CARDIOVASCULAR AND GENERAL HEALTH STATUS OF ADULTS WITH TRISOMY 21

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Background: There is a growing population of adults with Trisomy 21 (T21). Little data exists to assist practitioners in counseling patients and caregivers regarding expected long-term outcomes. We sought to assess cardiovascular status, as well as co-morbidities in this patient cohort.

Methods: A retrospective review of the electronic medical databases of 2 medical institutions with widespread coverage of the state was undertaken. Patients with a diagnosis of T21 who had a cardiology clinic visit or echocardiogram during 1995-2012 were identified. Patients of adult age (>21 years) were included. Demographic, surgical, and clinical variables were recorded. Correlates of adverse outcome defined as death, adult onset arrhythmia or heart failure, and adult cardiac surgery were sought.

Results: Ninety-four patients (49% female) with a median age of 30 (21-56) years and body mass index (BMI) of 29.1±7.6 m/kg² were identified. Congenital heart defects (CHD) requiring surgery in childhood were present in 68 (72%) patients, minor defects in 13 (14%) and no CHD in 13. Of the 81 patients with CHD, 43 had an isolated atrioventricular septal defect (AVSD), 7 isolated tetralogy (ToF), 2 AVSD with ToF, 2 single ventricle, 17 a VSD, and 10 had other defects. Of the 68 patients requiring surgery, definitive repair was performed in only 53 (78%). Adverse outcome was present in 38 (40%). There were 8 adult deaths at a mean age of 41.4±6.2 years. Thirty patients required 2.2 (1-14) hospitalizations for cardiorespiratory disease in adulthood. Of the 53 post-operative patients, 25% required repeat surgery in adulthood. Co-morbidities included hypothyroidism in 42 (45%), dementia in 4 (4%), sleep apnea in 34 (36%), pulmonary hypertension in 37 (39%) and obesity in 36 (38%). Of the 22 (23%) patients developing late arrhythmias, 13 required implantable cardioverter defibrillator or pacemaker. Patients with an adverse outcome had a higher BMI (31.3 vs. 27.8 m/ kg², p=0.03), and were more likely to have pulmonary hypertension (71% vs. 23%, p<0.0001).

Conclusions: Adults with Trisomy 21 are living into their 6th decade and have a high incidence of arrhythmias, need for repeat cardiac surgery, and pulmonary hypertension.