Aorta afwijkingen in de (niet) dagelijkse praktijk

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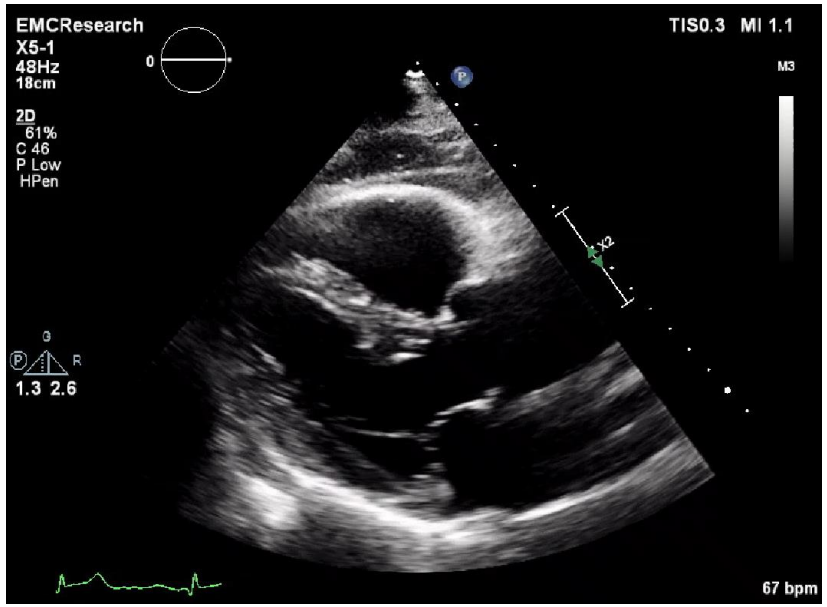
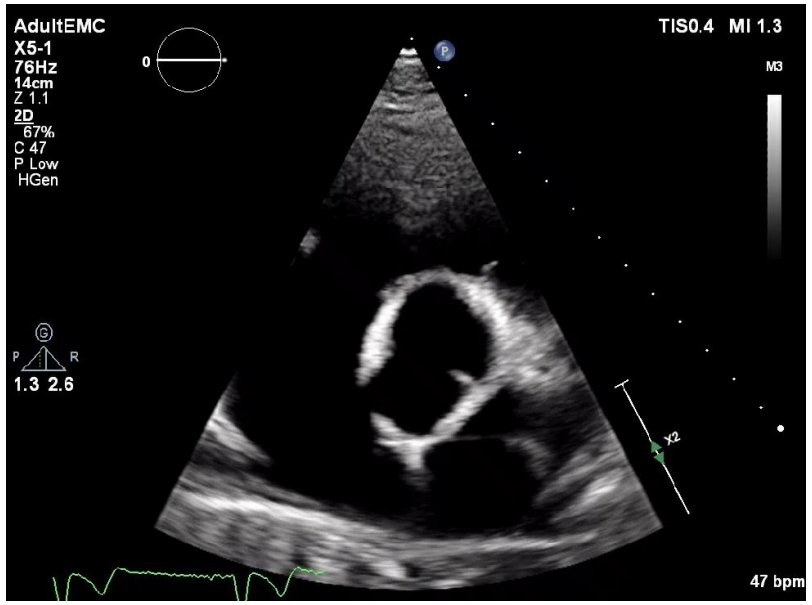
Casus (1)

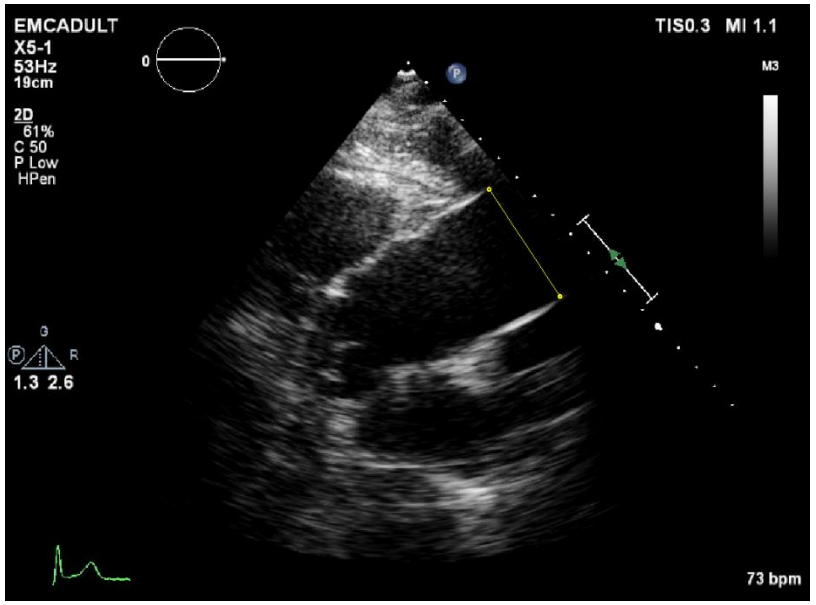
- 34 jarige vrouw, verwezen door de huisarts vanwege een ‘ruisje’
  - Cardiaal blanco
  - Familie anamnese niet bijdragend
- Geen cardiale klachten
- RR 115/69 mmHg (li=re), RR been 125/80 mmHg, pols 75/min RA
- Cor: S1S2, graad 2 systolische souffle tweede ICR

- ECG: SR, 70/min, normale as en geleiding, normale repolarisatie

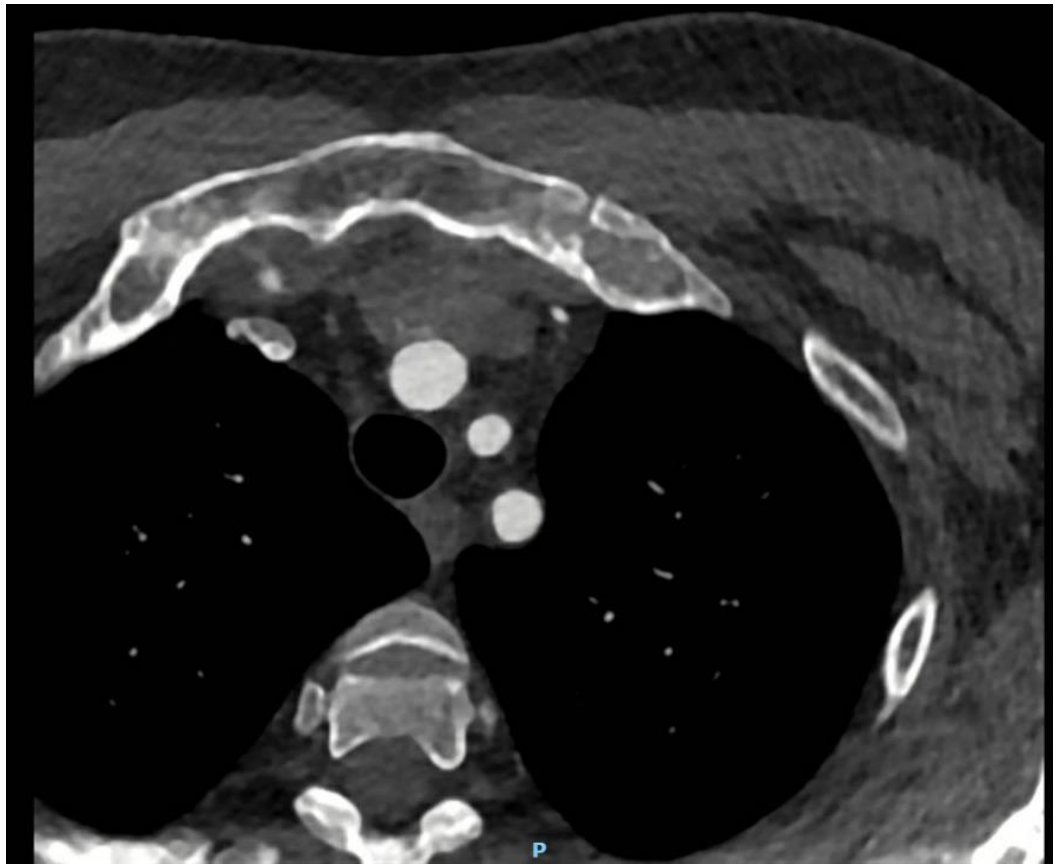
Casus (1)

