

Care of a Child with Turner Syndrome

1. General Presentation

Turner syndrome is one of the most common chromosomal abnormalities and an important cause of short stature and primary amenorrhea in young females. *Turner syndrome should be considered in any girl with short stature.* Turner syndrome refers to a collection of X chromosomal disorders resulting in a deletion or silencing of a particular set of genes on the short arm of the X chromosome. The most common type of Turner syndrome is the classic syndrome, representing 50% of all Turner syndrome cases. The classic syndrome involves the loss of one X chromosome, yielding a 45XO karyotype. Other forms of the syndrome include mosaic chromosomal complements of 45XO/46XX, X chromosome deletions, and ring X chromosomes.

2. Clinical Presentations

Clinical presentation can be variable and subtle especially for girls with the mosaic form of Turner syndrome. Characteristic clinical presentation includes (but is not limited to):

- Short stature despite normal growth hormone levels
- Dysmorphic features: webbed neck, short fourth metacarpal, nail dysplasia, high palate
- Sensorineural hearing loss
- Congenital lymphedema of the hands and feet
- Primary amenorrhea or early ovarian failure
- Ocular abnormalities: amblyopia, strabismus, ptosis, hypertelorism, epicanthus
- Renal abnormalities: horseshoe kidney, abnormal vascular supply
- Cardiovascular abnormalities: coarctation, valvular disease
- Cognitive, learning, and emotional disorders

3. Diagnosis of Turner Syndrome in a Child

A suspicion of Turner syndrome should be raised at birth from classic abnormalities such as lymphedema. Later on in childhood and adolescence, a suspicion of Turner syndrome should be raised when there is a failure to grow, failure to enter or complete puberty, or secondary amenorrhea.

A diagnosis of Turner syndrome can be arrived at through karyotype analysis. A DNA sample of peripheral blood lymphocyte showing a karyotype of 45XO is diagnostic. In the event that a patient presents with clinical features of Turner syndrome but a normal peripheral blood lymphocyte karyotype (46XX), a skin fibroblast tissue sample should be taken and karyotyped. Turner syndrome is the diagnosis if the skin fibroblast tissue sample shows 45XO. Also note that in the case of a patient with mosaicism resulting in Turner syndrome, the mosaicism distribution may also be dependent on the type of tissue.

4. Management of a Child with Turner Syndrome

a) Treat short stature

Treating short stature is important to maximize a girl's growth potential. Subcutaneous growth hormone should be administered when a girl with Turner syndrome falls below the 5th percentile for age. This often occurs between 2 and 5 years of age. Between ages 9 and 12, girls with extreme short stature require daily injections of growth hormone and an anabolic steroid (oxandrolone) before epiphyseal fusion to maximize final height. It should be noted that a low dose of oxandrolone should be used. At high doses of oxandrolone, unwanted effects of virilisation and early skeletal maturation may occur. The combination therapy should be continued until there is no remaining growth potential.

b) Treat primary amenorrhea

After the age of 12, estrogen therapy should be administered to induce normal pubertal development and maintain bone mineral density. Doses should begin at 1/10th the dosage of adults and be gradually increased within the next two to four years. Remember to discontinue oxandrolone when estrogen therapy is initiated.

After growth has stopped, hormone replacement therapy with both estrogen and progestin should be used to simulate normal menses. Note that cyclic progestin therapy is important to prevent endometrial hyperplasia caused by unopposed estrogen.

c) Treat lymphedema

Edema due to lymphatic obstruction is common in Turner syndrome. It should be managed with support stockings. Vascular surgery is rarely indicated. Surgery for girls with Turner syndrome should generally be avoided when possible since they have an increased predisposition to keloid formation.

d) Investigate and treat cardiovascular anomalies

An echocardiography or MRI is necessary at the time of diagnosis to screen for cardiovascular anomalies. Turner syndrome is often associated with coarctation of the aorta and bicuspid aortic valves. Repeat imaging is recommended every 5 to 10 years. Routine blood pressure in all extremities is also important to screen for hypertension. Cardiovascular anomalies should be treated as indicated. For example, coarctation of the aorta should be corrected surgically.

e) Assess for hypothyroidism

Thyroid function should be assessed annually. There is an increased risk of hypothyroidism in children with Turner syndrome.

f) Assess for metabolic syndrome

Fasting blood glucose, HbA1C, and lipids should be done annually beginning during adolescence. Girls with Turner syndrome have an increased risk of type 2 diabetes mellitus, obesity, dyslipidemia, and hypertension.

g) Assess for ocular abnormalities

Routine ophthalmologic evaluation is indicated. Ocular abnormalities are more common in this population. Strabismus can occur in girls with Turner syndrome and is most treatable early in life. Ptosis, amblyopia, cataracts, red-green color blindness and other ocular abnormalities can also occur.

h) Assess for renal anomalies

A renal ultrasound should be performed at the time of diagnosis. If there is any structural abnormality, children should be monitored yearly for UTI.

i) Assess and treat cognitive function and learning disabilities

Girls with Turner syndrome generally have normal intelligence. However, they may have difficulties with social cognition, nonverbal problem-solving tasks, and visual-spatial organization. There is also a higher incidence of attention-deficit disorder. Sometimes, self-esteem can also be poor. Preliminary studies have shown that estrogen and androgen replacement therapies can have some benefit on cognitive function in these girls. Family supports and social supports are important.

The list above includes the key management concerns of caring for a child with Turner syndrome. Due to the variability of Turner syndrome's clinical presentation, there are a variety of other medical conditions associated with Turner syndrome that may occur. These conditions should be treated as indicated.

Family Support Information:

Turner Syndrome Society of Canada

Tel: 1-800-465-6744

www.TurnerSyndrome.ca

References

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