

# Diagnosis of Turner Syndrome at Birth

## The Bungle Pack

kindly supported by **Tracey and Lainey Walters**

———— in memory of ————

*Corporal James Walters*

affectionately known as '**Bungle**'



Turner Syndrome Support Society

**The Turner Syndrome Support Society (TSSS) is proud and very grateful to have produced 'The Bungle Pack', in memory of James Walters, affectionately known as 'Bungle'.**

*“James was extremely passionate about TS and made sure that we were as prepared as we could be for all aspects of Lainey's condition. He would be immensely proud and completely overwhelmed at how far his baby girl has come, and to know that the money raised in his memory is making a difference, by educating parents, saving TS girls' lives and supporting individuals with TS, their families and healthcare professionals.”*

**Tracey Walters**

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**This booklet is written for healthcare professionals involved in the care of the child diagnosed at birth with Turner syndrome (TS).**

**The aim of the booklet is not to provide comprehensive medical guidance, but to include some helpful background information that will support discussions between healthcare professionals and the parents about their baby with TS.**

## Overview of Turner Syndrome

Turner syndrome (TS) is a relatively common chromosome abnormality affecting only females. In classical TS the girl has only one X chromosome in her cells (45X karyotype). Alternatively, the girl has some cells in her body with two X chromosomes, but other cells have only one, and this is called mosaicism.

There is wide variability in the severity of clinical features between individuals (Donaldson et al 2006). Although textbooks often refer to the marked phenotype (physical characteristic) that is evident at birth, many girls with TS are diagnosed after the age of puberty, or even as adults, which reflects the increasingly recognised mild phenotype.

Although TS is a lifelong condition, most individuals live long and healthy lives, but some may have several associated long-term conditions which require routine medical screening, regular monitoring, and follow-up.

### Cause of TS

The exact cause of TS is unknown. TS appears to occur as a random event during cell division, early in the development of the fetus. It is not usually inherited and the age of the mother does not increase the risk of TS. There are no clearly established risk factors and the recurrence of TS in subsequent pregnancies is rare.

### Incidence and prevalence

TS occurs in approximately 1 in every 2000 to 1 in every 2500 live female births (Nielsen et al 1991). The true prevalence of TS is difficult to ascertain for many reasons; for example some individuals have a mild phenotype and may remain undiagnosed. TS occurs with more or less the same prevalence in all ethnic groups and in different countries.

in memory of *Corporal James Walters*

## Physical features

Girls with TS may have distinctive features, some of which may be apparent at birth and some may appear as the child develops. Not all girls will have every problem listed:

- Short stature
- Webbed or broad neck
- Swelling of the hands and feet
- High arched palate with crowded teeth
- Low posterior hairline
- Deep forehead
- Atypical bone development affecting e.g. hands and elbows
- Red-green colour blindness
- Broad chest with widely spaced nipples
- Low set ears
- Abnormal finger and toe nails
- Short 4th metatarsal or metacarpal
- Wide carrying angle of the arms making it difficult to straighten the elbow
- Larger number of naevi compared to other members of the family
- Ptosis, strabismus, amblyopia, and cataracts are more common

## Diagnosis at birth

One of the most common early indicators of TS is lymphoedema at birth which affects the hands and feet. TS can be confirmed by the chromosomal analysis of peripheral blood or other tissue samples.

Following a positive diagnosis a comprehensive screening evaluation is required, involving a cardiac examination with echocardiogram or magnetic resonance imaging (MRI). This will identify malformations in the cardiovascular system including coarctation or narrowing of the aorta, and a bicuspid aortic valve, where the valve has only two cusps rather than three. Other medical tests include renal ultrasound, hypertension screening, clinical and laboratory evaluation of thyroid and liver function, as well as audiology testing. Early diagnosis of any problems will facilitate the best possible care of the child.

## Screening evaluation at birth

- Examine the child's hips for dysplasia
- Test hearing
- Consult with cardiologist who has experience in paediatric cardiology and TS
- Check blood pressure and peripheral pulses during each physical examination
- Compare arm and leg systolic pressure to evaluate for possible coarctation
- Renal ultrasound

## Communicating the diagnosis

Communicating a diagnosis can be a difficult process and involves a number of important skills and considerations. The challenges involved in communicating medical information cannot be successfully addressed with universal, 'one-size-fits-all' recommendations and should not be a 'one-off' event. Communication of the initial diagnosis will be a single point in the clinician/parent relationship that will continue through to treatment of the child and the overall management of the condition. The family will need careful education, counselling, and support to understand TS and its medical implications.