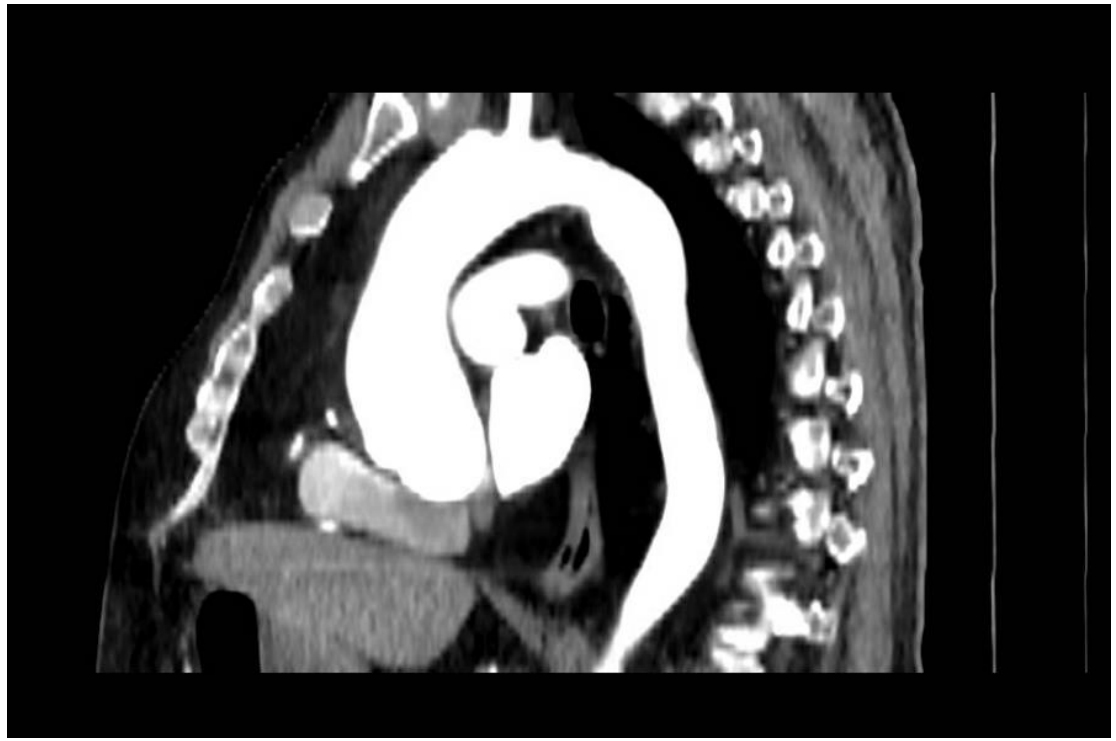

EMCResearch
X5-1
48Hz
18cm
2D
61%
C 46
P Low
HPen
TIS0.3 MI 1.1
M3
1.3 2.6
67 bpm


AdultEMC
X5-1
76Hz
14cm
Z 1.1
2D
67%
C 47
P Low
HGen
TIS0.4 MI 1.3
M3
1.3 2.6
47 bpm


EMCADULT
X5-1
53Hz
19cm
2D
61%
C 50
P Low
HPen
TIS0.3 MI 1.1
M3
1.3 2.6
73 bpm

Casus (2)



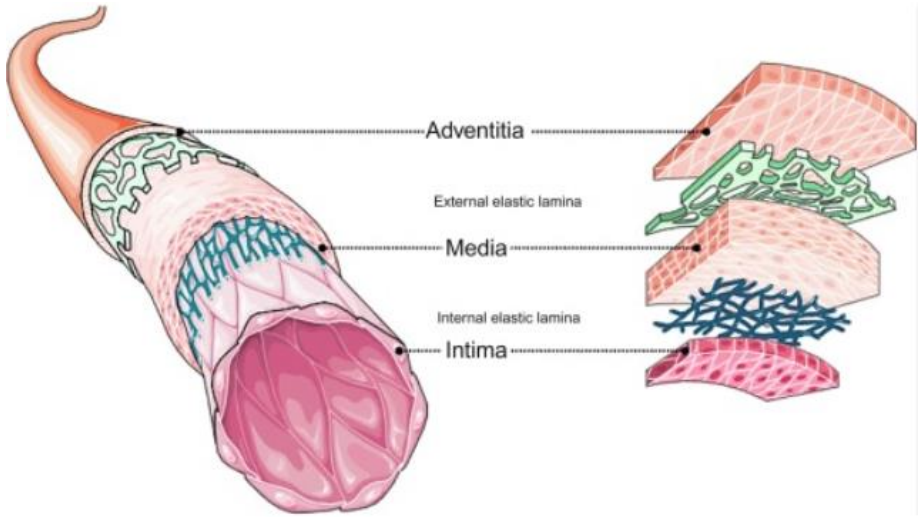


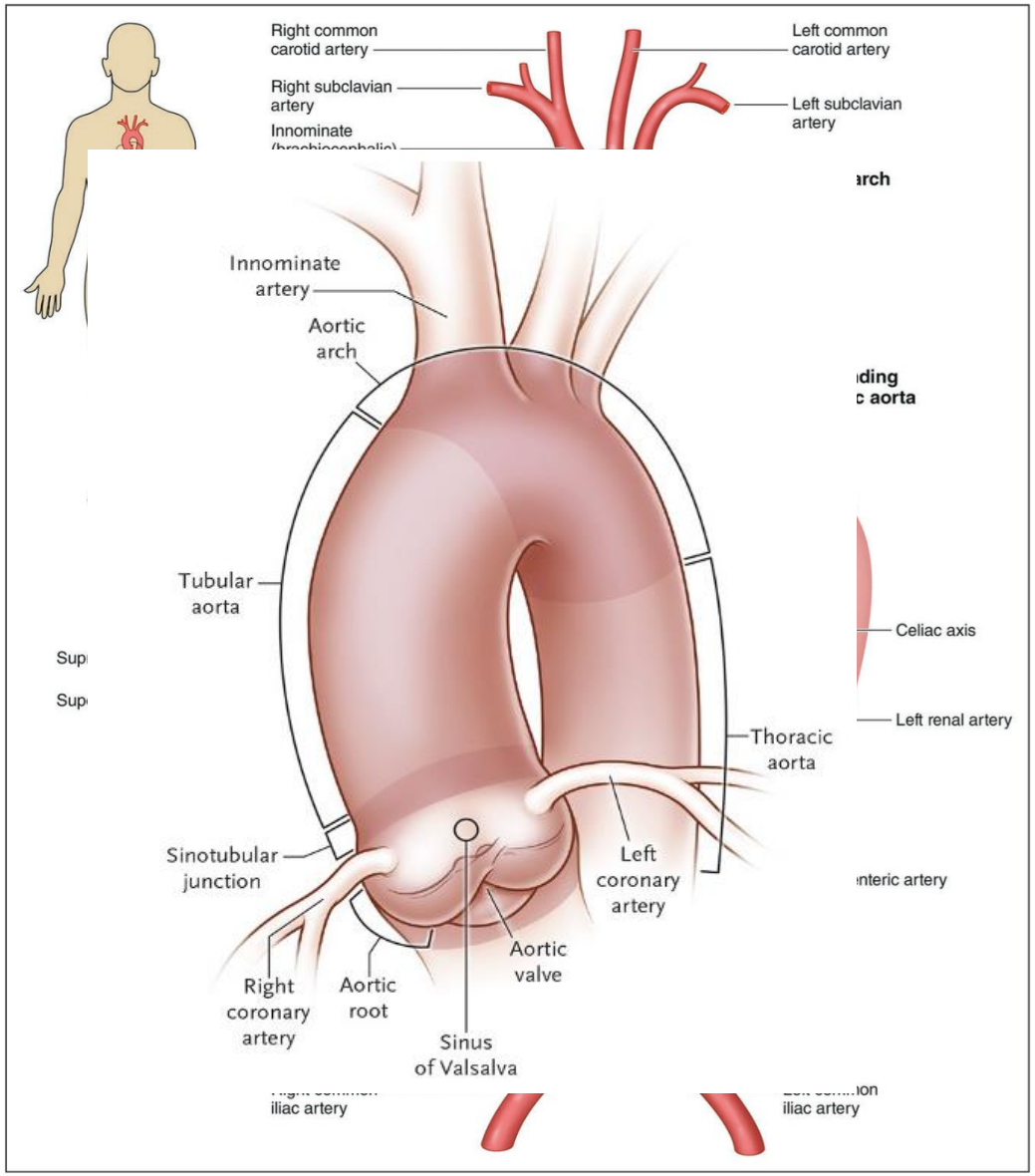
Aorta afwijkingen: inhoud

- Anatomie en definities
- Voorkomen en oorzaken van aorta afwijkingen
- Metingen: wat, op welke manier en hoe vaak?
- Syndromale aorta afwijkingen
- Complicaties: aorta dissectie
- Dagelijkse praktijk: sport en zwangerschap
- Behandeling van aorta afwijkingen

