.uk

The Human Fertilisation and Embryology Authority

information on IVF, egg donation and fertility clinics

21 Bloomsbury Street
London WC1B 3HF
Tel: 020 7291 8200
Fax: 020 7291 8201
Email: admin@hfea.gov.uk
www.hfea.gov.uk

BDF Newlife

BDF Centre
Hemlock Business Park
Hemlock Way
Cannock
Staffordshire WS11 7GF
Tel: 01543 468888
Website: www.bdfnewlife.co.uk

Restricted Growth Association

PO Box 4008
Yeovil BA20 9AW
Tel: 01935 841364
Email: office@restrictedgrowth.co.uk
Website: www.restrictedgrowth.co.uk

Useful websites

British Society for Paediatric Endocrinology and Diabetes (BSPED)

www.bsped.org.uk

Harlow Printing Limited

www.harlowprinting.co.uk

Health For All Children

www.healthforallchildren.co.uk

