Turner Syndrome

Turner syndrome occurs when an X-chromosome is lost, resulting in 45, XO. This lost chromosome results in the constellation of signs and symptoms delineating Turner syndrome, including short stature, shield chest, webbing of the neck, peripheral lymphedema, ovarian dysgenesis, streak ovaries, decreased estrogen in the setting of elevated FSH and LH, increased risks of dysgerminoma, bicuspid aortic valve and preductal coarctation of the aorta as well as horseshoe kidneys. These patients also experience menopause before menarche as a result of dysgenesis of the ovaries, also demonstrated by primary amenorrhea as these patients never establish menstruation.



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Signs and Symptoms

Female (XO)
Female with X and O
Turner Syndrome is due to a chromosomal abnormality in which all or part of one of the sex chromosomes is absent or abnormal. The most common form is 45, XO. The missing or altered X chromosome of Turner syndrome causes errors during fetal development of the ovaries with subsequent inadequacy of estrogen production to support reproductive and sexual maturation.

Short stature
Shorts Statue
Short stature is a common physical characteristic among these patients. These females generally find their maximum height approximately 20 cm below that of the average female.

Shield Chest
Shield Chest
Shield, or broad, chest is a characteristic physical abnormality in these patients with related, widely-spaced nipples.

Cystic Hygroma (Webbing of Neck)
Sisters-with-hydrant and Webbing on Neck
Cystic hygromas are common physical abnormalities in Turner syndrome that are due to congenital lymphatic malformations. Cystic hygromas are benign, but can be disfiguring with extra folds of skin extending from the tops of the shoulders to the sides of the neck. This provides Turner syndrome patients with their characteristic webbed neck look.

Lymphedema in Hands and Feet
Lime-edamame on Hands and Feet
Lymphedema of the hands and feet are common symptoms of Turner syndrome.

Ovarian Dysgenesis
Ovary Disc-genie
Turner Syndrome is characterized by abnormal ovarian development during embryonic growth and followed by inevitable, premature ovarian failure with the subsequent loss of reproductive functions.

Streak Ovary
Streak across Ovary
With ovarian dysgenesis, the immature and malformed gonadal tissue becomes functionless and fibrous, attracting the moniker, streak gonads

Decreased Estrogen
Down-arrow Easter-egg
Estrogen is primarily produced by the ovaries; however, it is insufficiently produced in Turner syndrome due to ovarian dysgenesis and leads to many of the clinical signs and symptoms found in these patients.

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Increased FSH

Up-arrow Fish

Ovarian dysgenesis leads to inability to produce adequate estrogen levels, and consequently, a loss of negative feedback regulation on the production of FSH and LH.

Increased LH

Up-arrow Luge

Ovarian dysgenesis leads to decreased estrogen levels, and consequently a loss of negative feedback regulating the production of FSH and LH.

Dysgerminoma

Disc-German-gnome

A dysgerminoma is a malignant germ cell tumor occurring in the ovaries. Women with Turner syndrome are at an increased risk for developing dysgerminomas.

Bicuspid Aortic Valve

Bike-cupid at A-orca Valve

A bicuspid aortic valve is a congenital condition where two of the aortic valvular leaflets fuse during development, resulting in a valve that is bicuspid instead of tricuspid. Up to 15% of adults with Turner syndrome have bicuspid aortic valves. This predisposes the valve to early failure and calcification, leading to valvular dysfunction via either stenosis or regurgitation.

Preductal Coarctation of the Aorta

Priest-duck Corking the A-orca

Preductal coarctation of the aorta is a congenital narrowing of the aorta proximal to the ductus arteriosus. This coarctation occurs when an intracardiac anomaly during fetal development decreases blood flow through the left side of the heart, leading to hypoplastic maturation of the aorta.

Horseshoe Kidney

Horseshoe Kidneys

A horseshoe kidney is a congenital disorder where the inferior poles of the kidney fuse together during development, forming a horseshoe-shape. Horseshoe kidney is often asymptomatic.

Menopause before Menarche

Man-paws before Man-arch

Women with Turner syndrome will experience menopause before menarche as a result of the ovarian dysgenesis and inability to properly produce and regulate the interplay between GnRH, FSH, LH and estrogen as the underdeveloped and malfunctioning ovaries cannot manufacture estrogen sufficiently.

Amenorrhea

Amen-tampon

Amenorrhea is the absence of a menstruation in a woman of reproductive age. Turner syndrome is a cause of primary amenorrhea, with menstruation never satisfactorily initializing due to inadequate estrogen production from ovarian dysgenesis.

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