Anatomie

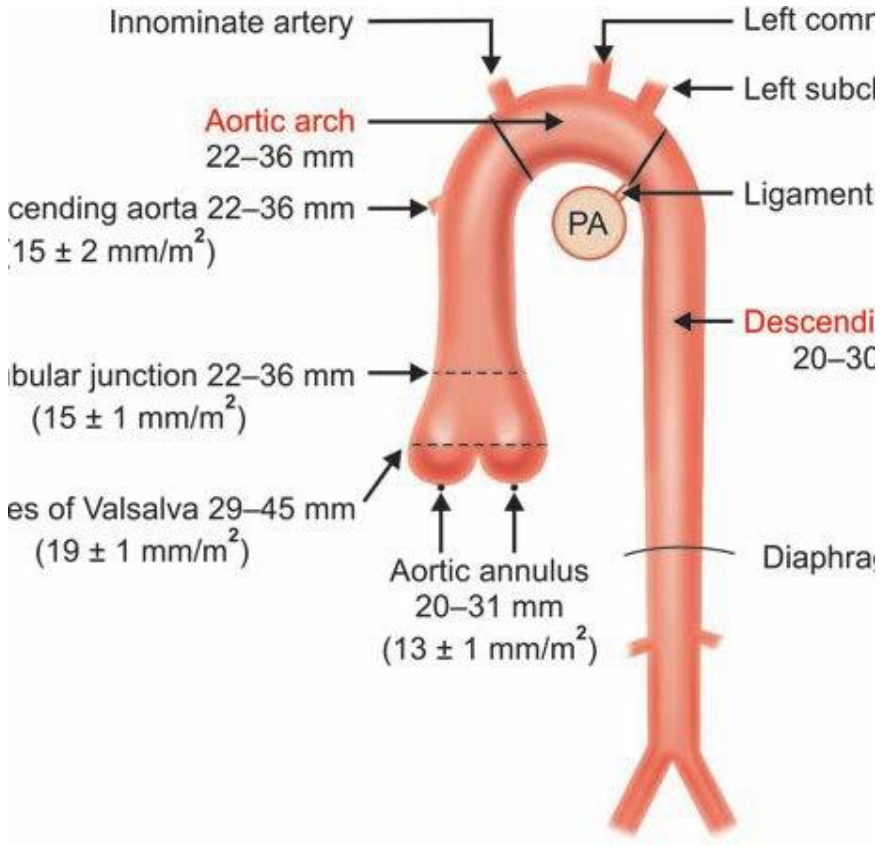
- Aorta: snelweg van de bloedsomloop
  - Gedurende leven 200 miljoen liter bloed!
- Aorta thoracalis en abdominalis
- Focus cardiologie: aorta wortel, ascendens en boog
- Histologie:





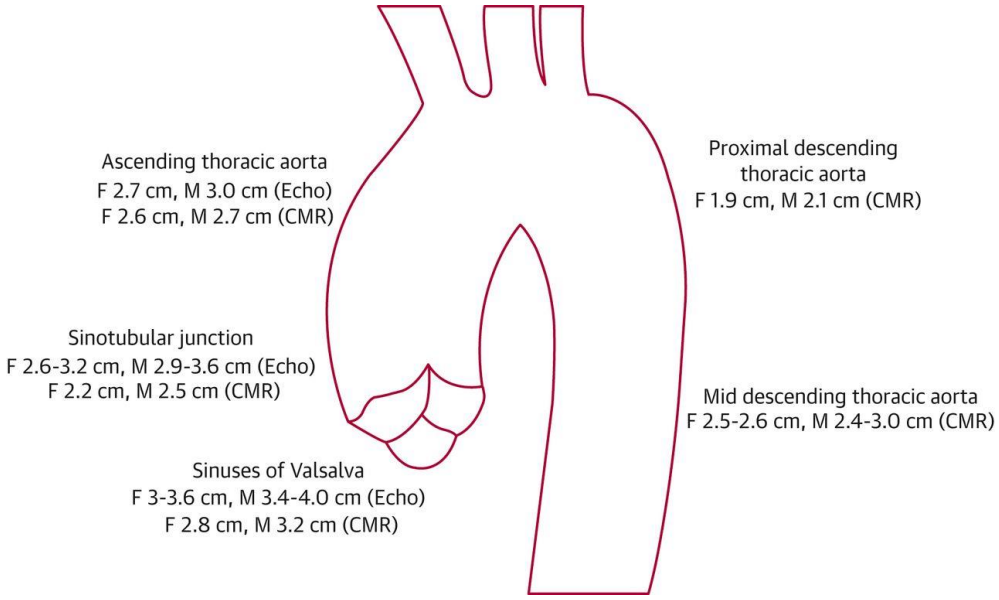
Anatomie

- Diameter aorta afhankelijk leeftijd, geslacht en locatie
- Normale groei 0.9 mm per decade mannen en 0.7 mm per decade vrouwen
- Functies van de aorta:
  - Conduit
  - Regulatie bloeddruk (druk receptoren)
  - Secundaire pomp (windketel fenomeen)



Definities

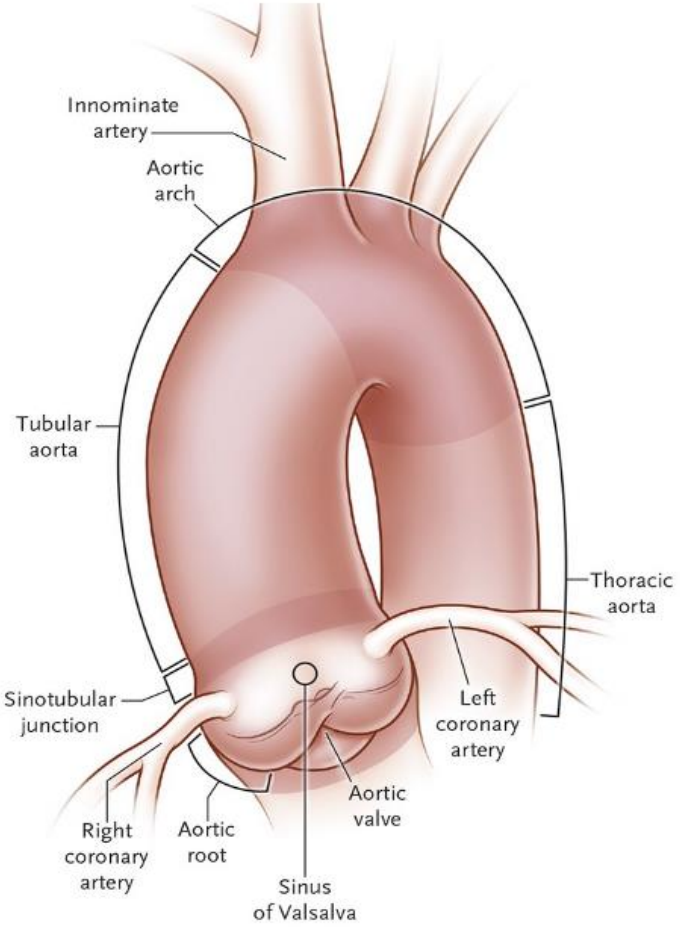
- Definitie aneurysma: dilatatie van tenminste 1.5 maal normale diameter
  - Normale diameter thoracale aorta varieert qua locatie en leeftijd van patiënt
- Meestal in buik aorta (66%) en minder vaak thoracale aorta (28%). Soms thoraco-abdominaal (10%)



