ADULT CONGENITAL HEART DISEASE
ACHD

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Rabin Medical Centre,
Campus Beilinson,
Sackler Faculty of Medicine,
University of Tel - Aviv
A Brief History of ACHD

- Joseph K. Perloff
- Jane Somerville
- John Morch

ACHD in North America – Transcending Borders
1975 and 1977: The birth of units specializing in Adult Congenital Heart Disease (ACHD)

Royal Brompton Hospital, Imperial College of Medicine (London, UK)

UCLA School of Medicine (USA)
SUCCESSORS

- 1983 University of Toronto Congenital Cardiac Centre for Adult (TCCCA) - 1959 (Drs. Morch, Evans)

- Mayo Clinic Adult Congenital Cardiac Clinic

- Boston, Cleveland, London, Chiba
A Brief History of ACHD

The Canadian Adult Congenital Heart (CACH) Network – founded 1991

ACHD in North America – Transcending Borders
ACHD in the US

Recent Developments in ACHD

The impressive growth of American ACHD Clinics.

ACHD in North America – Transcending Borders
EUROPE

Euro Heart Survey on Adult Congenital Heart Disease

79 centers
26 countries

Europe: 727 million inhabitants
1.2-2.7 million patients with ACHD

4168 patients

Engelbert PM, et al. Heart [Epub ahead of print].
### Scope of the problem. Epidemiology

**ACHD**

<table>
<thead>
<tr>
<th>Country</th>
<th>Population</th>
<th>80’s</th>
<th>2000</th>
<th>2008</th>
<th>2020</th>
</tr>
</thead>
<tbody>
<tr>
<td>United States</td>
<td>320,000,000</td>
<td>300,000</td>
<td>800,000</td>
<td>1,000,000</td>
<td>1,400,000</td>
</tr>
<tr>
<td>United Kingdom</td>
<td>60,000,000</td>
<td>150,000 (2005)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Canada</td>
<td>32,000,000</td>
<td>180,000</td>
<td></td>
<td>11,000 7,000 active (Toronto CCA)</td>
<td></td>
</tr>
<tr>
<td>Israel</td>
<td>7,500,000</td>
<td></td>
<td></td>
<td>~3,000 (ACHD Unit, Rabin)</td>
<td></td>
</tr>
</tbody>
</table>
Scope of the problem. Epidemiology

1. Each year approximately 40,000 newborn with congenital heart disease (CHD) in the US
2. Moderate to severe CHD, 6/ 1,000 live birth
3. Approximately 3 - 5/ 1,000 need some kind of cardiac care during their life*
4. Almost 1 in 150 young adults will have some form of CHD in the next decade

* Bicuspid aortic valve 19/ 1,000; tiny VSD 75/ 1,000
# Number of patients

## Regional ACHD centers

<table>
<thead>
<tr>
<th>6 largest national ACHD centers</th>
<th>Toronto</th>
<th>Brompton</th>
<th>Boston</th>
<th>UCLA</th>
<th>Mayo</th>
<th>Cleveland</th>
</tr>
</thead>
<tbody>
<tr>
<td>Active patients 2004</td>
<td>7,000</td>
<td>5,000</td>
<td>3,200</td>
<td>2,500</td>
<td>2,000</td>
<td>1,700</td>
</tr>
<tr>
<td>Median (pts/center)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>2,850</td>
</tr>
</tbody>
</table>

Niwa K. et al. *International Journal of Cardiology* 2004;96:211

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Courtesy Dr. A. Marelli, Quebec
Adult Cardiology vs. ACHD
Adult Cardiology vs. ACHD
Question 1

- Combined incidence of congenital heart defects (CHD)?
- Prevalence of CHD in adults/ kids?
- First time diagnosis of CHD in adults (%)?
- Survival of pts with CHD?
- Mortality: overall; surgical; sudden death?
- Recurrence rate of CHD: parents, siblings?
- Sex distribution of CHD: overall; type specific lesions?
- Genetic associations of CHD?
- Most frequent CHD: adults; kids?
- Specific lesions
INCIDENCE OF CONGENITAL HEART DISEASE (CHD)

- CHD is the most common group of human birth defects
- Combined incidence of CHD in humans

<table>
<thead>
<tr>
<th>Live birth</th>
<th>~ 0.4 – 0.8% (4-8/1000)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Spontaneously aborted fetuses</td>
<td>~ 10%</td>
</tr>
</tbody>
</table>

J.A. Towbin, J. Belmont, 2000; Wulfsberg EA et al., 1991; Ferencz et al., 1985
Question 2

- Combined incidence of congenital heart defects?
- **Prevalence** of CHD in adults/ kids?
- First time diagnosis of CHD in adults (%) ?
- Survival of pts with CHD ?
- Mortality: overall; surgical; sudden death?
- Recurrence rate of CHD: parents, siblings?
- Sex distribution of CHD: overall; type specific lesions?
- Genetic associations of CHD?
- Most frequent CHD: adults; kids?
- Specific lesions
Most studies on CHD frequency refer to birth prevalence with variable reported rates.

Prevalence of CHD has been determined in the general population of Quebec (Canada) using 18 years data (23-25% of population).

**Observed prevalence:**
- 11.89/1000 children
- 4.09/1000 adults
# Prevalence of severe and other CHD in the year 2000

<table>
<thead>
<tr>
<th></th>
<th>Adults alive in 2000</th>
<th>Children alive in 2000</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>N (%)</td>
<td>Per 1000 adults</td>
</tr>
<tr>
<td><strong>All CHD</strong></td>
<td>23563 (100)</td>
<td>4.09</td>
</tr>
<tr>
<td>Severe lesions</td>
<td></td>
<td></td>
</tr>
<tr>
<td>TOF or TA</td>
<td>1001</td>
<td>0.17</td>
</tr>
<tr>
<td>Transposition</td>
<td>834</td>
<td>0.14</td>
</tr>
<tr>
<td>Univentricular</td>
<td>235</td>
<td>0.04</td>
</tr>
<tr>
<td><strong>All severe</strong></td>
<td>2205 (9)</td>
<td>0.38</td>
</tr>
<tr>
<td>Other lesions</td>
<td></td>
<td></td>
</tr>
<tr>
<td>ASD</td>
<td>5076</td>
<td>0.88</td>
</tr>
<tr>
<td>VSD</td>
<td>4486</td>
<td>0.78</td>
</tr>
<tr>
<td>PDA</td>
<td>103</td>
<td>0.02</td>
</tr>
</tbody>
</table>

