CONGENITAL CV SURVIVORS AND CATCH 22

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OBJECTIVES

After this lecture the attendee should be familiar with contemporary issues for survivors of congenital heart disease treatment, which include developmental, psychosocial challenges, associated genetic and health issues as well as the long-term outlook.

SURVIVORS OF CONGENITAL HEART DISEASE

Approximately 40,000 infants in this country are born yearly with structural heart disease. This accounts for the leading cause of congenital malformations in the neonatal period. Significant advances in medical care, diagnostic techniques, anesthetic, perfusion and surgical management strategies over the past several decades now allow for survival of a great majority of these children into adulthood. This has resulted in a flourishing population of older children, young adolescents, and adults with congenital heart disease at a phenomenally increasing rate.

In the United States alone, it is estimated that there are over one million adults with unrepaired, palliated or "corrected" congenital heart disease and this cohort is growing at rate of 5% per year. In the recent past and for the first time in history, the number of adults with congenital cardiovascular malformations equals the number of children with these disorders.

It is estimated that nearly 40% of patients with congenital heart disease will require surgical intervention to palliate or "correct" their condition some time during their lifetime. As a result of the recognition of the detrimental effects associated with delayed interventions, there has been continued interest in primary early "correction". With ongoing medical and surgical refinements, and as survival rates and life expectancy further increase, it is anticipated that more members of this population will grow to become adults.

Clinical outcome in congenital heart disease is dependent on the anatomic diagnosis and the possibility of successful palliation or "correction". Palliated patients continue to have an abnormal circulation although the goals of the surgical intervention are to decrease the likelihood of the severe repercussions of the disease. Only patent ductus arteriosus ligation and early repair of an atrial septal defect are truly correctable lesions, and are associated with normal life expectancy. The expectation for the remaining surgical interventions is to improve cardiac function and hemodynamics, thus generally improving clinical outcome. In a significant number of patients this results in a favorable, although not completely normal quality of life. The fact is that in almost all patients with congenital heart disease some residual abnormality exists. In others, pathology may remain or develop after surgery related to their primary disease or treatment. This may lead to moderate to significant cardiovascular and respiratory impairment. The residua and sequelae may necessitate further medical or surgical therapy and be associated

with significant morbidity and early death. The management of these patients is guided by a number of factors, but to a significant extent, by the natural history of the defect and residual pathology. Factors to consider and potential morbidity following palliation or "repair" of congenital heart disease include: ventricular dysfunction, conduction disturbances and dysrhythmias, altered ventricular loading conditions, potential for bacterial endocarditis , risk of systemic air embolization, effects of chronic cyanosis, excessive pulmonary blood flow, pulmonary hypertension/pulmonary vascular changes, nerve palsies, and the perioperative stress response of the planned procedure.

In recognition of these considerations there has been ongoing concern regarding the care of survivors of congenital cardiovascular disease. Issues that have lately also received increasing attention regarding this patient population include: insurability, who are the most suitable health care providers, where should they be cared for (pediatric facilities, adult hospitals, centers specializing in congenital heart disease), the unique psychological needs of this patient populations, and others.

It is anticipated that an escalating number of palliated and "repaired" patients will need to undergo surgical procedures related or unrelated to their heart disease requiring anesthetic care. Understanding the anatomy, pathophysiology, hemodynamic and functional consequences of these defects, as well as potential long-term issues will likely allow for improved care of these patients. The session will focus on contemporary outcomes of patients affected by congenital heart disease and challenges of the survivors as they transition into adulthood.

CATCH 22

The acronym CATCH 22 describes a syndrome that comprises a constellation of abnormalities including Cardiac defects, Abnormal facies, Thymic hypoplasia, Cleft palate, and Hypocalcemia. Other problems that may affect these individuals include velopharyngeal insufficiency and mild conductive hearing loss. The CATCH 22 syndrome encompasses a wide spectrum of disorders caused by a microdeletion on the long arm of chromosome 22, otherwise known as the 22q11 deletion syndrome (22q11DS). Most individuals, in the range of 85-90%, have de novo mutations.

The 22q11 deletion syndrome is estimated to affect between 1 in 2,000–5,000 individuals, representing a relatively common genetic disorder. This heterogenous group of disorders include: Shprintzen syndrome, velo-cardio-facial syndrome, conotruncal face anomaly, and DiGeorge syndrome. This last entity has been considered by some to represent a severe form at the end of the wide and complex spectrum.

Significant heterogeneity is recognized in the clinical features and manifestations of the 22q11DS, variably affecting the central nervous system, cardiovascular system, endocrine system, auditory system, ocular system, skeletal muscles and the skin. Among patients with 22q11DS the most prevalent defects include cardiac malformations, speech delay, and immunodeficiency. A high frequency of cognitive and developmental problems, learning disabilities, and behavioral abnormalities are observed. A known association is recognized between 22q11DS and psychiatric disease. Affected individuals have a 25-30% risk for the development of schizophrenia in adulthood, as well as an increased risk for psychiatric problems

that include bipolar disorders, obsessive-compulsive conditions, and other schizo-affective illnesses.

The cardiovascular malformations most commonly seen in these disorders include conotruncal defects such as interrupted aortic arch, truncus arteriosus, tetralogy of Fallot, and pulmonary atresia with ventricular septal defect. Although the presence of 22q11.2 deletion syndrome in many reports does not worsen the surgical prognosis, it has been documented that a proportion of children within the CACTH 22 spectrum have a very poor developmental outcome following cardiac surgery. This is not thought to be attributable to the cardiac condition and its treatment alone, and may represent either a pre-existing component of the syndrome or an interaction between the syndrome and its treatment.

The presentation will review the clinical features of the CATCH 22 syndrome, highlighting the cardiac manifestations, as well update the knowledge on the cognitive/and or neuropsychiatric deficits affecting this group of patients. Available data regarding neurodevelopmental outcomes will be discussed.

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