Marfan Syndrome

Cardiovascular Manifestations

- Most common: progressive aortic root dilatation
- Mitral valve prolapse and regurgitation

Progressive root enlargement (Marfan's)

Jan '04
Nov '04
Sep '05

41 yo man
Marfan's

ECG gating

Marfan Syndrome

- Incidence 1:5000
- Clinical Diagnosis based on Ghent Criteria
- Most often a defect in FBN1 gene
  - Codes for fibrillin-1 protein
  - Molecular testing neither sensitive nor specific
- 25% without family history and represent new mutations
Clinical 3D and 4D Imaging of the Thoracic Aorta
In Marfan’s Disease

ROLE OF RADIOLOGY

MODERN COMPUTED TOMOGRAPHY (CT)
- CT-Angiography (CTA)
- 3D and ‘4D’ CT Angiography (EKG-gated CTA)

3D and 4D VISUALIZATION
- diagnosis, surveillance
- treatment planning
- education

ECG gated CTA of the Thorax
(16-channel MDCT)

‘Gated Chest’
- entire thoracic aorta
- not thinnest collimation (1.25mm)
- no beta-blockers, no subling. nitro
- recon. 10 phases (0–90% of RR interval)
- no/limited ECG-pulsing (30-75%)

Coronary CTA
- heart
- thinnest collimation (0.625mm)
- beta-blockers, and subling. nitro
- recon. 1 diastolic phase (65% of RR)
- ECG pulsing on (dose reduction)

‘gated Chest’ with ECG-tube current modulation

diastole CINE 10 phases systole

ECG-pulsing (30-70%), 20% dose

s/p Ross procedure (pulmonary- to aorta autograft)
susp. leak/pseudoaneurysm

s/p Ross procedure (pulmonary- to aorta autograft)
susp. leak/pseudoaneurysm

diastole systole
cine loop
64 channel ECG gated CTA + coronary of the Thorax

‘Gated Chest’  Coronary CTA
• entire thoracic aorta  • heart
• not thinnest collimation (1.25mm)  • thinnest collimation (0.625mm, 0.75mm)
• no beta-blockers, no subling. nitro  • beta-blockers, and subling. nitro
• recon. 10 phases (0–90% of RR interval)  • recon. 1 diastolic phase (65% of RR)
• no/limited ECG-pulsing (30–75%)  • ECG pulsing on (dose reduction)

Stanford Indications for (routine) ECG gated CTA of the Chest
• preoperative evaluation of patients with aortic root aneurysm
• postoperative (pre-discharge) evaluation of patients with aortic root aneurysms
• acute aortic syndromes

NOT indicated: routine surveillance / follow-up of aortic root aneurysms, chronic dissection
→ echocardiography, MRI

3D and 4D Pre- and Postop. CT of the Thoracic Aorta

Learning Objectives

TECHNICAL
• 3D/4D CT data acquisition (EKG-gated CTA)
• 3D/4D Clinical Visualization
  Stanford setup (Client-server solution)

CLINICAL
• aortic root surgical anatomy / pathology
• pre- / post-operative aortic root imaging

68 y.o. man
• w. cervical arch, pseudocoarctaion
• large left sinus–of–Vals. aneu
• Coronary artery disease ?

short LAD occlusion

w. cervical arch, pseudocoarctaion
large left sinus–of–Vals. aneu
Coronary artery disease ?
**Interactive 3D/4D Interpretation**

**Server/Client System**

(*TeraRecon, San Mateo, CA*)

**Server** does all the rendering ('big box')
- up to 4 dedicated rendering boards, can handle 28,000 images in real time
- up to 20 clients can log in

**Thin Client** (small program)
- communicates with server
- runs on any PC computer: e.g. next to PACS, conference rooms, physician offices, at home, hotel, ... 100eds

(*Similar systems available by many other vendors*)

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**Interactive Visualization – Interpretation**

- 3D thoracic Aorta
  - VR (candy-cane view)
- Measurements
  - MIP (5mm) ~ a-p
  - MIP (5mm) ~ lat.
  - (MPR orthogonal)
- Coronary Anatomy
  - VR
- Sinuses + valve
  - VR 'transparent'
  - MinIP (inverted)
  - 3 Chamber view

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**3D and 4D Pre- and Postop. CT of the Thoracic Aorta**

**Learning Objectives**

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**Thoracic Aorta Surgical Anatomy**

Descending Aorta
Transverse Aorta
Ascending Aorta
Aortic Root

* Sino-tubular Junction
# Sinuses of Valsalva

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**Thoracic Aorta Surgical Anatomy**

