Guideline
Tetralogy of Fallot

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Guideline Tetralogy of Fallot

Why was this guideline chosen?

- most frequent cyanotic congenital heart disease (2.5% of all CHD)

- untreated high mortality (10 years survival 24%)


Bertranou EG, Am J Cardiol 1978;42:458
**Important feature**

anterior and superior deviation of the outlet septum resulting in

- subvalvar pulmonary stenosis with ? hypoplasia of PA- valve and pulmonary arteries
- non-restrictive malignment subarterial VSD
- overriding aorta
- RV- hypertrophy

Michel Rigby, London 1994
The severity of RVOTO determines the extent of R-L shunting on ventricular level and therefore the extent of cyanosis.

- **Severe RVOTO**
  - results in deep cyanosis due to a large R-L shunt

- **Mild RVOTO**
  - does not cause cyanosis ("pink Fallot") and may result in heart failure due to large L-R shunting on ventricular level
Associated anomalies

- secundum atrial septal defect
- **right aortic arch (25%)**
- major aorto-pulmonary collaterals (MAPCAS)
- complete AVSD (frequently associated with trisomy 21)
- anomaly of coronary arteries with e.g. anomalous LAD from RCA crossing the RVOT (5%)
Di George Syndrome
micro deletion 22q11.2
- in 9-17% of all uncomplicated Tetralogy of Fallot (TOF)
- in 60-70% in patients with TOF and right aortic arch

Recommendation
FISH analysis for micro deletion 22q11.2
in all patients after consent
of the parents (strong consensus)
Important findings!

• systolic murmur
  - caused by PS
  - intensity inversely correlated with severity of PS
  - VSD no murmur!

• central cyanosis
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Diagnosis

Echocardiography is cornerstone for the diagnosis. Following findings are important to record:

• **Anatomy**
  - malalignment subarterial VSD
  - override of aorta and anomalies of aortic arch
  - *RVOTO with subvalvar and valvar PS*
  - *bifurcation of PA with origin of RPA and LPA*
  - unifocal or multifocal PA- perfusion
  - coronary arteries

• **Function**
  - Doppler gradient of RVOTO and branch pulmonary arteries
Differential diagnosis

- pulmonary atresia with VSD
- absent pulmonary valve syndrome
- DORV (aorta > 50% from RV, discontinuity between aortic and mitral valve)
Diagnostic cardiac catheterisation is usually not indicated (strong consensus)

**Indication**

- suspicion of
  - anomalous origin and/or perfusion of pulmonary arteries
  - anomalous coronary artery crossing RVOT
- after previous palliation

origin of LPA from ascending aorta

origin of LAD from RCA
Necessary investigations

• at each clinical visit
  - pulse oxymetry: if repeatedly pO₂-saturation < 80%
    treatment mandatory!

• once pre-op
  (as reference for post-op follow-up examinations)
  - chest X-ray
  - ECG

Usually not indicated

• MRI/ CT-scan
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Emergency: cyanotic spells

**Cause**
- acute obstruction of RVOT and/or
- acute decrease of peripheral resistance

**Findings**
- increase of cyanosis
- tachycardia
- seizures (neurological sequels!)
- auscultation
decreased intensity of systolic heart murmur
due to reduced flow into PA
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Cyanotic spells

1. Emergency treatment

- increase of peripheral resistance (knee-to-chin position)
- administration of oxygen
- sedation
  - morphine s.c./i.v. 0.1-0.2 mg/kg
  - midazolam i.v. 0.1 mg/kg
  - rectal/nasal 0.5 mg/kg
  - ketamine i.v. 1-2 mg/kg
  - i.m. 5 mg/kg
- administration of volume
  (i.v. bolus of 10 ml/kg e.g. NaCl 0,9%, can be repeated)
- admission to ICU with
  - intubation
  - i.v. noradrenaline
  - i.v.ß-blocker e.g. esmolol 50-200 µg/kg/min

2. Subsequent treatment

- oral propranolol (2-6 mg/kg/d)
- no delay of surgical or interventional treatment
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Treatment options

Corrective surgery
• enlargement of RVOT and
• VSD closure

Time of corrective surgery
• elective at the age 4 - 12 months
• in neonates possible but higher morbidity and mortality than in infants > 4 months

Palliation
• required if corrective surgery is not possible in severely cyanosed infants to improve the perfusion of the lungs
1. **Enlargement of RVOTO depends on**
   - if diameter of PA- valve < -2 SD
     - resection of infundibular stenosis a. ?valvotomy
     - usually no transannular patch
   - if diameter of PA- valve > -2SD
     - usually transannular patch

To avoid significant pulmonary valve insufficiency
a residual RVOT gradient is accepted

2. **VSD closure**
   - via right ventriculotomy or transatrial approach
Primary corrective surgery not be feasible due to e.g.
- hypoplastic pulmonary arteries
- significant co- morbidity
- prematurity
- abnormal coronary artery crossing the RVOT

Possibilities of palliation
- surgically
  - systemic- pulmonary shunt (e.g. BT- shunt)
  - enlargement of RVOT
- interventional cardiac catheterisation
  - balloon valvuloplasty
  - PDA stenting
  - stent in RVOT

→ No agreement which type of palliation is preferable
**Palliative treatment** is performed in our centre..