A. Marelli et al., Circulation 2007
Extrapolation: Quebec Data

Annual Increase ~ 1,000 Adults per Year

Prevalence of Adults with CHD
(4.09 per 1000)
~ 96,000

Prevalence of severe CHD in Adults
(0.38 per 1000)
~ 9,000

Courtesy of Dr. E. Oechslin, Toronto
OVERALL PREVALENCE OF CONGENITAL HEART DEFECTS VARIES BY RACE AND ETHNICITY

1. Non - Hispanic White 14.4 /1000 live births

2. Non - Hispanic Black  12.8 /1000 live births

3. Others (American Indian, Pacific Asian)                      12.5 /1000 live births

4. Hispanics 8.8 /1000 live births

3075 congenital CV malformations of 2303 children from a birth population of 235,230

Tania Mangones et al., IJC, 2010
Question 3

- Combined incidence of congenital heart defects?
- Prevalence of CHD in adults/ kids?
- **First time diagnosis of CHD in adults (%) ?**
- Survival of pts with CHD ?
- Mortality: overall; surgical; sudden death?
- Recurrence rate of CHD: parents, siblings?
- Sex distribution of CHD: overall; type specific lesions?
- Genetic associations of CHD?
- Most frequent CHD: adults; kids?
- Specific lesions
DIAGNOSIS OF CHD

1. Approx. 60% in babies
2. Children, 30%
3. Adults, 10%
Question 4

- Combined incidence of congenital heart defects?
- Prevalence of CHD in adults/ kids?
- First time diagnosis of CHD in adults (%)?
- Survival of pts with CHD?
- Mortality: overall; surgical; sudden death?
- Recurrence rate of CHD: parents, siblings?
- Sex distribution of CHD: overall; type specific lesions?
- Genetic associations of CHD?
- Most frequent CHD: adults; kids?
- Specific lesions
# Complex Congenital Heart Disease

<table>
<thead>
<tr>
<th>Year of birth</th>
<th>Survival rate</th>
</tr>
</thead>
<tbody>
<tr>
<td>1940-1959</td>
<td>10%</td>
</tr>
<tr>
<td>1960 – 1979</td>
<td>50%</td>
</tr>
<tr>
<td>1980 - 1989</td>
<td>80%*</td>
</tr>
<tr>
<td>1990 – 2000s</td>
<td><strong>85 – 95%</strong></td>
</tr>
</tbody>
</table>

Warnes et al., JACC, 2001 (modified)
GROWING POPULATION OF ADULT PATIENTS WITH CHD

40s – 70s
80% - 90% kids
10% - 20% adults

90s – 2000
50% kids
50% adults

Next decade or two
40% kids
60% adults

Today
all CHD, adults/ kids 55/ 45%; severe CHD 49/51%
Changing Age Distribution of Severe CHD 1985 - 2000


Courtesy of Dr. E. Oechslin, Toronto
Age Distribution

n = 4168
CHD is a Continuum from Fetal Life until Adulthood
Survival Rate in CHD and Milestones in Cardiac Surgery

%


20 25 40 55 70 80 85

BT-Shunt Fallot-OP Atrial Switch-OP Fontan-OP Arterial Switch-OP

Courtesy of Dr. H. Kaemmerer, Munich
Question 5

- Combined incidence of congenital heart defects?
- Prevalence of CHD in adults/ kids?
- First time diagnosis of CHD in adults (%)?
- Survival of pts with CHD?
- **Mortality**: overall; surgical; sudden death?
- Recurrence rate of CHD: parents, siblings?
- Sex distribution of CHD: overall; type specific lesions?
- Genetic associations of CHD?
- Most frequent CHD: adults; kids?
- Specific lesions
## EuroHeart Survey. Mortality

<table>
<thead>
<tr>
<th></th>
<th>n</th>
<th>Number of death</th>
<th>5 yr mortality</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td>CV</td>
<td>Non CV</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Kaplan Meier estimate (%)</td>
<td>95% CI (%)</td>
</tr>
<tr>
<td>ASD sec.</td>
<td>882</td>
<td>5</td>
<td>4</td>
</tr>
<tr>
<td>VSD</td>
<td>628</td>
<td>6</td>
<td>2</td>
</tr>
<tr>
<td>TOF</td>
<td>811</td>
<td>8</td>
<td>3</td>
</tr>
<tr>
<td>Coarctation</td>
<td>551</td>
<td>2</td>
<td>1</td>
</tr>
<tr>
<td>TGA</td>
<td>363</td>
<td>9</td>
<td>0</td>
</tr>
<tr>
<td>Marfan syndrome</td>
<td>287</td>
<td>3</td>
<td>2</td>
</tr>
<tr>
<td>Fontan circulation</td>
<td>198</td>
<td>12</td>
<td>4</td>
</tr>
<tr>
<td>Cyanotic defects</td>
<td>390</td>
<td>44</td>
<td>10</td>
</tr>
<tr>
<td>Overall</td>
<td>4110</td>
<td>89</td>
<td>29</td>
</tr>
</tbody>
</table>

EHJ, 2006
EuroHeart Survey.

ACHD Mortality

Figure 1 Kaplan–Meier survival curves for the eight defects.

EHJ, 2006
# Clinical activity and cardiac surgery

<table>
<thead>
<tr>
<th>Settings</th>
<th>UCLA</th>
<th>Toronto</th>
<th>Cleveland</th>
<th>Boston</th>
<th>Mayo</th>
<th>Royal Brompton</th>
</tr>
</thead>
<tbody>
<tr>
<td>Adm./yr*</td>
<td>350</td>
<td>660</td>
<td>100</td>
<td>180</td>
<td>300</td>
<td>450</td>
</tr>
<tr>
<td>Pregn./yr</td>
<td>23</td>
<td>60</td>
<td>30</td>
<td>15</td>
<td>18</td>
<td>80</td>
</tr>
<tr>
<td>Surgery</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Oper./yr</td>
<td>170</td>
<td>150</td>
<td>50</td>
<td>70</td>
<td>100</td>
<td>100</td>
</tr>
<tr>
<td>Re-do</td>
<td>80%</td>
<td>35%</td>
<td>25%</td>
<td>29%</td>
<td>56%</td>
<td>60%</td>
</tr>
<tr>
<td>Postop. mort.</td>
<td><strong>2%</strong></td>
<td><strong>1.5%</strong></td>
<td><strong>2%</strong></td>
<td><strong>1.5%</strong></td>
<td><strong>3%</strong></td>
<td><strong>1.5%</strong></td>
</tr>
</tbody>
</table>

* Ratio: # hospit./ total # of pts followed, averaged 10%

K. Niwa et. al., IJC, 2004
Surgery is corrective, if......

- ...atrial function is normal
- ...life expectancy is normal!
- ...there is no need for therapeutic measures during follow-up!
January 1989

The Journal of THORACIC AND CARDIOVASCULAR SURGERY

Honored Guest’s Address

Do we really correct congenital heart defects?