Definities

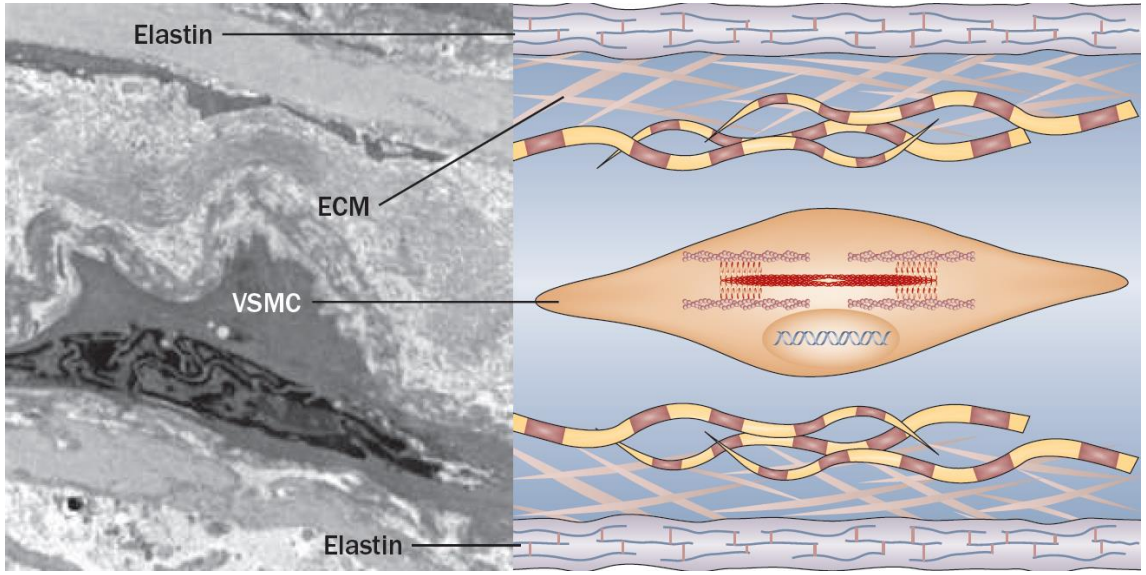
Thoracale aneurysmata:

- Aorta ascendens (van aortaklep tot anonima): 60%
- Aorta boog (alle thoracale aneurysmata met betrokkenheid brachiocephale vaten): 10%
- Aorta descendens (distaal linker subclavia): 40%
- Thoraco-abdominaal: 10%



Oorzaak aorta aneurysma / dissectie

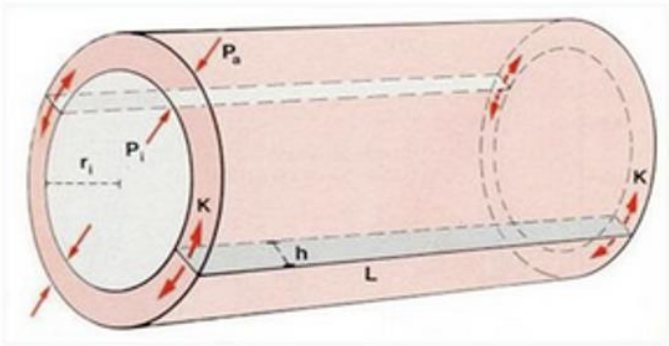
- Histologie:
  - Gladde spiercellen dropout
  - Elastische vezels degeneratie
- Dit zie je ook bij veroudering en bij hypertensie



Oorzaak aorta aneurysma / dissectie

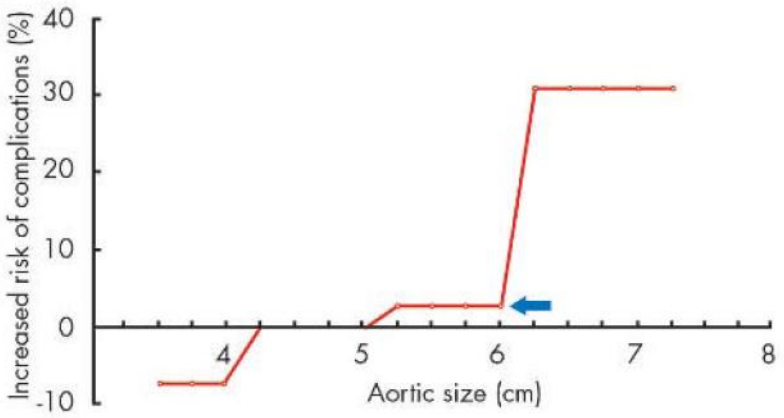
Wet van La Place: $K = P\frac{r}{2d}$

K = Wandspanning
P = Druk
r = Radius
d = Wanddikte

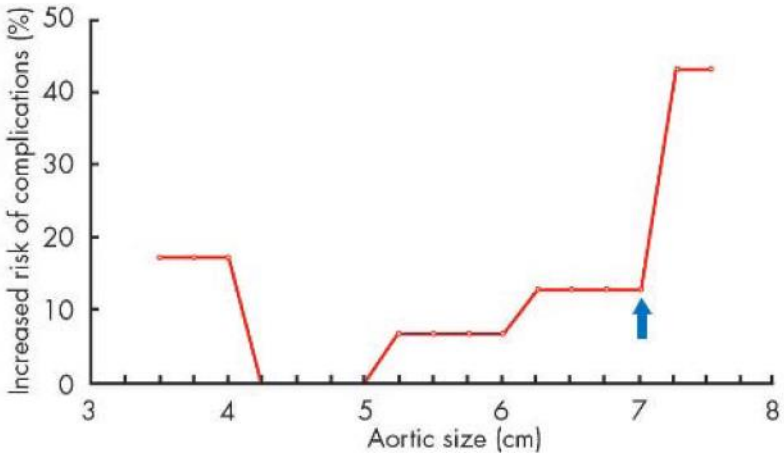


Een toename van de binnenradius van het vat, of een stijging van de druk in het vat (hypertensie!) leidt tot een verhoging van de wandspanning en een afname van de wanddikte