(only one answer per centre)

- never \(N=1\)
- always surgical palliation \(N=6\)
- always catheter intervention \(N=3\)
- both catheter intervention and surgical palliation \(N=13\)

→ most centres offer surgical as well as interventional palliation

→ only 1 centre always performes corrective surgery

Survey by H. Bertram 2011
During the last 5 years our centre performed **interventional palliation** in patients with severe RVOTO as … (up to 3 answers per centre)

<table>
<thead>
<tr>
<th>Option</th>
<th>Cases</th>
</tr>
</thead>
<tbody>
<tr>
<td>never</td>
<td>N=1</td>
</tr>
<tr>
<td>only balloon valvuloplasty</td>
<td>N=17</td>
</tr>
<tr>
<td>RVOT stenting, too</td>
<td>N=11</td>
</tr>
<tr>
<td>ductal stenting, too</td>
<td>N=12</td>
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</tbody>
</table>

→ many centres do catheter interventions in pts. with severe RVOTO
→ balloon valvuloplasty in pts. with severe RVOTO is quite common (17/22)
→ stenting the RVOT or the arterial duct has been performed in ~ 50 % of centres

Survey by H. Bertram 2011
Experience in Toronto

- 11 symptomatic young infants
- median valve diameter 3.7 mm (Z-score – 6.7)
- implantation of coronary stent
  - no procedural complications
  - median increase in saturation from 73% to 94%

G. Dohlen, Heart 2009;95:142-147
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Possibilities of Palliations

- various palliations are feasible
- a preference cannot be given → decision of each cardiac centre

- severe hypoxaemia
- severe RVOTO
- hypoplastic pulmonary arteries
- severe RVOTO
- significant accompanying comorbidities*

- no clinical stabilization: tcSO2 < 80 %
or
- hypoxaemic spells

- clinical stabilization: tcSO2 > 85%
- no hypoxaemic spells

balloon valvuloplasty
RVOT stenting
ductal stenting
surgical palliation

prostaglandin E

- early corrective surgery
- outpatient visits every 2-3 weeks; preoperative angiography

corrective surgery performed electively at age 4-12 months
Important complications
• arrhythmia
• pulmonary valve insufficiency

Follow-up examinations
regular (once a year) and life-long with:
• ECG (QRS width > 180 msec or increase > 3.5 msec/year)
• echocardiography
• Holter ECG (every 3 y)
• MRI (if RV volume loading due to pulmonary insufficiency)
• cardio-pulmonary exercise test (every 5 y in patients > 10 y of age)
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Untreated patients

• signs of chronic hypoxemia

• risks
  (increasing with age, haemoglobin, iron deficiency)
  - endocarditis
  - brain abscess
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Prevention
• not possible

Affected families
• genetic counselling
• fetal echocardiography

Recommendation
• delivery of the child with prenatally diagnosed TOF in a hospital with a Department of Paed. Cardiology/Surgery

Fetus with TOF
<table>
<thead>
<tr>
<th>Beteiligte Fachgesellschaften / Organisationen</th>
<th>Vertreter / Experte</th>
</tr>
</thead>
</table>
| Deutsche Gesellschaft für Pädiatrische Kardiologie | Prof. Dr. med. Jochen Weil (Sprecher), Hamburg  
Prof. Dr. med. Sven Dittrich, Erlangen  
Prof. Dr. med. Peter Ewert, Berlin  
PD Dr. med. Nikolas Haas, Bad Oeynhausen  
Prof. Dr. med. Thomas Paul, Göttingen  
Prof. Dr. med. Angelika Lindinger, Homburg  
PD Dr. med. Alfred Hager, München  
Priv. Doz. Dr. med. Carsten Rickers, Kiel  
PD Dr. med. Harald Bertram, Hannover |
| Deutsche Gesellschaft für Thorax-, Herz- und Gefäßchirurgie | Dr. Alexander Horke, Stuttgart  
PD Dr. Jörg Sachweh, Duisburg |
| Bundesverband Herzkranker Kinder e. V. | Dr. Dr. Sicco Henk van der Mei, Giessen |
| Arbeitsgemeinschaft Niedergelassener Kinderkardiologen | Dr. med. Karl-Robert Schirmer, Hamburg  
Dr. med. Marc Schlez, Neustadt |
| Arbeitsgemeinschaft der an allgemein-pädiatrischen Kliniken tätigen pädiatrischen Kardiologen | Dr. Jörg Franke, Kempten  
Dr. med. Irene Ruschke, Chemnitz |
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