J. Stark, FRCS, FACS, FACC, London, England
Corrective Surgery....

- Atrial septal defect
- Ventricular septal defect
- Patent ductus arteriosus

... if treated during childhood!!
‘The surgeon fixed my heart:
I am cured’
CHD Patients in Ontario

Medium / High Risk for Complications: 50%

31,000 Children/Adolescents

38,000 Adults (>18 yrs)

Courtesy of Dr. E. Oechslin, Toronto
Mechanism of death
(197 deceased pts, 1981-1996)

E. Oechslin et al., AJC, 2000
Mechanism of Death by Diagnosis

E. Oechslin et. al., AJC, 2000
Mechanism of Death by Diagnosis

E. Oechslin et al., AJC, 2000
Sudden Cardiac Death in ACHD

- Late SCD (after CHD surgery)
- 0.9/1000 patient – years (significantly higher than in general population)
  1. Arrhythmic events
  2. Embolic events
  3. Acute ventricular failure
  4. Aneurysm rupture
  5. Eisenmenger syndrome

L. Harris, S. Belaji (in M. Gatzoulis et al.) ACHD, 2003
# Event rate of SCD in ACHD

<table>
<thead>
<tr>
<th>2.2/1000; 90% of cases</th>
<th>0.14/1000</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. TOF</td>
<td>• Left to right shunt lesions</td>
</tr>
<tr>
<td>2. CTGA</td>
<td>• Pulmonary valve stenosis</td>
</tr>
<tr>
<td>3. Coarctation (LVH)</td>
<td></td>
</tr>
<tr>
<td>4. Aortic stenosis</td>
<td></td>
</tr>
<tr>
<td>• Residual stenosis</td>
<td></td>
</tr>
<tr>
<td>• Coronary emboli</td>
<td></td>
</tr>
<tr>
<td>• Cerebral emboli</td>
<td></td>
</tr>
<tr>
<td>• IE</td>
<td></td>
</tr>
</tbody>
</table>

L. Harris, S. Belaji (in M. Gatzoulis et al.) ACHD, 2003
Circumstances of death in ACHD

<table>
<thead>
<tr>
<th>Pts (n)</th>
<th>Pts.-yrs</th>
<th>Death, n (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>8,595</td>
<td>26,500</td>
<td>231 (2.7)</td>
</tr>
</tbody>
</table>

**Main causes (%)**
- HF - 26%
- SCD - 22%

<table>
<thead>
<tr>
<th>Out of hospital (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>35 (rural 55 vs. urban 32)</td>
</tr>
</tbody>
</table>

**Season**
- Fall – slightly higher (NS)

**Sudden CV death (%)**

<table>
<thead>
<tr>
<th>During exercise (%)</th>
<th>Out of hospital (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>~ 40%</td>
<td>8%</td>
</tr>
<tr>
<td></td>
<td>62%</td>
</tr>
</tbody>
</table>

Non-sudden CV death ~18% out of hospital

Zomer AC et al., IJC, 2010
One - year Hospitalization Rate

Severe CHD | Other CHD | Quebeck population

Hospitalizations per 100 pts, 1999 - 2000

Emergencies

11% Cardiovascular
6% Infective
83% Others

Kaemmerer H, Oechslin E, ... Hess J. J Thorac Cardiovasc Surg 2003; 126:1048-52

Courtesy of Dr. E. Oechslin, Toronto
Cardiovascular Emergencies

- Arrhythmias: 11%
- Heart failure: 53%
- Cerebral ischemia: 6%
- Other cardiovascular: 7%
- Others: 2%
- Syncope: 2%
- Infections: 2%

Courtesy of Dr. E. Oechslin, Toronto

Kaemmerer H, Oechslin E, ... Hess J. J Thorac Cardiovasc Surg 2003; 126:1048-52
# Arrhythmias – ACHD patients

<table>
<thead>
<tr>
<th>Rhythm disturbance</th>
<th>Associated lesion</th>
</tr>
</thead>
<tbody>
<tr>
<td>Bradycardias</td>
<td></td>
</tr>
<tr>
<td>SN dx</td>
<td>CTGA - Mustard</td>
</tr>
<tr>
<td></td>
<td>CTGA - Senning</td>
</tr>
<tr>
<td></td>
<td>Fontan</td>
</tr>
<tr>
<td></td>
<td>Sinus venosus defect</td>
</tr>
<tr>
<td></td>
<td>Heterotaxy syndrome</td>
</tr>
<tr>
<td>AVB</td>
<td>AV septal defects</td>
</tr>
<tr>
<td></td>
<td>cCTGA</td>
</tr>
<tr>
<td>Surgically/ interventionaly induced AVB</td>
<td>VSD</td>
</tr>
<tr>
<td></td>
<td>SA stenosis</td>
</tr>
<tr>
<td></td>
<td>AVVR</td>
</tr>
</tbody>
</table>

ACC/ AHA ACHD Guidelines, 2008
<table>
<thead>
<tr>
<th>Rhythm disturbance</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Tachycardias</strong></td>
<td></td>
</tr>
<tr>
<td>WPW</td>
<td>Ebstein anomaly</td>
</tr>
<tr>
<td></td>
<td>cCTGA</td>
</tr>
<tr>
<td>IART (atrial flutter)</td>
<td>CTGA – Mustard</td>
</tr>
<tr>
<td></td>
<td>CTGA – Senning</td>
</tr>
<tr>
<td></td>
<td>Fontan</td>
</tr>
<tr>
<td></td>
<td>TOF</td>
</tr>
<tr>
<td>AFib</td>
<td>MV disease</td>
</tr>
<tr>
<td></td>
<td>AS</td>
</tr>
<tr>
<td></td>
<td>TOF</td>
</tr>
<tr>
<td>VT</td>
<td>Post palliation – single ventricle</td>
</tr>
<tr>
<td></td>
<td>TOF</td>
</tr>
</tbody>
</table>
Question 6

