Anomalies of the Fetal Heart

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Abnormal Cardiac Chambers

- Ebstein anomaly
- Tricuspid atresia
- Cardiac tumors
Anomalies of the Outflow Tracts

- Tetralogy of Fallot
- Common Arterial Trunk
- Double Outlet Right Ventricle
Ebstein Anomaly
Ebstein Anomaly

- Apical displacement of septal and posterior leaflets of tricuspid valve
- Part of right ventricle (RV) is atrialized
- Dysplastic tricuspid valve
- Severe tricuspid regurgitation

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Ebstein Anomaly

- Wide spectrum
- Minor form to severe form
- Associated anomalies:
  - Pulmonary stenosis or atresia
  - VSD
  - ASD
Ebstein Anomaly

Spectrum of Disease
Ebstein Anomaly

Ultrasound Findings

• Enlarged heart
• Dilated RA
• Attachment of TV septal leaflet to RV wall (essential for diagnosis)
• Paradoxic septal movement
• RV outflow obstruction
Cardio-thoracic Ratio

Ebstein Anomaly

- 0.6 associated with pulmonary hypoplasia
Ebstein Anomaly
Ebstein Anomaly
Ebstein Anomaly

Ultrasound Findings

• Holosystolic TR
• Jet originates deep in RV
• PSV > 175 cm/sec
Ebstein Anomaly

Color Doppler
Ebstein Anomaly
Ebstein Anomaly
Ebstein Anomaly
Ebstein Anomaly

Pulsed Doppler
Ebstein Anomaly

Early Gestation
Ebstein Anomaly

Associated Anomalies

• RV outflow obstruction in 60%
• ASD in ~ 60%
• SVT common post repair
• Most are isolated

Br Heart J 1974;36:417–427
Ebstein Anomaly

*Poor Prognostic Factors (fetus)*

- Massive Cardiomegaly
- Decreased RV outflow
- Fetal hydrops
Outcome (fetal series):

- 45% dying in utero
- Overall 80-90% mortality

Tricuspid Atresia
Tricuspid Atresia

- Absence of a right atrioventricular connection
- Diminutive right ventricle (RV)
- Widely patent foramen ovale
- Ventricular septal defect (VSD)
- Right ventricular outflow obstruction

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Tricuspid Atresia

- Inlet-type VSD always present
- Size of RV is related to size of VSD
- Widely patent foramen ovale or ASD
- Flow to RV thru VSD in late diastole
Tricuspid Atresia

Ultrasound Findings

- Small RV, normal contractility
- No RV myocardial thickening
- Thickened echogenic TV
- Slightly dilated RA
- Large foramen ovale
- Malaligned atrial and ventricular septae
Tricuspid Atresia
Tricuspid Atresia
Tricuspid Atresia
Tricuspid Atresia
# Tricuspid Atresia

<table>
<thead>
<tr>
<th>Type</th>
<th>Prevalence</th>
<th>Great arteries</th>
</tr>
</thead>
<tbody>
<tr>
<td>Type 1</td>
<td>70 – 80 %</td>
<td>Normal</td>
</tr>
<tr>
<td>Type 2</td>
<td>12 – 25 %</td>
<td>D-Transposition</td>
</tr>
<tr>
<td>Type 3</td>
<td>Rare</td>
<td>CAT, L-Transposition</td>
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</tbody>
</table>
Cardiac Tumors
Cardiac Tumors

- Rhabdomyomas (80-90%)
- Teratomas
- Fibromas
- Myxomas
- Rhabdomyosarcomas
- Hamartomas
- Others
Rhabdomyomas
Rhabdomyomas
Rhabdomyomas
Rhabdomyomas
Tuberous Sclerosis
Tetralogy of Fallot

Subaortic malaligned VSD

Overriding dilated aortic root

Narrow stenotic PA

RV hypertrophy is not present in fetus

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Tetralogy of Fallot

- Incidence ~ 1 in 3600 live births
- Accounts for 3-7 % of infants with CHD
Tetralogy of Fallot

- Classic form (~ 80 %)
- Pulmonary atresia with VSD
- Absent pulmonary valve
Tetralogy of Fallot

Ultrasound Findings:

- Four chamber view normal
- Left axis deviation
Tetralogy of Fallot

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Tetralogy of Fallot
Tetralogy of Fallot

