



Turner Syndrome Support Society

Did you know almost **80% of girls** were first seen by **ENT specialists** before they were diagnosed with **Turner Syndrome?**

Your awareness could make a huge difference to a family!

Can you help the Turner Syndrome Support Society [UK]?

We are the national organisation supporting those people who have Turner Syndrome [TS] by providing accurate up to date information on TS and living with TS. One of our aims is to improve awareness and understanding about TS. Another is to improve the age of diagnosis and give girls the opportunity to benefit from the treatment available to them following diagnosis. This is where we need your help.

Short Stature

Repeat grommets
in short girls

Tonsils/Adenoids/
Sleep Apnoea

High arched palate/
small jaw



Recurrent Otitis with
MediaLow set ears

Retraction pockets
or cholesteatoma
in short girls

Excessive saliva
overcrowded teeth

Short thick neck
described as
webbed neck

Turner syndrome what is it?

Turner syndrome [TS] is a common chromosome abnormality affecting 1 in 2000 females. It is caused by the complete or partial deletion of one of the X chromosomes in some or all of the cells of the body. There are a number of physical features and characteristics associated with TS but it is rare that they will all appear in one child. Short stature, recurrent otitis media and delayed puberty are particularly common. Providing diagnosis is made early enough there is the possibility of growth hormone treatment for short stature or timely induction of puberty with oestrogen. Unfortunately, the diagnosis of TS is often delayed occurring in adolescence in 50%. While the majority of girls with TS lead healthy, happy and normal lives, the journey is more difficult for those with late diagnosis particularly if it had been missed in earlier visits to a doctor. From speaking to families following a diagnosis of TS almost 80% were first seen by an ENT specialist. If you are aware of TS you can make a real difference to the long-term quality of life of the child! A referral to a Paediatric Endocrinologist can confirm or exclude TS.

Disadvantages of delayed diagnosis of TS

Girls with TS have specific difficulties which, if recognised early, can be compensated for by simple measures. Some of the educational problems relating to TS may be the result of unrecognised hearing impairment and early detection would enable simple measures such as placement in class. The early introduction of growth hormone leads to near normalisation of final height for girls with TS and timely introduction of oestrogen is important for bone development. In addition there may be other benefits such as reduced incidence of ear infection events (Lanes R., Gunczler P. (2000).

Recognising TS in an ENT clinic

ENT visits are an important opportunity to make an early diagnosis of TS for two reasons. Firstly, second to short stature ENT problems are the most common feature of TS during childhood. Secondly, recurrent and severe otitis media occur in a young age group who can still benefit from earlier administration of growth hormone and oestrogen. Even in those patients who do not have obvious phenotypic features (See below), close observation can suggest a diagnosis of TS.

Many girls subsequently diagnosed with TS were first seen in an ENT clinic. These missed opportunities arise because the TS phenotype might not be immediately apparent. Below is a list of features which will help to identify girls who are at risk. With regard to short stature, simply ask about her height ranking in the classroom! We suggest this quick checklist be performed for every girl attending with severe or recurrent otitis media and that if any of these features are evident then a blood sample for karyotype should be requested.

Specific Associated Features in Turner Syndrome

- short stature
- nail dysplasia
- high arched palate
- short fourth metacarpal
- low set ears
- low posterior hair line
- webbed neck
- strabismus

The presence of key features of TS should be considered in all young girls with otitis media. ENT surgeons should always consider possible underlying causes for recurrent OM (especially if refractory) and should refer to a paediatrician for a full assessment to exclude TS as well as the other conditions

Recommended reading “Diagnosing Turner Syndrome” available on www.tss.org.uk

Cholesteatoma has a high prevalence in Turner syndrome, DBN Lim1, EJ Gault1, H Kubba2, MSC Morrissey2, DM Wynne2, MDC Donaldson - Acta Pædiatrica ISSN 0803-5253

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