Risico op complicaties versus diameter aorta



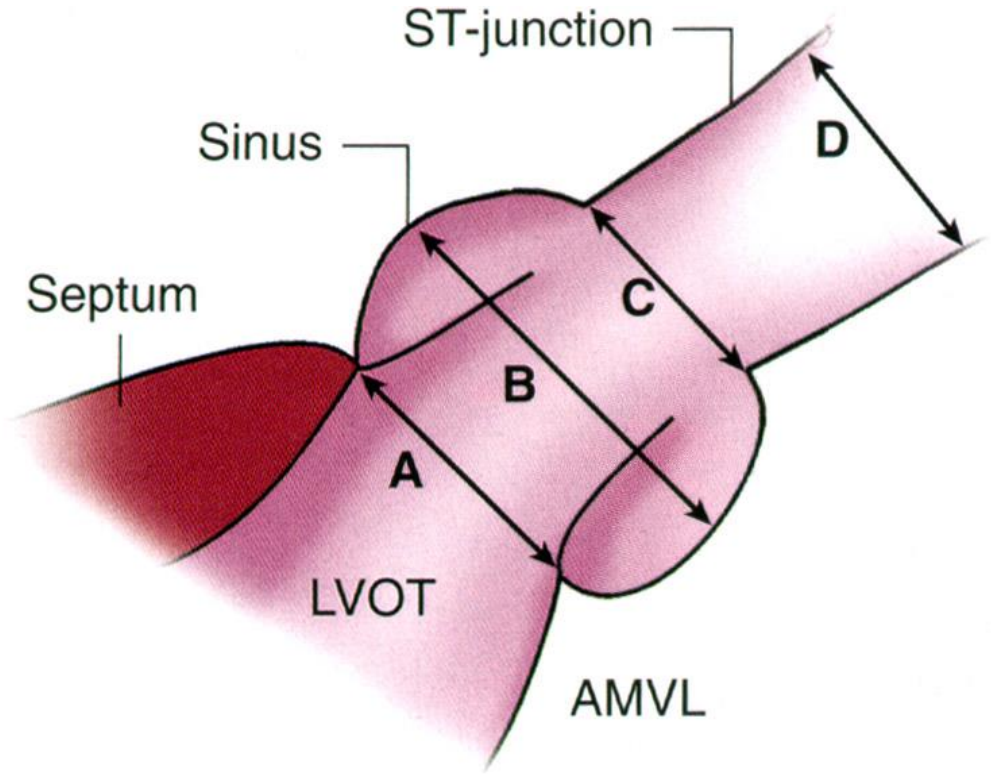
Ascending aorta

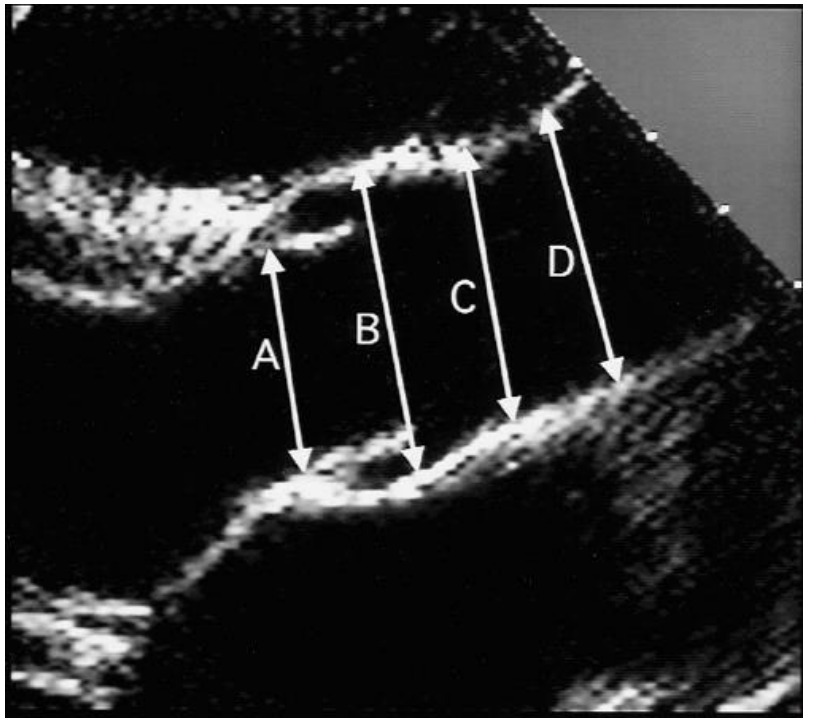


Descending aorta

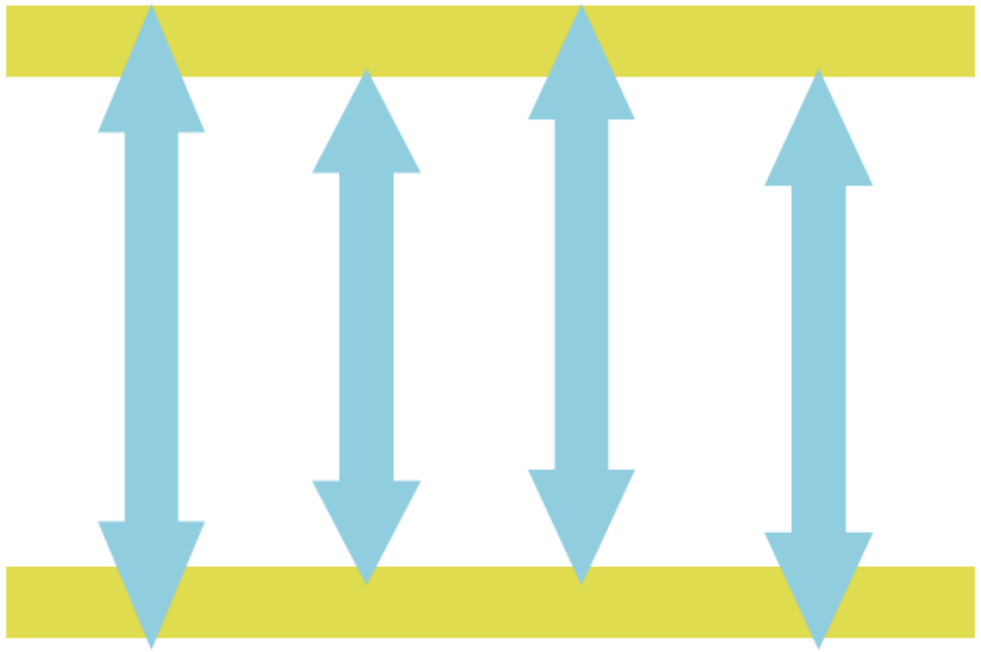
Elefteriades et al, J Am Coll Cardiol 2010;55

Wat moet je meten? Echo

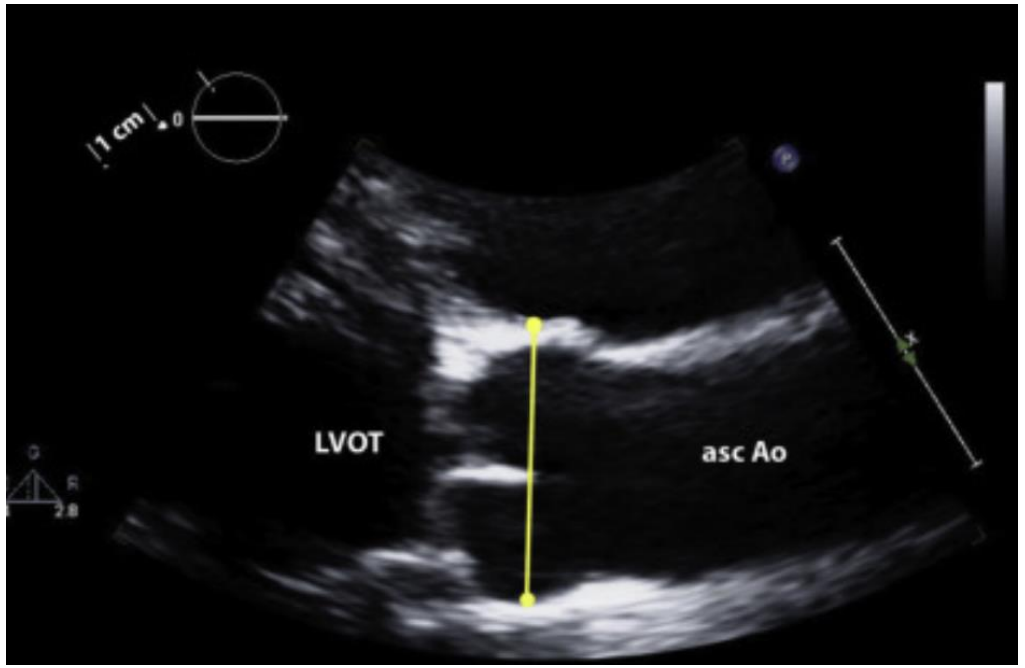




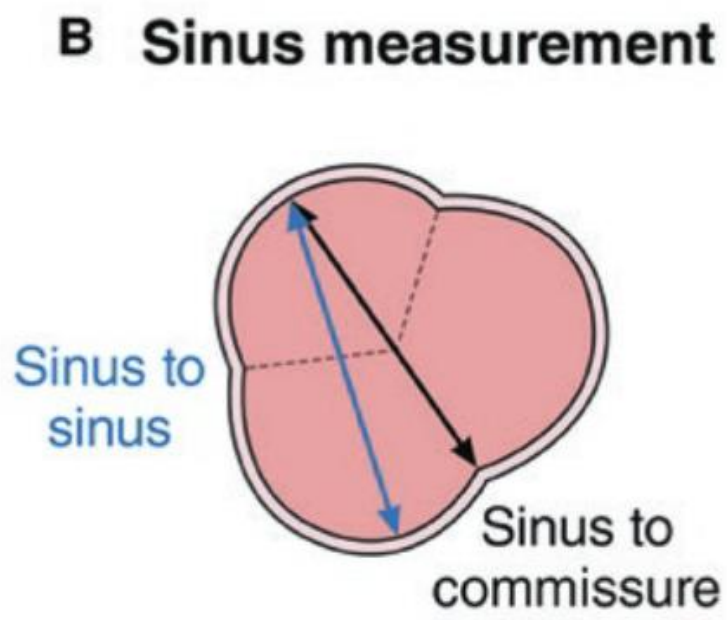
Hoe moet je meten?