- Combined incidence of congenital heart defects?
- Prevalence of CHD in adults/ kids?
- First time diagnosis of CHD in adults (%)?
- Survival of pts with CHD?
- Mortality: overall; surgical; sudden death?
- Recurrence rate of CHD: parents, siblings?
- Sex distribution of CHD: overall; type specific lesions?
- Genetic associations of CHD?
- Most frequent CHD: adults; kids?
- Specific lesions
### CHD Recurrence Risk

**Baltimore – Washington Infant Study (BWIS)**

<table>
<thead>
<tr>
<th></th>
<th>Siblings (%) with CHD</th>
<th>One affected child</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>All forms of CHD</strong></td>
<td><strong>3 - 5</strong></td>
<td>-</td>
</tr>
<tr>
<td><strong>Subgroup of patients</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>- Lt. sided obstructive</td>
<td>4.5 – 13.8</td>
<td>-</td>
</tr>
<tr>
<td>lesions</td>
<td></td>
<td></td>
</tr>
<tr>
<td>- PAS</td>
<td></td>
<td></td>
</tr>
<tr>
<td>- Ebstein’s anomaly</td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Recurrence risk of CHD</strong></td>
<td></td>
<td><strong>At least x3 baseline risk</strong></td>
</tr>
</tbody>
</table>

Huhta James C. et al., 2006
# Risk of Recurrent Disease in Offspring of Parents with CHD

<table>
<thead>
<tr>
<th>Lesion</th>
<th>Mother affected</th>
<th>Father affected</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Risk of transmission (%)</td>
<td>Number of cases</td>
</tr>
<tr>
<td>AVSD</td>
<td>11.6</td>
<td>5/43</td>
</tr>
<tr>
<td>AS</td>
<td>8.0</td>
<td>36/248</td>
</tr>
<tr>
<td>Coarctation</td>
<td>6.3</td>
<td>14/222</td>
</tr>
<tr>
<td>ASD</td>
<td>6.1</td>
<td>59/969</td>
</tr>
<tr>
<td>VSD</td>
<td>6.0</td>
<td>44/731</td>
</tr>
<tr>
<td>PS</td>
<td>5.3</td>
<td>24/453</td>
</tr>
<tr>
<td>PDA</td>
<td>4.1</td>
<td>39/828</td>
</tr>
<tr>
<td>TOF</td>
<td>2.0</td>
<td>6/301</td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td><strong>5.8</strong></td>
<td><strong>222/3795</strong></td>
</tr>
</tbody>
</table>

Nora JJ, JACC 1994; 23: 1468 – 71
## EuroHeart Survey

**Percentage of women with at least one full term pregnancy**

<table>
<thead>
<tr>
<th>Pathology</th>
<th>Ever pregnant</th>
<th>General Dutch Population %</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>%</td>
<td>N observed</td>
</tr>
<tr>
<td>ASD sec.</td>
<td>50</td>
<td>232/462</td>
</tr>
<tr>
<td>VSD</td>
<td>40</td>
<td>111/281</td>
</tr>
<tr>
<td>TOF</td>
<td>36</td>
<td>129/354</td>
</tr>
<tr>
<td>Coarctation</td>
<td>40</td>
<td>76/191</td>
</tr>
<tr>
<td>TGA</td>
<td>30</td>
<td>41/137</td>
</tr>
<tr>
<td>Marfan syndrome</td>
<td>36</td>
<td>50/137</td>
</tr>
<tr>
<td>Fontan circulation</td>
<td>2</td>
<td>2/88</td>
</tr>
<tr>
<td>Cyanotic defect</td>
<td>18</td>
<td>42/238</td>
</tr>
<tr>
<td><strong>Overall</strong></td>
<td><strong>36</strong></td>
<td><strong>683/1888</strong></td>
</tr>
</tbody>
</table>

EHJ, 2006
Predictors of maternal cardiac complications (newly identified predictors in bold)

<table>
<thead>
<tr>
<th>Variable</th>
<th>Odds ratio</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td>History of arrhythmias</td>
<td>4.3</td>
<td>0.0011</td>
</tr>
<tr>
<td>Other (cardiac) medication before pregnancy</td>
<td>4.2</td>
<td>&lt;0.0001</td>
</tr>
<tr>
<td>NYHA functional class</td>
<td>2.2</td>
<td>0.0298</td>
</tr>
<tr>
<td>LVOT obstruction</td>
<td>12.9</td>
<td>&lt;0.0001</td>
</tr>
<tr>
<td><strong>Systemic AV valve regurgitation (moderate/severe)</strong></td>
<td>2.0</td>
<td>0.0427</td>
</tr>
<tr>
<td><strong>Pulmonary AV valve regurgitation (moderate/severe)</strong></td>
<td>2.3</td>
<td>0.0287</td>
</tr>
<tr>
<td>Mechanical valve prosthesis</td>
<td>74.7</td>
<td>0.014</td>
</tr>
<tr>
<td><strong>Cyanotic heart disease</strong></td>
<td>3.0</td>
<td>&lt;0.0001</td>
</tr>
</tbody>
</table>

LVOT=Left ventricular outflow tract

Drenthen W et al. European Society of Cardiology Congress 2007; September 1-5, 2007; Vienna, Austria.
### Predictors of neonatal complications

<table>
<thead>
<tr>
<th>Variable</th>
<th>Odds ratio</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td>Twin or multiple gestation</td>
<td>5.4</td>
<td>0.0014</td>
</tr>
<tr>
<td>Smoking during pregnancy</td>
<td>1.7</td>
<td>0.0070</td>
</tr>
<tr>
<td>Cyanotic heart disease</td>
<td>2.0</td>
<td>0.0003</td>
</tr>
<tr>
<td>Mechanical valve prosthesis</td>
<td>13.9</td>
<td>0.0331</td>
</tr>
<tr>
<td>Other (cardiac) medication before pregnancy</td>
<td>2.2</td>
<td>0.0009</td>
</tr>
</tbody>
</table>

