Down Syndrome and Congenital Heart Disease

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Disclosures

• Data Safety and Monitoring Board for Native RV Outflow Tract Transcatheter Valve - Medtronic, Inc.
Down Syndrome and Congenital Heart Disease

• Trisomy 21 (Down syndrome):
  – Most common chromosomal anomaly associated with congenital heart disease
  – Down syndrome patients account for up to 10% of all congenital heart disease patients
  – Congenital heart disease occurs in 40-50% of Down syndrome patients
Down Syndrome and Congenital Heart Disease

• Types of congenital heart defects:
  – Atrioventricular canal defects (aka endocardial cushion defects or AV canal defects)
    • Primum ASD 8%
    • Complete AV canal (87%)
    • Isolated cleft in left AV valve (5%)
  – Ventricular septal defects
  – Tetralogy of Fallot (Ventricular septal defect plus right ventricular outflow tract obstruction)
  – Patent ductus arteriosus
Down Syndrome and Congenital Heart Disease

- Important considerations in children with Down syndrome and congenital heart disease
  - Accelerated development of pulmonary vascular obstructive disease (PVOD) with left to right shunts
  - Airway obstruction due to tonsil and adenoid tissue can exacerbate pulmonary hypertension due to airway obstruction
Complete AV Canal

Figure 41.
Endocardial cushion defect of the complete variety. View from the interior of the left side of the heart. Characteristically, there is a crescent-shaped defect in the lowermost part of the atrial septum. An atrioventricular valve common to both sides of the heart and a subvalvular ventricular septal defect are other features of the complex.
Complete AV Canal Diagnosis

- Echocardiogram generally sufficient
- Heart Catheterization
  - To clarify echo anatomy
  - To assess PVR response to O2 or NO
- Important anatomic features
  - Additional VSD’s
  - Number of LV papillary muscles
  - Pulmonary and systemic venous connections - heterotaxy
  - Origins of great vessels
  - Size of ventricles and relationship of AV canal to ventricles
Complete AV Canal Indications for Repair

- Congestive heart failure
- Failure to thrive
- Pulmonary hypertension
Complete AV Canal Timing of Repair

- Congestive heart failure refractory to medical therapy regardless of age or size

- Complete repair before 6 months of age
  - Down syndrome - early onset of pulmonary vascular disease (PVOD can occur by 9 months)
  - All patients - seasonal viral respiratory illnesses (RSV)
Ventricular Septal Defect
Ventricular Septal Defects

- Infundibuloventricular
- Infundibular
- Posterior (AV canal type)
- Muscular
- Malalignment

Ventricular Septal Defects
Pulmonary Vascular Disease
Tetralogy of Fallot
Tetralogy of Fallot
# Down Syndrome and AV Canal Defects

## Outcomes of Surgical Repair 2011-13

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<tr>
<th>Defect</th>
<th>n</th>
<th>Survival</th>
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<tbody>
<tr>
<td>Complete AV Canal</td>
<td>81*</td>
<td>98%</td>
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<tr>
<td>Transitional AV Canal</td>
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<tr>
<td>Primum Atrial Septal Defect</td>
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*One death in premature infant*
<table>
<thead>
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<th>Condition</th>
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<th>Survival</th>
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<tr>
<td>Ventricular septal defect</td>
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<td>Tetralogy of Fallot</td>
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<tr>
<td>Patent ductus arteriosus</td>
<td>43</td>
<td>100%</td>
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Outcomes for Other Defects (2011-2013)
Down Syndrome and Congenital Heart Disease

• Congenital heart disease is common in patients with Down Syndrome
• Without surgery, congenital heart disease is a major source of morbidity and premature death
• Surgical repair of most congenital heart disease can be accomplished with low risk and improves survival and quality of life