Descending Aorta
Transverse Aorta
Ascending Aorta
Aortic Root

* Sino-tubular Junction
# Sinuses of Valsalva
73 y/o retired RN
ascending aortic aneurysm

Normal diameter of thoracic aorta

<table>
<thead>
<tr>
<th>Anulus</th>
<th>normal 23-27mm</th>
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<tbody>
<tr>
<td></td>
<td>&gt;27mm auloaortic ectasia</td>
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Thoracic aorta (incl. sinuses and STJ)

| normal | age-, sex-, body size dependent |
| 'ectatic' | appears enlarged, but ≤4cm |
| >4.0 cm | aneurysm (root, asc., tra., desc.) |

Treatment indication (elective)

| >4.5-5.0cm | (syndromic patients) |
| >6.0+cm | (degenerative) |
| rapid diameter increase (>5mm/year) |

Ao. root valve insufficiency

Thoracic Aortic Aneurysms (TAA)
Etiology and associated conditions

- atherosclerotic
- genetic (congenital wall defect)
  - Marfan's
  - Bicuspid aortic valve
- chronic dissection
- infectious (mycotic, syphillis)
- vasculitis
- post-traumatic

very common

rarer

75 y/o woman w. atherosclerotic aneurysm of the thoracic aorta (normal root)
Thoracic Aortic Aneurysms (TAA)

Etiology and associated conditions

congenital (intrinsic, defect of aortic wall):

- **Marfan’s**
  - FBN-1 gene
  - fibrillin= extracellular matrix fiber regulates TGF-β
  - prevalence (0.01-0.02 %)

- **Bicuspid AV**
  - fibrillin, MMP 2
  - prevalence (1-2 %)

- **Ehlers-Danlos IV**
  - COL 3A1 gene (procollagen III)

- **Loeys-Dietz**
  - TGFBR1 or TGFBR2 gene

TGF: transforming growth factor, MMP: matrix metalloproteinase.

Prevalence 1-2%

Complications (>33%)

- valve degeneration and stenosis
- endocarditis
- aortic root dilatation (50% of young pts.)
  - ao. root aneurysm

BAV disease (bicuspid aortic valve disease)

- Prevalence 1-2%
- Complications (>33%)
  - valve degeneration and stenosis
  - endocarditis
  - aortic root dilatation (50% of young pts.)
  - ao. root aneurysm

33 yo woman, Marfan’s Disease
gated chest

Aortic root aneurysm

33 yo woman, Marfan’s gated chest

Aortic Root Aneurysm, Mitral Valve Prolapse

BAV disease (bicuspid aortic valve disease)

- Prevalence 1-2%
- Complications (>33%)
  - valve degeneration and stenosis
  - endocarditis
  - aortic root dilatation (50% of young pts.)
  - ao. root aneurysm

BAV - aneurysm, interrupted desc. ao.
**Bicuspid Aortic Valve Disease**

- Abnormal origin of right coronary artery

**Marfan's disease**
- Aortic root aneurysm, mild AR
- Coronary fistula /w systemic feeders

**Thoracic Aorta Surgical Options**

**Aortic Root**
- Anuloplasty
- Composite graft
  - Contains valve
  - Coronary reimplantation
- Valve Sparing Surgery
  - Resect sinuses down to valve insertions
  - Preserve native valves
  - Coronary reimplantation

**Valve Sparing Aortic Root Procedures**

Tirone David's "Reimplantation" Technique

**47 yo man**
Marfan's and BAV

**41 yo man**
Marfan's
Estimate correct annulus diameter (d):
\[ d = (h_{leaflet} \times 1.35) \times \text{mod. from David & Feindel (1992): } d = \left( h_{leaflet} \times 2 \right) \times 0.67 + (2 \times A_{wall}) \]
e.g. 18mm leaflet: 24mm annulus

Clinical 3D and 4D Imaging of the Thoracic Aorta
Case 5: WH&R

27 year old man
Bicuspid aortic valve + root aneurysm
TT-Echo: valve prolaps and severe AR, with LV dilatation;
not ideal candidate for valve-sparing AO root repair

Gated CTA of chest
\cdot preoperative assessment of ao. root
\cdot valve reconstruction attempted

Bicuspid Aortic Valve
Raphe 'L-R' sinus
Severe prolaps of R/L-cusp
**Bicuspid Aortic Valve**

Severe prolaps of R/L-cusp due to rupture of commissural suspensory 'chord'

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**3D and 4D Pre- and Postop. CT of the Thoracic Aorta**

**SUMMARY**

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**Thank you ...**

Department of Radiology
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