Drenthen W et al. European Society of Cardiology Congress 2007; September 1-5, 2007; Vienna, Austria.
Incidence of adverse cardiac events late after pregnancy according to pregnancy - related adverse cardiac events

Adverse cardiac events: cardiac death, cardiac arrest, pulmonary edema, sustained symptomatic tachyarrhythmia and/or bradyarrhythmia required treatment or stroke/ TIA

Heart, 2010
Incidence of adverse cardiac events late after pregnancy according to maternal risk score

Risk predictors:
- NYHA > 2/resting cyanosis
- Subaortic/ subpulmonary ventricular dysfunction and/or severe PR
- Left heart obstruction
- Cardiac events during or before pregnancy

Heart, 2010
Absolute contraindications for pregnancy

- **Eisenmenger syndrome** (30% - 50% maternal mortality)
  Termination of pregnancy is safer, preferably with cardiac anesthesia

- Complex cyanotic CHD

- Marfan syndrome with dilated aortic root

- Severe aortic stenosis

- Untreated/ severe coarctation

- Systemic ventricular dysfunction
QUESTION 7

- Combined incidence of congenital heart defects?
- Prevalence of CHD in adults/ kids?
- First time diagnosis of CHD in adults (%)?
- Survival of pts with CHD?
- Mortality: overall; surgical; sudden death?
- Recurrence rate of CHD: parents, siblings?
- **Sex distribution of CHD: overall; type specific lesions?**
- Genetic associations of CHD?
- Most frequent CHD: adults; kids?
- Specific lesions
MEN VS. WOMEN

- Dutch Registry - CONCOR
- 7414 pts
- Average age of pts: 34 y.o men, 36 y.o women
<table>
<thead>
<tr>
<th>Outcome</th>
<th>Odds ratio (95% CI)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Death</td>
<td>0.79 (0.57–1.09)</td>
</tr>
<tr>
<td>Pulmonary hypertension</td>
<td>1.33 (1.07–1.65)</td>
</tr>
<tr>
<td>Systemic hypertension</td>
<td>1.08 (0.88–1.33)</td>
</tr>
<tr>
<td>Aortic outcomes</td>
<td>0.67 (0.50–0.90)</td>
</tr>
<tr>
<td>Cerebrovascular vascular accident/transient ischemic attack</td>
<td>0.88 (0.66–1.18)</td>
</tr>
<tr>
<td>Endocarditis</td>
<td>0.53 (0.40–0.70)</td>
</tr>
<tr>
<td>Pacemaker</td>
<td>0.91 (0.73–1.14)</td>
</tr>
<tr>
<td>Implantable cardioverter defibrillator</td>
<td>0.45 (0.26–0.80)</td>
</tr>
<tr>
<td>Arrhythmia</td>
<td>0.88 (0.77–1.02)</td>
</tr>
</tbody>
</table>

## SEX DISTRIBUTION OF CHD

<table>
<thead>
<tr>
<th></th>
<th>1985</th>
<th>2000</th>
<th>P</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Female</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Prevalence of CHD</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>57%</strong> Female</td>
<td>4.83/1000</td>
<td>4.55/1000</td>
<td>&lt; 0.0001</td>
</tr>
<tr>
<td>Male</td>
<td>3.94/1000</td>
<td>3.61/1000</td>
<td></td>
</tr>
<tr>
<td>All CHD</td>
<td>1985</td>
<td>2000</td>
<td></td>
</tr>
<tr>
<td><strong>Severe CHD</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>1985</td>
<td>0.25/1000</td>
<td>0.41/1000</td>
<td>0.0001</td>
</tr>
<tr>
<td>2000</td>
<td>0.16/1000</td>
<td>0.35/1000</td>
<td></td>
</tr>
</tbody>
</table>

A. Marelli et al., Circulation 2007
### Sex distribution of specific lesion

<table>
<thead>
<tr>
<th></th>
<th>2000</th>
<th>p</th>
<th>1985</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Shunt lesions</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Female</td>
<td>2.13/1000</td>
<td>p</td>
<td>Female</td>
<td>p</td>
</tr>
<tr>
<td>Male</td>
<td>1.46/1000</td>
<td>&lt; 0.0001</td>
<td>1.59</td>
<td>0.93</td>
</tr>
<tr>
<td><strong>ASD, VSD, PDA, AVSD</strong></td>
<td>0.03/1000</td>
<td>0.01</td>
<td>0.05/1000</td>
<td>0.11</td>
</tr>
<tr>
<td><strong>Transp. complex</strong></td>
<td>0.05</td>
<td>&lt; 0.001</td>
<td>0.08</td>
<td>&lt; 0.001</td>
</tr>
<tr>
<td><strong>Coarct.</strong></td>
<td>0.05</td>
<td></td>
<td>0.08</td>
<td></td>
</tr>
</tbody>
</table>
Smoking and CHD

- Largest case control study in the US, tracking 5,000 cases of CHD and the same number of controls in 10 states
- 35,000 – 40,000 infants born every year in the US with CHD
- If all women of reproductive age stop smoking there will be (–) 2,000 kids born with CHD
Smoking and CHD

- Women who are smoking during early pregnancy are 60% more likely to have babies with CHD [(than those who do not smoke) (OR 1.6)]
- Smokers 80% more likely to have babies with septal or right sided defects
- Of women who had kids with CHD, 34% reported they smoked some time from the month prior to conception thru the end of the first trimester (comp. to 25% of women whose children did not have heart problems)
Maternal Diabetes Mellitus and CHD

- DM is known to have a teratogenic effect on the cardiovascular system
- Reported risk of malformation in published studies is 1.7% to 4.0%, but a prospective population-based study of live born infants of diabetic mothers has not previously been reported.
- Recently published study, comparing the prevalence of structural cardiovascular malformations in live babies born to mothers with preexisting diabetes with those born to nondiabetic mothers.
Maternal Diabetes Mellitus and CHD

- Information from all live births between 1995 and 2000 in the former Northern Health Region of England
- 192,618 babies were born, of whom 609 had diabetic mothers
- Congenital structural defects of the heart, including transposed arteries, were found in 22 of the babies with diabetic mothers (3.6%) and in 1417 babies with mothers without diabetes (0.74%)
- The odds ratio for a CV malformation with maternal diabetes was 5.0 (95% CI 3.3 to 7.8).
Maternal Diabetes Mellitus and CHD