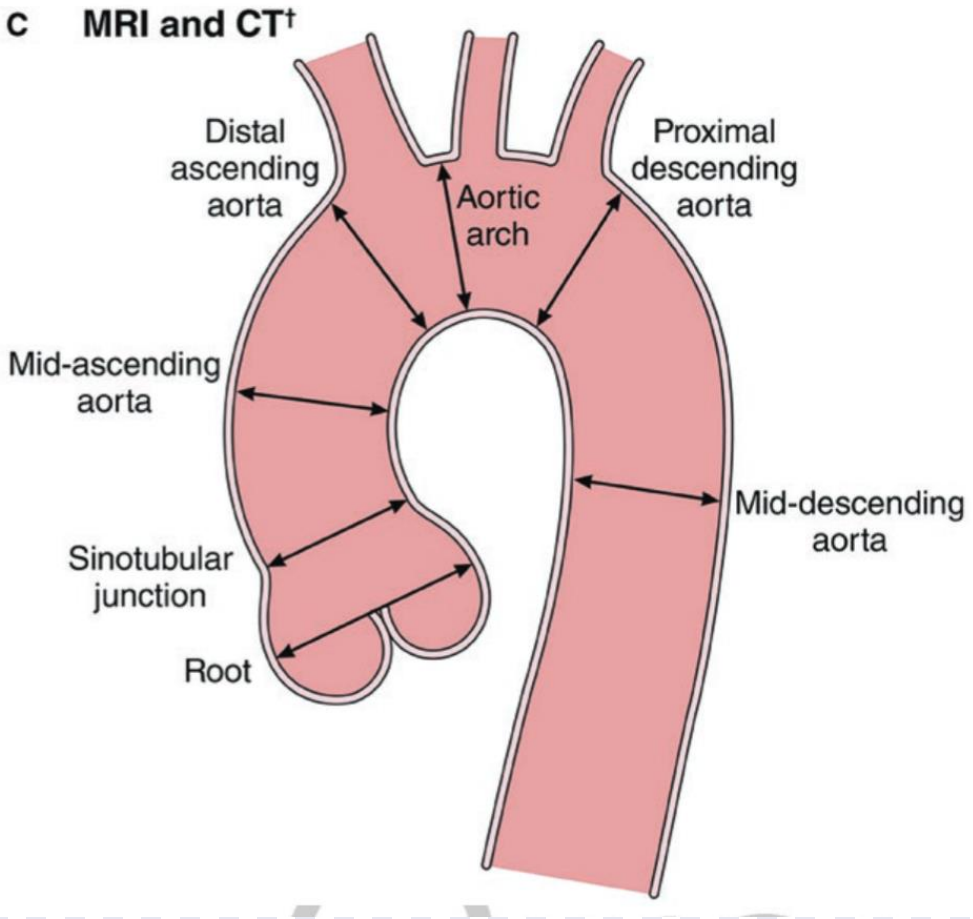


Leading edge to leading edge
Einde diastole



Wat moet je meten? CT





Grote variatie!

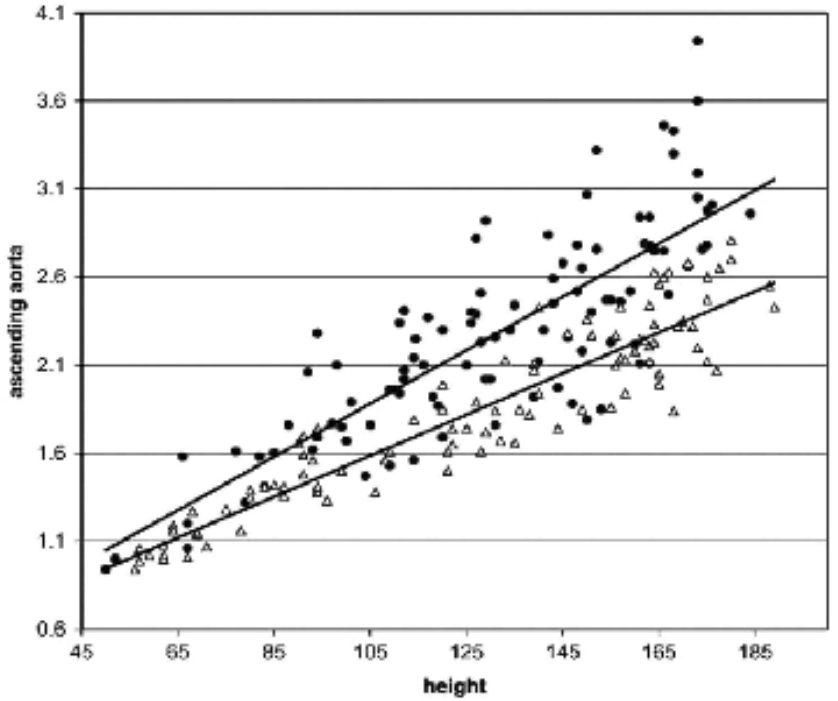
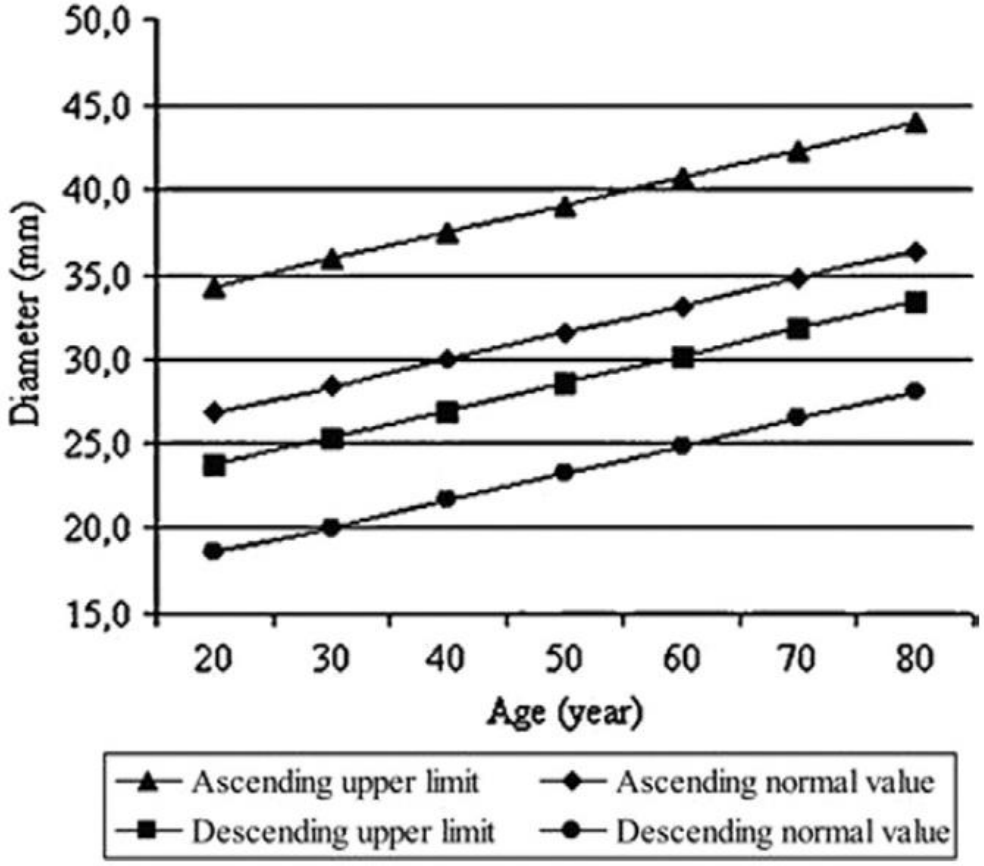


Figure 4. Ascending aortic diameter (cm) as a function of height (cm) among children with BAV (n=101) vs controls with TAV (n=97). The upper slope (circles) represents the BAV group ($y=0.0151x+0.2926$), and the lower slope (triangles) represents the TAV group ($y=0.0117x+0.362$). Reprinted from Beroukhim et al,[65] with permission from Elsevier. Copyright 2006.