Ultrasound Findings:

- Five chamber view abnormal
- Aortic dextroposition
- Dilated aortic root (3rd trimester)
- Perimembranous subaortic VSD
- Infundibular pulmonary stenosis
Tetralogy of Fallot

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Tetralogy of Fallot
Tetralogy of Fallot

5-Chamber View
Tetralogy of Fallot
Tetralogy of Fallot
Tetralogy of Fallot

A Normal

B TOF
Tetralogy of Fallot

5-Chamber View

3-Vessel View

LV, RV, AO, VSD, PA, AO, RV, RA
Tetralogy of Fallot
Tetralogy of Fallot

3-Vessel Trachea View
Tetralogy of Fallot - PA

3-Vessel Trachea View
Tetralogy of Fallot

STIC – TUI in Color Doppler
Tetralogy of Fallot

STIC-Glass Body in Color Doppler
Tetralogy of Fallot

Common Associated Cardiac Anomalies

- Patent foramen ovale/ ASD in 85%
- Right sided Aortic arch in 25%
- Persistent LSVC in 11%
Tetralogy of Fallot

**Rare Associated Cardiac Anomalies**

- Atrioventricular canal defect in < 5%
- Abnormal coronary circulation in < 5%
- Anomalous pulmonary venous connections in < 1%
Tetralogy of Fallot

Associated Extracardiac Anomalies

• Chromosomal abnormalities in 30%
• Anomalies of anatomic organs, common
• Deletion 22q11 in 10-15%
  • Right Aortic arch
  • Thymic hypopgenesis / agenesis
• Extracardiac anomalies
• Polyhydramnios
Poor Prognostic Factors

- Decelerated growth of the PA
- Accelerated growth of the Ao
- Cessation of forward flow in PA
- Reversed flow in DA
- TOF with pulmonary atresia
- Absent pulmonary valve
- Associated chromosomal anomalies
- Associated extracardiac anomalies
- Small LV
- Associated abnormal venous connections
Common Arterial Trunk (CAT)

- Ventricular septal defect
- Single arterial trunk (CAT)
- PA originates from CAT
- Absent ductus arteriosus

CAT gives rise to systemic, coronary and pulmonary circulations

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Common Arterial Trunk

- Incidence ~ 1.6 % of newborns with CHD
- Occur in 1.07 of 10,000 live births
- More common in fetuses of diabetic mothers
Common Arterial Trunk

- Truncus arteriosus
- Truncus arteriosus communis
- Aorticopulmonary trunk

Classification is based upon origin of pulmonary arteries
Common Arterial Trunk

- Type 1: Main PA arises from CAT (A1)
- Type 2: Direct origin of RPA and LPA from CAT, close anatomically (A2)
- Type 3: Direct origin of RPA and LPA from CAT, distant anatomically (A2)
- Type 4: RPA and LPA arise from aortic arch or descending aorta (PA with VSD)

A3: single PA – A4: Aortic arch abnormalities

Collet & Edwards 1949 – Van Praagh 1965
Common Arterial Trunk

CAT Type A4

Van Praagh & Van Praagh, 1965
Common Arterial Trunk

Origin of CAT

• Biventricular origin in 2/3 of cases
• RV origin in 1/3 of cases
• LV origin in rare cases
Common Arterial Trunk

Valves of CAT

- Three leaflets in 69%
- Four leaflets in 22%
- Two leaflets in 9%
- Five, six or more in rare cases

Thickened with regurgitation and occasional stenosis
Common Arterial Trunk

- Four chamber view normal
- Cardiac levorotation
- Five chamber view abnormal
- Overriding large vessel
- Thickened dysplastic valves
- Absent PA from RV
Common Arterial Trunk

Identifying origin of PA from the CAT confirms the diagnosis
Common Arterial Trunk

4-Chamber View
Common Arterial Trunk

4-Chamber View

5-Chamber View
Common Arterial Trunk

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Common Arterial Trunk

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Common Arterial Trunk

Dysplastic Valves of CAT

Insufficiency
Common Arterial Trunk

Valves of CAT
Common Arterial Trunk
Common Arterial Trunk
Common Arterial Trunk

3-Vessel Trachea View
Common Arterial Trunk

3-Vessel Trachea View
Common Arterial Trunk

GA=13w2d

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Common Arterial Trunk

3-D reconstructed plane in power Doppler
Common Arterial Trunk

Type 1 (A1)