- More than 3 times excess of the following heart malformations in babies of diabetic mothers:
  1. Transposition of the great arteries (TGA)
  2. Truncus arteriosus (TA)
  3. Tricuspid atresia (TrA)
- The 5 times excess of cardiovascular malformations overall
- Implies that these specific malformations are perhaps at least 15 times more prevalent [in babies born to diabetic mothers] than in offspring of nondiabetic pregnancies
Question 8

- Combined incidence of congenital heart defects?
- Prevalence of CHD in adults/ kids?
- First time diagnosis of CHD in adults (%)?
- Survival of pts with CHD?
- Mortality: overall; surgical; sudden death?
- Recurrence rate of CHD: parents, siblings?
- Sex distribution of CHD: overall; type specific lesions?
- Genetic associations of CHD?
- Most frequent CHD: adults; kids?
- Specific lesions
# Prevalence of Genetic Problems in CHD

<table>
<thead>
<tr>
<th>Category</th>
<th>Prevalence</th>
</tr>
</thead>
<tbody>
<tr>
<td>Chromosomal abnormalities</td>
<td>4-8% of CHD</td>
</tr>
<tr>
<td>Single gene syndromes (AD, AR, X-linked; high recurrent rate)</td>
<td>1-3% of CHD</td>
</tr>
<tr>
<td>Known teratogens</td>
<td>1-2% of CHD</td>
</tr>
<tr>
<td>Multifactorial (probably)</td>
<td>87 – 94%</td>
</tr>
</tbody>
</table>

Deletions, missense mutations, duplications within a gene

Jeffrey A. Towbin, John Belmont, 2000; Wulfsberg EA et al., 1991
Mary S. Minette and David J. Zahn, 2006
HEART FAILURE IN ACHD PATIENTS

Typical ACHD substrates for late heart failure in ACHD patients

1. Severe AS/ AR BAV and variants, subvalvular or supravalvular pathology, superimposed coarctation
2. Severe congenital mitral stenosis/ regurgitation
3. Uncorrected ASD/ AVSD
4. cCTGA
5. CTGA (after correction)
6. TOF with early - era surgery, long standing shunt, or severe PR
7. Single- ventricle physiology
8. Fontan

ACC/ AHA ACHD Guidelines, 2008
### HEART FAILURE IN ACHD PATIENTS

Other possible pathogenetic factors for heart failure in ACHD patients

1. Prolonged cyanosis
2. Prolonged pressure/ volume overload
3. Poor myocardial intraoperative preservation
4. Large ventricular septal patch or incision/ scar
5. Residual obstruction/ shunts
6. Arrhythmias
7. Obesity

ACC/ AHA ACHD Guidelines, 2008
Other possible factors for heart failure development in ACHD patients

<table>
<thead>
<tr>
<th>1. Acquired valvular disease</th>
<th>2. CAD</th>
</tr>
</thead>
<tbody>
<tr>
<td>3. Systemic hypertension</td>
<td>4. DM</td>
</tr>
<tr>
<td>5. IE</td>
<td>6. Pregnancy</td>
</tr>
<tr>
<td>7. Chronic lung disease</td>
<td>8. Chemotherapy/ mediastinal irradiation</td>
</tr>
<tr>
<td>9. Illicit drug</td>
<td>10. Acquired renal/ liver disease</td>
</tr>
<tr>
<td>11. Sleep apnea</td>
<td>12. Thyroid problems</td>
</tr>
</tbody>
</table>

ACC/ AHA ACHD Guidelines, 2008
1. Thorough evaluation before anticipated surgery
2. Consultation with ACHD expert & cardiac anesthesiologist
3. Moderate & high-risk patients, non-cardiac surgery (even minor) at experienced ACHD center
   - Significant residual defects
   - Fontan physiology
   - Severe pulmonary hypertension, including Eisenmenger
   - Cyanotic CHD
   - Congestive heart failure
   - Valve disease
   - Arrhythmias

ACC/ AHA ACHD Guidelines, 2008
## Noncardiac Surgery

<table>
<thead>
<tr>
<th>High risk</th>
</tr>
</thead>
<tbody>
<tr>
<td>Pulmonary hypertension, primary or secondary</td>
</tr>
<tr>
<td>Cyanotic congenital heart disease New York Heart Association class III or IV</td>
</tr>
<tr>
<td>Severe systemic ventricular dysfunction (ejection fraction less than 35%)</td>
</tr>
<tr>
<td>Severe left-sided heart obstructive lesions</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Moderate risk</th>
</tr>
</thead>
<tbody>
<tr>
<td>Prosthetic valve or conduit</td>
</tr>
<tr>
<td>Intracardiac shunt</td>
</tr>
<tr>
<td>Moderate left-sided heart obstruction</td>
</tr>
<tr>
<td>Moderate systemic ventricular dysfunction</td>
</tr>
</tbody>
</table>

ACC/ AHA ACHD Guidelines, 2008
Heart/ Lung Transplantation

1. Pts with uncorrectable or previously repaired or palliated CHD associated with significant pulmonary vascular disease (ex. single ventricle physiology)
2. Simple cardiac defect (ASD, VSD, PDA) can often be repaired at lung Tx
3. Combined heart – lung TX is most appropriate in the presence of more complex intracardiac abnormalities
4. Chronic cyanosis – fatal hemorrhagic complications
5. Complex anatomy/ vascular problems/ access
6. Previous thoracotomies (less problematic)

ACC/ AHA ACHD Guidelines, 2008
Heart/Lung Transplantation

- Survival after heart Tx
  1. Predicted post Tx half-life (median survival) for the entire cohort of pediatric and adult heart recipients is 10 yrs with a half-life of 13 yrs for those who survive the first yr
  2. Having an ACHD as an indication for Tx increases the risk during the first yr by 2–fold
- Survival after lung Tx: approx. 75% at 1 yr, 60% at 2 yrs
  1. Actuarial survival at 10 yrs after heart/lung Tx is 20%
  2. The outcome for heart/lung Tx is similar to that for lung Tx

ACC/AHA ACHD Guidelines, 2008
Eisenmenger physiology. Cyanosis

1. Patients with Eisenmenger physiology/ cyanosis should avoid situations that could predispose them to clinical deterioration
   - Pregnancy contraindicated
   - Dehydration
   - Moderate and severe strenuous exercise (particularly isometric exercise)
   - Acute exposure to excessive heat
   - Exposure to altitudes greater than 5,000 feet
   - Phlebotomy. Iron deficiency
   - Prolonged flights - precaution measures