Tadros e.a. Circulation 2009;119:880-890

Hoe vaak moet je meten?

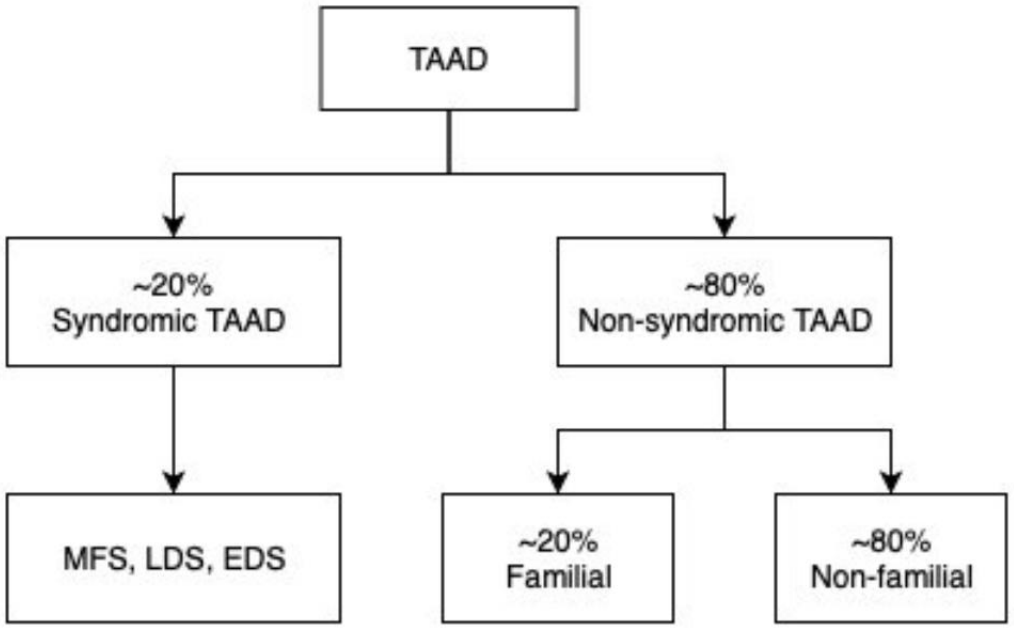
COR	LOE	RECOMMENDATIONS
1	C-LD	1. In patients with a dilated thoracic aorta, a TTE is recommended at the time of diagnosis to assess aortic valve anatomy, aortic valve function, and thoracic aortic diameters.[1-4]
2a	C-LD	2. In patients with a dilated thoracic aorta, a CT or MRI at the time of diagnosis is reasonable to assess thoracic aortic anatomy and diameters.[1,3,5-7]
2a	C-LD	3. In patients with a dilated thoracic aorta, follow-up imaging (with TTE, CT, or MRI, as appropriate based on individual anatomy) in 6 to 12 months is reasonable to determine the rate of aortic enlargement; if stable, surveillance imaging every 6 to 24 months (depending on aortic diameter) is reasonable.[1,3,4]

Isselbacher et al, JACC 2022

Incidentie aorta aneurysma

- Aorta aneurysma is de 19e doodsoorzaak
- Incidentie aneurysma aorta thoracalis: 6/100.000
- Prevalentie niet symptomatisch TAA 0.16%-0.34%
  - Met name in zesde en zevende decade, 2-4 maal zo vaak bij mannen
- Waarschijnlijk echter veel hoger, omdat veel aneurysmata geen klachten geven of gemist worden bij overlijden!
- Meestal helemaal geen klachten!
  - High index of suspicion bij acute klachten

Indeling



Verworven
- Atherosclerose
- Hypertensie
- Takayasu arteritis
- Syfilis
- Tuberculose
- HIV

Familiaal syndromaal
- Marfan syndroom
- Ehlers-Danlos syndroom Type IV
- Turner syndroom
- Loeys-Dietz syndroom
- Bicuspide aortaklep

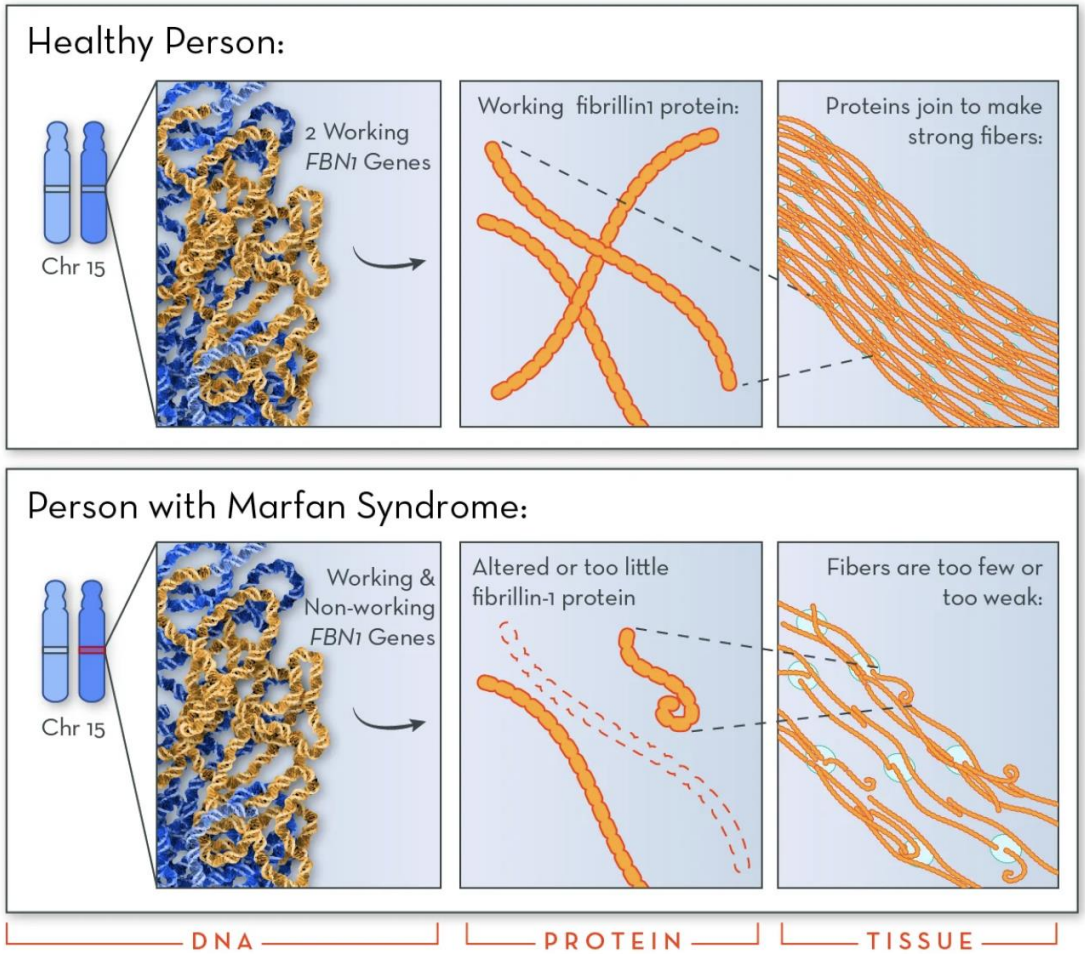
Familiaal non-syndromaal
- Aorta syndroom

