Patient Guide to Turner Syndrome and the Heart

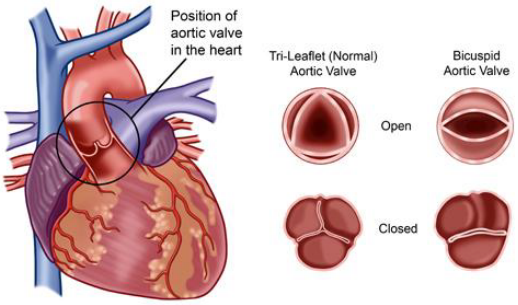
The Heart in Turner Syndrome

Cardiovascular abnormalities are one of the most common complications in girls and women with Turner Syndrome (TS). Congenital heart defects, or heart defects present at birth, occur in approximately 35% of girls with TS. Aortic defects in TS are associated with lymphatic anomalies. Adults living with TS may also be at a higher risk of developing cardiovascular complications, such as hypertension (high blood pressure), atherosclerosis (hardening of the arteries), and aortic dilatation and dissection (expansion or tearing of the main artery in the body). These complications may not always cause symptoms; therefore it is important for girls and women with TS to have cardiac screening throughout their lives.

Congenital Heart Defects in Turner Syndrome

Bicuspid Aortic Valve

The heart consists of four chambers and four valves. The chambers pump blood and the valves open and close to allow the forward flow of blood to the body and the lungs, and also to prevent the backward flow of blood as the heart is contracting. The aortic valve is a one-way valve between the heart and the aorta, the main artery from the heart that distributes oxygen-rich blood to the body.The normal aortic valve is has three small flaps, or leaflets. A bicuspid aortic valve is present when the valve only has two leaflets. It may function adequately for many years without causing any complications. Over time, calcium deposits on or around the valve leaflets may lead to aortic valve stiffening and narrowing (stenosis). This condition can cause the valve to leak (insufficiency). Bicuspid aortic valve is also associated with complications such as infections of the leaflet lining (endocarditis). The most common cardiac problem found in TS, bicuspid aortic valve (BAV) affects up to 30% of women with Turner syndrome.



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Hypoplastic Left Heart Syndrome

Hypoplastic Left Heart Syndrome (HLHS) is rare, and can occur in a small minority of patients with Turner syndrome. In a newborn with this condition, the structures on the left side of the heart are severely underdeveloped. Treatment usually requires multiple cardiac surgeries or less frequently, heart transplant. Long-term outcomes are 65-70% survival at 5 years.

Partial Anomolous Pulmonary Vein Connection

Tips & Recommendations for Heart Health in Girls and Women Living with TS

All girls and women with TS should undergo cardiovascular risk screening, as well as an ECG, Echocardiogram, or MRI if indicated. The frequency of these tests will depend on whether underlying problems have been detected.

Appropriate follow-up of heart defects and frequent monitoring and management of diabetes, lipid abnormalities, and blood pressure are essential for patients with TS.

Heart-healthy lifestyle modifications, such as diet and exercise, are important for maintaining long-term cardiovascular health.

Regular moderate physical activity is recommended. Some exercises may be contraindicated in those with underlying heart problems (e.g. vigorous activity or weight lifting) Consult your doctor before starting any exercise regimen.

Be an advocate for your own health, and learn about your own heart health status. If you have a heart condition you may want to consider wearing a medic alert bracelet or carrying a wallet card describing the problem.

Do not put off seeing your cardiologist, even if you have never had any problems. Heart issues can arise throughout the lifespan, so being proactive is important.

