The Heart in Turner Syndrome

Turner Syndrome Foundation Workshop
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1. Heart disease in TS
    a. At least 23 – 50% of all TS individuals
    b. Major cause of early mortality in TS adults
2. Congenital heart disease
    a. Bicuspid aortic valve (14 – 30%)
        i. No effect
        ii. Stenosis (obstruction)
        iii. Regurgitation (leakage)
        iv. Aortic dilation
    b. Aortic coarctation (7 – 18%)
        i. Association with elongated arch (49%)
        ii. Association with aortic dilation
        iii. Association with hypertension
    c. Atrial and ventricular septal defects (1 – 8%)
    d. Partially anomalous pulmonary venous connection (1 – 13%)
    e. Left superior vena cava (up to 13%)
3. Acquired heart disease
    a. Hypertension (30 – 50%)
        i. Increased frequency with age
    b. Aortic dilation (20 – 30%)
        i. Risk factors in TS
            1. Bicuspid aortic valve
            2. Aortic coarctation
            3. Hypertension
            4. Intrinsic problem with aortic wall
        ii. Increased risk for aortic dissection and rupture
            1. Rare (1 – 2%)
            2. Proximal aortic diameter ≥ 2.5 cm/m^2
            3. High mortality and morbidity
            4. Occurs earlier in TS women
            5. > 20 years of age

The Heart in Turner Syndrome (Continued)

6. Risk factors
    a. Hypertension
    b. Congenital heart disease
    c. Pregnancy
7. 11% without known risk factors

c. Coronary artery disease
    i. Premature atherosclerosis
        1. Hypertension
        2. Diabetes
        3. Obesity
        4. Increased cholesterol level
        5. Estrogen deficiency
    ii. Genetic predisposition

4. Recommendations (Bondy, NIH, 2007)
    a. Cardiovascular screening (all patients at time of diagnosis)
        i. Evaluation by cardiologist with expertise in congenital heart disease
        ii. Comprehensive exam including blood pressure in all extremities
        iii. Clear imaging of the heart, aortic valve, aortic arch, and pulmonary veins
            1. Echo usually adequate for infants and young girls
            2. MRI and echo for older girls and adults
            3. Electrocardiogram
    b. Monitoring (follow-up depends on clinical situation)
        i. For patients with apparently normal cardiovascular system and age-appropriate blood pressure
            1. Re-evaluation with imaging at timely occasions (at transition to adult clinic, before attempting pregnancy, or with appearance of hypertension)
            2. Girls who have only had an echo should undergo an MRI when old enough to cooperate with the procedure
            3. Otherwise, imaging every 5 to 10 years
        ii. For patients with cardiovascular pathology
            1. Treatment and monitoring to be determined by cardiologist
            2. Education about risk, compliance, and presenting symptoms for associated complications
            3. Counseling about pregnancy and exercise (no isometric exercise or contact sports)