AO
PA
CAT
RV
LV

3-D reconstructed plane in inverse mode
Common Arterial Trunk

Type A4 with IAA

3-D reconstructed plane in inverse mode
Common Arterial Trunk

Common Associated Cardiac Anomalies

- Absent DA in 50%
- When present, DA is patent postnatally in 70%
- Right Aortic Arch in 20-30%
- Interrupted Aortic Arch in 15%
- Absence of one of branch PA in 16%
- Variation in coronaries in 30%
Common Arterial Trunk

Associated Extracardiac Anomalies

- Common and seen in ~ 40% of CAT
- Numerical chromosomal anomalies in 4.5%
- 22q11 deletion in 30-40%
- Reported in diabetic mothers
# Common Arterial Trunk

## Differential Diagnosis

<table>
<thead>
<tr>
<th></th>
<th>CAT type 1</th>
<th>TOF</th>
</tr>
</thead>
<tbody>
<tr>
<td>Malaligned ventricular</td>
<td>Present</td>
<td>Present</td>
</tr>
<tr>
<td>septal defect and aortic override</td>
<td>Markedly dilated</td>
<td>Present</td>
</tr>
<tr>
<td>Aortic root size</td>
<td>Arising from the common arterial trunk</td>
<td>Normal to dilated</td>
</tr>
<tr>
<td>Pulmonary trunk</td>
<td>No pulmonary trunk arising from ventricle</td>
<td>Narrow, separately arising from ventricle with patent pulmonary valve</td>
</tr>
<tr>
<td>Ductus arteriosus</td>
<td>Absent in 50%</td>
<td>Narrow, antegrade flow</td>
</tr>
<tr>
<td>Aortic valve/trunkal</td>
<td>Valve with one to six leaflets</td>
<td>Normal aortic valve</td>
</tr>
<tr>
<td>valve</td>
<td>Often dysplastic and insufficient</td>
<td>No regurgitation</td>
</tr>
<tr>
<td>Chromosomal aberrations</td>
<td>22q11 deletion in 30%-40%, other trisomies in 4%-5%</td>
<td>22q11 deletion in 10%-15%, other trisomies in 30%</td>
</tr>
<tr>
<td>Prognosis in postnatal isolated cases</td>
<td>Good</td>
<td>Good to excellent</td>
</tr>
<tr>
<td></td>
<td>Reoperations of pulmonary conduit required</td>
<td></td>
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</tbody>
</table>
Common Arterial Trunk

Outcome of Fetal Series

<table>
<thead>
<tr>
<th>Compiled data</th>
<th></th>
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</thead>
<tbody>
<tr>
<td>Number of cases</td>
<td>87</td>
</tr>
<tr>
<td>Pregnancy termination (%)</td>
<td>34 (39%)</td>
</tr>
<tr>
<td>In utero deaths</td>
<td>4 (4.5%)</td>
</tr>
<tr>
<td>Live births</td>
<td>48 (55%)</td>
</tr>
<tr>
<td>Neonatal and infant deaths (%)</td>
<td>20 (23%)</td>
</tr>
<tr>
<td>Survival in all cases (%)</td>
<td>28 (32%)</td>
</tr>
<tr>
<td>Survival in continuing cases (%)</td>
<td>28/52 (53%)</td>
</tr>
<tr>
<td>Survival in all liveborns (%)</td>
<td>28/48 (58%)</td>
</tr>
</tbody>
</table>

Pediatr Cardiol 2009;30:256
Common Arterial Trunk

Outcome of Neonatal Series

Study involving 50 infants with CAT, operated on from 2 days to 6 months of age, Actuarial survival of 96% at 3 years was reported.