ACC/ AHA ACHD Guidelines, 2008
Cardiac conditions associated with the highest risk of adverse outcome from endocarditis for which prophylaxis with dental procedures is reasonable

<table>
<thead>
<tr>
<th>Condition</th>
<th>Congenital Specific Condition*</th>
</tr>
</thead>
<tbody>
<tr>
<td>Previous infective endocarditis</td>
<td>Unrepaired cyanotic CHD, including palliative shunts and conduits</td>
</tr>
<tr>
<td>Prosthetic cardiac valve or prosthetic material used for cardiac valve repair</td>
<td>Completely repaired congenital heart defect with prosthetic material or device, whether placed by surgery or by catheter intervention, during the first 6 months after the procedure†</td>
</tr>
<tr>
<td></td>
<td>Repaired CHD with residual defects at the site or adjacent to the site of a prosthetic patch or prosthetic device that inhibit endothelialization</td>
</tr>
<tr>
<td></td>
<td>Cardiac transplant recipients who develop cardiac valvulopathy</td>
</tr>
</tbody>
</table>

ACC/ AHA ACHD Guidelines, 2008  
AHA Guidelines 2007  
ACC/AHA Guidelines 2008
QUESTION 9

- Combined incidence of congenital heart defects?
- Prevalence of CHD in adults/ kids?
- First time diagnosis of CHD in adults (%) ?
- Survival of pts with CHD ?
- Mortality: overall; surgical; sudden death?
- Recurrence rate of CHD: parents, siblings?
- Sex distribution of CHD: overall; type specific lesions?
- Genetic associations of CHD?
- Most frequent CHD: adults; kids?
- Specific lesions
## Classification system

<table>
<thead>
<tr>
<th>Physiology</th>
<th>Morphology/ ACHD types</th>
<th>Coronary</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Acyanotic</strong></td>
<td>Shunts</td>
<td><strong>ALCAPA</strong></td>
</tr>
<tr>
<td><strong>Cyanotic</strong></td>
<td>Left sided</td>
<td><strong>LVOTO</strong></td>
</tr>
<tr>
<td><strong>Eisenmenger</strong></td>
<td>Right sided</td>
<td><strong>RVOTO</strong></td>
</tr>
<tr>
<td><strong>Systemic RV</strong></td>
<td>Conotruncal</td>
<td></td>
</tr>
<tr>
<td><strong>Fontan</strong></td>
<td>Ebstein anomaly</td>
<td></td>
</tr>
<tr>
<td><strong>Single ventricle variants</strong></td>
<td>“Situs anomalies”</td>
<td></td>
</tr>
<tr>
<td><strong>Simple or complex</strong></td>
<td>Vascular</td>
<td></td>
</tr>
</tbody>
</table>

**Repaired/ palliated/unrepaired**
SHUNT? WHAT ELSE?
Shunt? What else?
## Cyanosis...

<table>
<thead>
<tr>
<th>Cyanosis</th>
<th>Upper extremities</th>
<th>Lower extremities</th>
<th>Examples</th>
</tr>
</thead>
<tbody>
<tr>
<td>Differential cyanosis</td>
<td></td>
<td></td>
<td>- Coarctation</td>
</tr>
<tr>
<td></td>
<td>Red</td>
<td>Blue</td>
<td>- Interrupted aortic arch</td>
</tr>
<tr>
<td>Reversed differential</td>
<td>Blue</td>
<td>Red</td>
<td>- DORV (Taussig - Bing anomaly)</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>- CTGA</td>
</tr>
</tbody>
</table>

**Differential cyanosis:**
- Lt arm mild cyanosis (unoxgenated blood to LSA)
- Right arm cyanosis (PA to right PDA)

**Reversed differential cyanosis:**
- LV blood subpulmonary VSD → PA → PDA → DA
- CTGA (PDA, intact IVS):
- Blood from PA → aorta thru PDA (distal to the LSA)
Summary

• Mortality and morbidity are shifting away from the young and towards the adult

• Workload is increasing

Impact on Health Care System!
Summary

- The **demographics** of CHD population have changed:
  - Increasing number of adults with CHD

- **Distribution** of CHD:
  - Prevalence of pts with **severe CHD** is increasing more rapidly in adults than in children
  - Equalizing numbers of adults and children with severe CHD
Congenital Heart Disease

- Heterogeneous population – the patient profile is changing
- Most pts are not 'fixed'
  - Life-long risk for complications
- Expert care with knowledge and expertise in:
  - Anatomy and physiology
  - Long-term outcome
- Multi-disciplinary team approach in designated centres
Structural heart problems with unique morphology and physiology
1. Shunts
2. Fine balance between pulmonary (& systemic) blood flow
3. Morphological right ventricle (RV) in systemic position
4. Systolic and diastolic RV dysfunction
5. Frequent multiple associated lesions, even relatively simple
   - Sometimes (infrequent), associated genetic/ familial syndromes (including non-cardiac problems)
   - Continuum of disease, from fetal life into adult age with regular adult problems (cardiac and non-cardiac) on top of baseline CHD
   - Significantly modified natural history by multiple surgical/ interventional procedures (with very good results, but nearly never totally repaired/completely healed)
CONGENITAL HEART DISEASE (CHD) IT IS A STORY ABOUT:

- Continuously growing adult population
- Good survival rate but substantial morbidity and mortality, especially in pts with moderate/ high risk and complex lesions (including non-cardiac ones)
  1. CHF
  2. Cyanosis
  3. Pulmonary arterial hypertension
  4. Arrhythmias/ EP treatment
  5. Infective endocarditis
  6. Non-cardiac interventions
- Reasonably good tolerance of pregnancy, in general
- Not negligible recurrence risk
Patients with **named conditions or operations** are complex

Specialized centre
ACHD TSUNAMI: 

Catch The WAVE!