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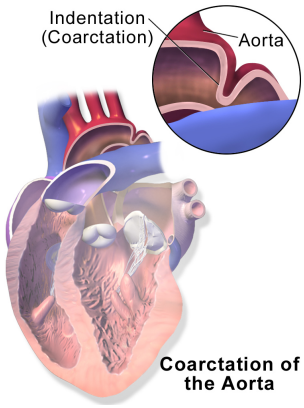
Coarctation of the Aorta

The aorta is the major blood vessel carrying oxygenated blood to the body. From the main pumping chamber of the heart, the aorta courses up towards the head, giving off branches that supply the head and arms with blood. It then curves downward, and finally divides into two main arteries into the legs. Coarctation of the Aorta is the narrowing of the upper aorta. It occurs in approximately 10-15% of girls with TS. Due to the narrowing, the heart must work harder to pump blood through the aorta. Without treatment, this can cause enlargement, and eventually failure of the heart. If severe, the coarctation may need surgical repair during the newborn period. Otherwise, it may be treated by expanding the narrowing with a small balloon inserted into the aorta (angioplasty) in older infants.



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Acquired Cardiac Conditions

Ascending Aorta Dilatation & Dissection

In TS, dilatation of the aorta usually involves its initial portion, in particular the tubular portion (ascending aorta). Ascending aortic dilatation is thought to occur in approximately 10-25% of older patients with Turner syndrome, particularly those with bicuspid aortic valves (BAV), but can occur without any other cardiac lesion. As the aorta progressively dilates, the wall becomes thinner and weaker. This may result in a tear (dissection) in the internal lining of the aortic wall, and can lead to heart attack or stroke. Most patients will remain asymptomatic, however symptoms of severe chest pain radiating through to the back can be a medical emergency. Frequent cardiac imaging is recommended. Current studies are exploring if medications such as beta-blockers or angiotensin receptor blockers may be helpful.

Hypertension

High blood pressure (hypertension) is commonly seen in about 50% of adults with TS. Although the mechanism is unclear, it appears that there may be something specific to TS that increases the risk. Screening for hypertension should be done on a regular basis.

Atherosclerosis

Atherosclerosis (hardening & narrowing of the arteries) also appears to be increased in TS. However, it remains unclear if there is something specific to TS that is contributing. Diet changes or medications to reduce cholesterol levels should be discussed with your health care .

Pregnancy Concerns for Women with TS

According to current guidelines, spontaneous or assisted pregnancy in TS should be considered only after thorough cardiac evaluation. Alarming reports of fatal aortic dissection during pregnancy and the postpartum period have raised concern about the safety of pregnancy in TS. A history of surgically repaired cardiovascular defect, the presence of bicuspid aortic valve, or current evidence of aortic dilatation or systemic hypertension are considered contraindications to pregnancy. TS patients who do become pregnant are at increased risk for eclampsia and diabetes as well as aortic dissection, and require specialist monitoring throughout gesta- tion and the postpartum period.

References

1) Turner Syndrome: Across the Lifespan. Edited by Jill Hamilton and Irena Hozjan. The Turner Syndrome Society of Canada. Published by Castlemore Graphics, Brampton, 2008.

2) Assessing and Addressing Cardiovascular Risk in Adults with Turner Syndrome- Turtle et al, Clinical Endocrinology, May 2013

3) Management of cardiovascular disease in Turner Syndrome (Report)- Joshua Thomas and Anji T Yetman, Expert Review of Cardiovascular Therapy, Dec 2009.

4) Congenital Cardiovascular Disease in Turner Syndrome- Carolyn A. Bondy, Congenital Heart Disease, January/Februrary 2008.

5) Aortic Dilatation and Dissection in Turner Syndrome- Carolyn A. Bondy, Circulation, September 2007.

6) Turner, know your body! An information book on Turner Syndrome- Novo Nordisk 2009. Edited by Claus H. Gravholt.

7.) Association Between Fetal Lymphedema and Congenital Cardiovascular Defects in Turner Syndrome. Loscalzo ML, Van PL, Ho VB, Bakalov VK, Rosing DR, Malone CA, Dietz HC, Bondy CA. Association between fetal lymphedema and congenital cardiovascular defects in Turner syndrome. Pediatrics. 2005; 115: 732–735

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