A number of factors will influence the success or otherwise of communicating the diagnosis including the personality of the clinician and parents. Healthcare professionals should take into consideration that different people react differently to the diagnosis and they should tailor their communication accordingly. In addition it is important to take note of the people involved and their ability to understand medical information. All information should be presented in an easy to understand manner.

As soon as the diagnosis is confirmed, the clinician should speak to the parents in person. A quiet, private room is required to convey the diagnosis with time to communicate at the parents' pace and with no interruptions. The language should be as simple as possible, medical jargon avoided, and the uncertainty faced by the parents acknowledged. The parents need to be fully informed about the prognosis, complications, and quality of life of individuals affected with TS, as well as of recent advances in management. The parents need to know that short stature and infertility are likely, intellectual disability is unlikely, congenital anomalies may be present, and some girls may have learning difficulties. The treatments and available interventions should be discussed, such as growth-enhancing therapy and hormone replacement, but with medical supervision, psychosocial counselling, and support, girls with TS may lead healthy, productive lives (Frias et al 2003).

A plan of action should be discussed to reassure parents that their child will receive the best possible care. The parents should be given time to ask any questions that they may have, and further support provided by other healthcare professionals if required. Printed materials about the condition should be available and information about the Turner Syndrome Support Society (TSSS) should be provided.

## Specialist medical care

TS may present the girl/woman, her family and the healthcare team with a wide range of genetic, developmental, endocrine, cardiovascular, psychosocial and reproductive issues.

Children with TS should be referred to a hospital that has expertise in TS and an interdisciplinary approach to the management of the condition. The paediatric endocrinologist will manage an overall care plan with regular assessments to monitor growth, puberty development, and referrals to the relevant medical speciality for treatment as necessary. Treatment will focus on problems that occur; some features of TS will not require any intervention and others may be improved or treated with surgery, medicine or other relevant therapies.

More than 95% of adult women with TS have short stature (Saenger et al 2001) and over 90% have signs of ovarian failure (Bondy 2007). Clinics for adults with TS are established in the UK where they monitor the associated long-term conditions including cardiovascular disease, hypertension, obesity, type 2 diabetes, hypothyroidism, sensorineural hearing loss, and osteoporosis.

The medical care of girls with TS requires ongoing assessment and periodic review of specific problems at appropriate ages. The TSSS provides a Paediatric and Adult Health Checklist which lists all the tests required for paediatric and adult TS management. It is a useful record for people with TS and their parents and is designed to slot into the patient record and act as a 'prompt' to ensure that the individual is treated according to her individual needs.

*“We spoke to the TSSS and it was extremely reassuring and honest. Through the Society, we have since met many girls and ladies with TS, and their families, and it's fantastic to see how well many girls with TS do. We are confident that, with the right support, Charlotte has a bright future ahead of her.”* Catherine, Charlotte's mother

## Family stories

The TSSS has so many stories about families with amazing daughters that have TS which are available on its website [www.tss.org.uk](http://www.tss.org.uk). Here is Charlotte's story...

### Charlotte's story

Charlotte was born five days early after a very quick labour. Before leaving the hospital the doctor expressed concern about Charlotte's swollen hands and feet, reassured her parents but asked them to return to the out-patient clinic two weeks later. Despite the reassurance her parents were concerned and 'googled' puffy feet at birth. The results suggested Turner syndrome (TS) and Charlotte's parents were both alarmed and distressed.

When they returned to the hospital her parents were informed that Charlotte might have TS. A blood test was done and six long weeks later the diagnosis was confirmed. The paediatrician discussed the condition, potential issues, and explained the next steps. Over the next few weeks Charlotte had ultrasound scans on her kidneys, head, and an echocardiogram which all revealed no concerns. Initially the paediatrician continued to manage Charlotte's care but she was then referred to the endocrinology department at her local children's hospital.

Charlotte is now a happy and lively 18-month-old. She is small for her age, but so far, any issues related to her TS have been minimal. She has very small, upturned toenails and her feet are still quite swollen. She was a slightly fussy eater, but now eats a variety of finger foods quite happily. She walked and talked a little later than her brother, but not unusually so. She's not been a great sleeper, but then many babies aren't!



## References

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## Further information

### **Turner Syndrome Support Society**

[www.tss.org.uk](http://www.tss.org.uk)

### **Turner Syndrome International Group**

[www.tsint.org](http://www.tsint.org)

### **Turner Syndrome Support Society UK**

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