J Thorac Cardiovasc Surg 2000;119:508
Double Outlet RV

“DORV is a type of ventriculoarterial connection in which both great vessels arise either entirely or Predominantly from the right ventricle”

Congenital Heart Surgery Nomenclature and Database Project – Ann Thorac Surg 2000
Double Outlet RV

- Ventricular septal defect
- Great arteries arise from RV
- Varying relationship of great arteries

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Double Outlet RV

- Incidence ~ 1 – 1.5 % of newborns with CHD
- Occur in ~ 1 of 10,000 live births
- More common in fetuses of diabetic mothers
Double Outlet RV

- Spatial relationship of great arteries
- Location of VSD
- Presence of pulmonary & aortic stenosis

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### Double Outlet RV

**Spatial relationship of great arteries**

<table>
<thead>
<tr>
<th>Relationships of great arteries in DORV</th>
<th>Description</th>
</tr>
</thead>
</table>
| Right posterior aorta to the pulmonary artery (tetralogy of Fallot-type DORV) | • Rare form of DORV  
• Normal relationship of great arteries |
| Right anterior aorta to the pulmonary artery (D-transposition-type DORV) | • Second most common type of DORV  
• VSD either subaortic or subpulmonary  
• Subgroup called Taussig-Ring form of DORV |
| Left anterior aorta to the pulmonary artery (L-transposition-type DORV) | • Rare form of DORV  
• Left course of the aorta in the thorax  
• VSD either subaortic or subpulmonary |
| Right lateral aorta to the pulmonary artery (side by side) | • Most common form of DORV  
• Aorta to the right of pulmonary artery  
• Subaortic-type VSD is most common |

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### Double Outlet RV

#### Anatomic positions of VSD

<table>
<thead>
<tr>
<th>Anatomic positions of VSD</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Subaortic type</td>
<td>• VSD located closer to the aortic valve than the pulmonary valve</td>
</tr>
<tr>
<td></td>
<td>• Most common type</td>
</tr>
<tr>
<td>Subpulmonary type</td>
<td>• VSD located closer to the pulmonary valve than the aortic valve</td>
</tr>
<tr>
<td></td>
<td>• Typically supraventricular in location</td>
</tr>
<tr>
<td></td>
<td>• Second most common type</td>
</tr>
<tr>
<td>Subaortic and subpulmonary type (doubly committed)</td>
<td>• Large VSD</td>
</tr>
<tr>
<td></td>
<td>• VSD closely related to both semilunar valves</td>
</tr>
<tr>
<td></td>
<td>• Rare type</td>
</tr>
<tr>
<td>Remote type (nonrelated)</td>
<td>• VSD is distant from and nonrelated to both semilunar valves</td>
</tr>
</tbody>
</table>

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Double Outlet RV
Double Outlet RV

Oblique plane (5-chamber and 3-VT)
Double Outlet RV

Oblique plane (short axis)
Double Outlet RV

Oblique plane (short axis)
Double Outlet RV

Oblique plane (5-chamber and 3-VT)
Double Outlet RV
Double Outlet RV

Oblique plane (14 weeks)
Double Outlet RV

3-D reconstructed plane in inverse mode
Double Outlet RV

Common Associated Cardiac Anomalies

- Pulmonary stenosis in 70%
- Mitral atresia
- Atrial septal defects
- Aortic stenosis
- Aortic coarctation
- Persistent LSVC
- Venous anomalies / heterotaxy
Double Outlet RV

Associated Chromosomal Anomalies

- Common, range of 12-40%
- Trisomy 18, 13 & 22q11
# Double Outlet RV

## Prognostic Factors

<table>
<thead>
<tr>
<th>Cardiac findings</th>
<th>Good prognosis</th>
<th>Poor prognosis</th>
</tr>
</thead>
<tbody>
<tr>
<td>Aortic arch</td>
<td>Normal-sized aortic arch</td>
<td>Tubular aortic arch hypoplasia</td>
</tr>
<tr>
<td>Pulmonary artery</td>
<td>Patent pulmonary artery</td>
<td>Pulmonary atresia</td>
</tr>
<tr>
<td>Ventricle</td>
<td>Normal-sized ventricles</td>
<td>Hypoplastic left ventricle</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Single ventricle anatomy</td>
</tr>
<tr>
<td>Atrioventricular valve</td>
<td>Normal formed atrioventricular valves</td>
<td>Mitral atresia</td>
</tr>
<tr>
<td>anatomy</td>
<td></td>
<td>Atrioventricular septal defect</td>
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<tr>
<td>Situs</td>
<td>Normal situs</td>
<td>Situs ambiguous</td>
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