- Arrhythmias
- Heart Failure
- Pulmonary Hypertension
- Re-Intervention
- Psychosocial Issues
- Reproduction
- Premature Death
CATCH 22

- **Cardiac disease**
- **Abnormal facies**
- **Thymic hypoplasia**
- **Cleft palate**
- **Hypocalcemia**
- **22q11 del. (mid 1990’s)**

**Figure 1**: Gene/EST map of the D22S1933 interval in 22q11. Genes and ESTs (triangles) are indicated to the left and right, respectively, of the line representing the 22q11 region. TBX1 is a member of the T-box family of transcription factors (Cheffer et al. 1996). “COMT” denotes the catechol-O-methyl transferase gene (Grossman et al. 1992). The genetic markers, D22S1638, D22S941, D22S1648, D22S944, and D22S1623 are indicated (squares). The critical region is boxed.

C Carlson 1997
MULTIPLE NAMES - THE SAME SYNDROME

- IAA, TA, TOF & parathyroid and thymic hypoplasia: classic findings of DiGeorge syndrome (DGS)

- Conotruncal defects (esp. with misalignment VSD) & velopharyngeal insufficiency, learning difficulties: more common in velo-cardiofacial syndrome (VCFS) and conotruncal anomaly face syndrome (CAFS)

- Cayler cardiofacial, Opitz G/BBB, Shprintzen, Sedlackova (syndromes) etc.

Bettina F. Cuneo, 2001
22Q11.2DEL

- 90% pts with full “DGS phenotype”
- 70% pts with “VCFS”
- 15% pts with isolated conotruncal defect
- De novo cases approx. 90%

- Inherited – approx. 10% (parents more mildly affected than kids and CHD are rare)

- Recurrence risk approx. 50% (AD)
DIAGNOSIS

- Microdeletion of 22q 11 (submicroscopic)
  - 1.5 – 3 MB deletion: dozens of genes and multiple repeated sequences

- No single gene mutation as a case of the syndrome

- **FISH test** highly specific
  - Approx. 90% pts - FISH positive
  - Subgroup (approx. 10%) pts - FISH negative
    * small amount of deletion or (an)other syndrome/s
An example of a normal FISH test for a balanced structural rearrangement

Chromosome 4 telomere - red
Chromosome 22 - yellow
Chromosome 22 telomere - green

www.givf.com
Figure 1. Idiogram of chromosome 22 illustrating the commonly deleted region (DGCR, hatched box) and the approximate location of the probes used for deletion analysis. Probes for loci D22S75 (N25) and D22S66 (160b) lie within the DGCR. Cos 82 and pH17, the control probes, map to the distal long arm of chromosome 22.
### TESTS FOR DIAGNOSIS OF 22Q11 DEL

<table>
<thead>
<tr>
<th>Test method</th>
<th>Mutations detected</th>
<th>Mutation detection rate</th>
<th>Test availability</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>FISH</strong></td>
<td>Deletion 22q11.2 DGCR</td>
<td>&gt; 95%</td>
<td>Clinical testing</td>
</tr>
<tr>
<td><strong>Direct DNA</strong></td>
<td>Smaller 22q11 del or point mutation</td>
<td>&lt; 5%</td>
<td>Research only</td>
</tr>
</tbody>
</table>

*Mutation analysis, scanning sequence analysis, molecular genetic testing*

OMIM. PubMed
SCREENING FOR 22Q11 DEL

- CHD – conotruncal
- Learning difficulties or mental retardation
- Characteristic facies
- Hypernasal speech and/or palatal abnormalities
- History of immune problem (thymic problem)
- History of hypoparathyroidism/ hypocalcemia

A. Bassett, E. Chow, 1999
OTHER CONGENITAL PROBLEMS IN 22Q11 DEL

- Blood disorders: bleeding, coagulopathies
- Immune disorders: infections
- Parathyroid: hypocalcemia
- Renal
- Hernias
- Talipes
- Orthopedic
- ENT
- Ophthalmologic
- Mental retardation/ Learning diffic./ Speech dist./ Developmental…

Modified from A. Bassett, E. Chow, 1999
### WHICH ADULT SHOULD BE SCREENED?

<table>
<thead>
<tr>
<th>Commonly associated cardiovascular lesions</th>
<th>Extracardiac manifestations</th>
</tr>
</thead>
<tbody>
<tr>
<td>TOF</td>
<td>Psychiatry history</td>
</tr>
<tr>
<td>PA-VSD</td>
<td>Mental retardation</td>
</tr>
<tr>
<td>TA</td>
<td>Nonverbal learning disability</td>
</tr>
<tr>
<td>IAA (typeB)</td>
<td>Dysmorphic facial features</td>
</tr>
<tr>
<td>Isolated arch anomalies</td>
<td>History of cleft palate</td>
</tr>
<tr>
<td></td>
<td>Hypernasal speech</td>
</tr>
<tr>
<td></td>
<td>Hypocalcemia</td>
</tr>
</tbody>
</table>

Modified from Luc M Beauchesne et al., 2005
WHICH ADULTS SHOULD BE SCREENED?

- Conotruncal heart defects (TOF & co)
- Global dysmorphic facies
- Voice abnormalities (eg. hypernasality)
- History of learning difficulties
- Young age (esp. < 30 y.o.)

Personal communication. Dr. A. Bassett, 2006
Who should be screened?

Modified from Luc M Beauchesne et al., 2005
CONCLUSIONS

- The incidence and prevalence of 22q 11del syndrome ranging from 1:2000 to 1:6000

- Significant clinical overlap with other genetic syndromes; genetic testing important in some cases (22q11del)

- Multisystemic disease required multidisciplinary team involved in the care of this population

- Important implications for surgical treatment and management

- Awareness required
1. **ACC/AHA 2008 Guidelines for the Management of Adults With Congenital Heart Disease** (JACC, 2008)

2. **Canadian Cardiovascular Society (CCS) 2009 Consensus Conference on the management of adults with congenital heart disease.** (Canadian Journal of Cardiology, 2010)

3. Proceedings of the 32-nd Bethesda Conference: Care of the Adult with Congenital Heart Disease (JACC, 2001)

Guidelines

1. Congenital Cardiac Defects Committee of the AHA Section on Cardiovascular Disease in the Young. Collaborative care for adults with congenital heart disease, Circulation, 2002)


Guidelines


2. AHA Scientific Statement … Indications for Heart Transplantation in Pediatric Heart Disease (Circulation, 2007)

3. 2008 Focused Update Incorporated into the ACC/ AHA 2006 Guidelines for the Management of Patients with Valvular Heart Disease

Management of Congenital Valvular Heart Disease in Adolescents and Young Adults; Management of Valvular Disease in Pregnancy
Questions? Uncertainties?

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