Syndromaal

Syndromic		
Marfan syndrome	15q21.1	*FBN1*
Marfan-like syndrome	3p24-25	*TGFBR2*
Loeys–Dietz syndrome	3p24-25 9q33-34	*TGFBR2*, *TGFBR1*
Ehlers–Danlos syndrome	2q24.3-31	*COL3A1*
BAV–TAA syndrome	9q34-35, others	*NOTCH1* Unidentified
Arterial tortuosity syndrome	20q13.1	*SLC2A10*
Turner syndrome	45,X0	Unidentified
Noonan syndrome	12q24.1 2p21-22 12p12.1	*PTPN11* *SOS1* *KRAS*
Polycystic kidney disease	16p13.3 4q21-22	*PKD1* *PKD2*

Marfan syndroom

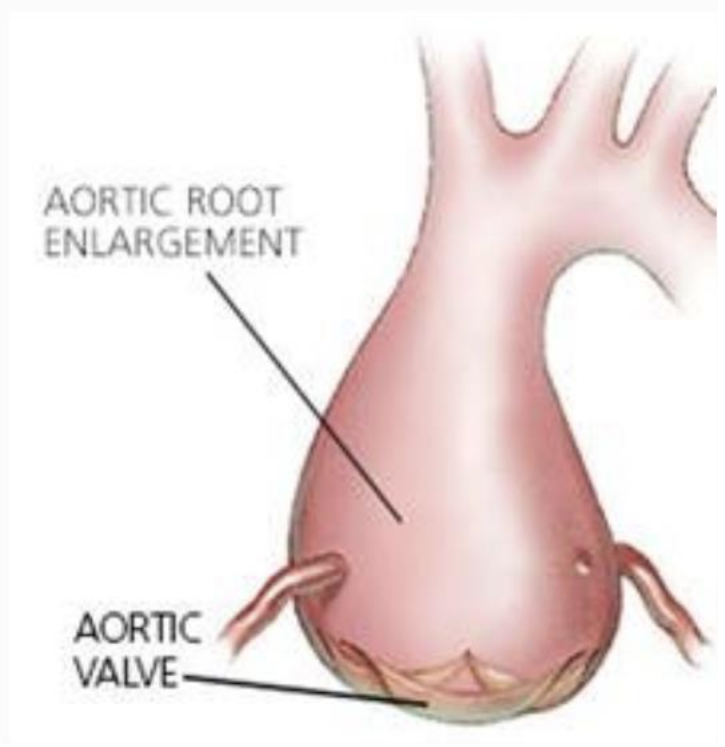
- Frequent voorkomende syndromale aortopathie
- Bindweefselafwijking, mutaties in fibrilline 1 gen (FBN1)
  - Matrix component voor zowel elastische als niet-elastische weefsels
- Autosomaal dominante aandoening
- Incidentie 1:3000-5000 personen
- Grote variatie voorkomen afwijkingen in cardiovasculair, oculair en musculoskeletaal systeem



Marfan syndroom

Cardiale afwijkingen:

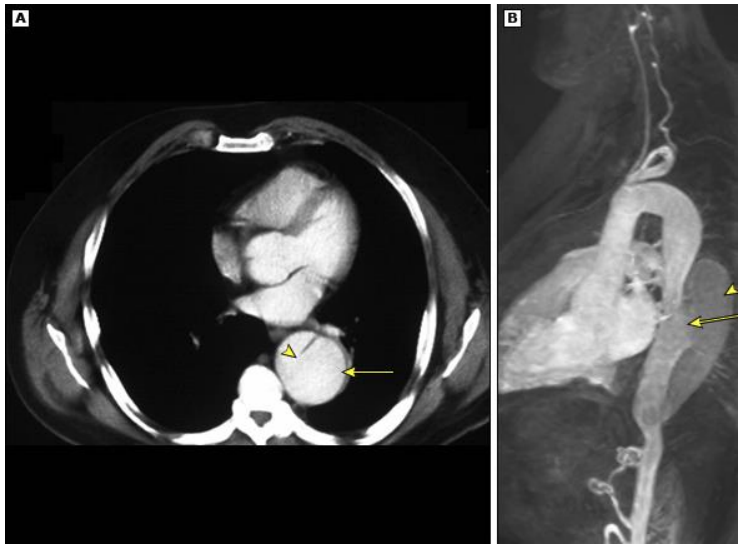
- Aorta pathologie
  - Aorta wortel dilatatie, voornaamste oorzaak mortaliteit Marfan
    - Aneurysma kan overal voorkomen
  - Slechte correlatie ernst aorta afwijkingen en oculaire en musculoskeletale afwijkingen



Marfan syndroom

Aorta pathologie

- Aanwezig bij 60%-80% volwassenen met Marfan
- Marfan aanwezig bij 50% patiënten dissectie <40 jaar
- Jaarlijks risico dissectie echter 0.5% per jaar indien aorta <50 mm
- Potentieel grote gevolgen voor zwangerschap!



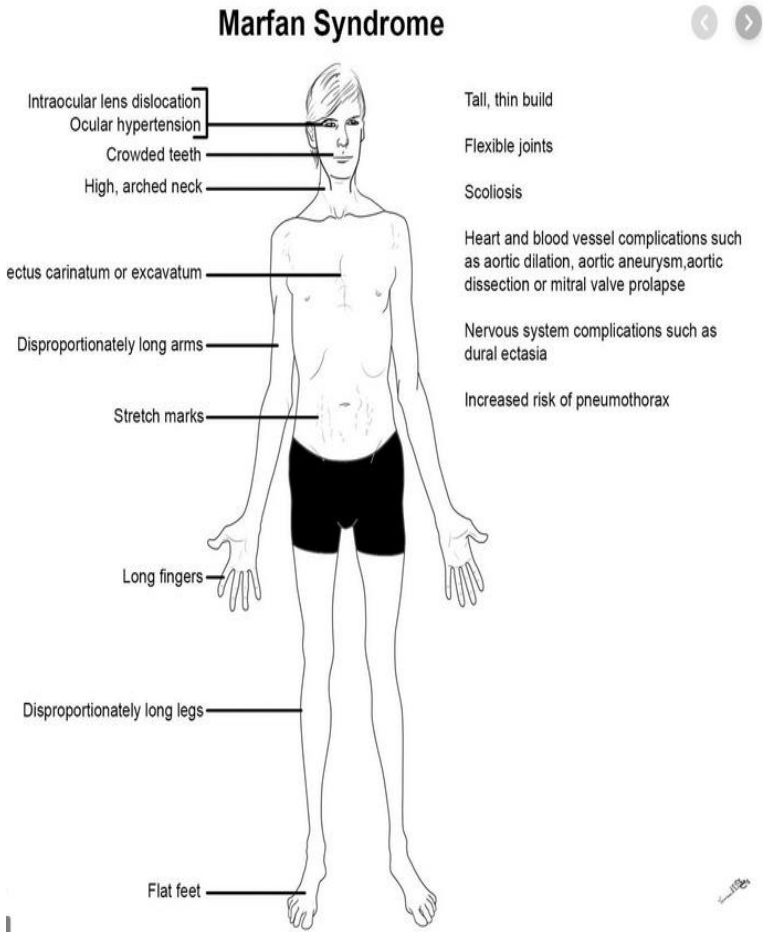
Jondeau et al, Circulation 2012)

Marfan syndroom

Andere cardiale afwijkingen:

- Mitralisklep prolaps bij 40-54%
  - Meestal zeer milde insufficiëntie
  - Bij 25% progressief
- Soms ook tricuspidalisklep aangedaan

- Niet cardiale uitingen komen veel voor



Marfan syndroom: behandeling

- Medicamenteus
  - Bètablokker
  - ACE remmer of ARB?
- Chirurgie

Table 12 **Indications for aortic surgery in Marfan syndrome**[67]

Indications	Class[a]	Level[b]
Patients should undergo surgery when aortic root maximal diameter is:		
• >50 mm	I	C[c]
• 46–50 mm with		
- family history of dissection or	I	C
- progressive dilation >2 mm/year as confirmed by repeated measurement or	I	C
- severe AR or MR or	I	C
- desire of pregnancy	I	C
• Patients should be considered for surgery when other parts of the aorta >50 mm or dilation is progressive	IIa	C

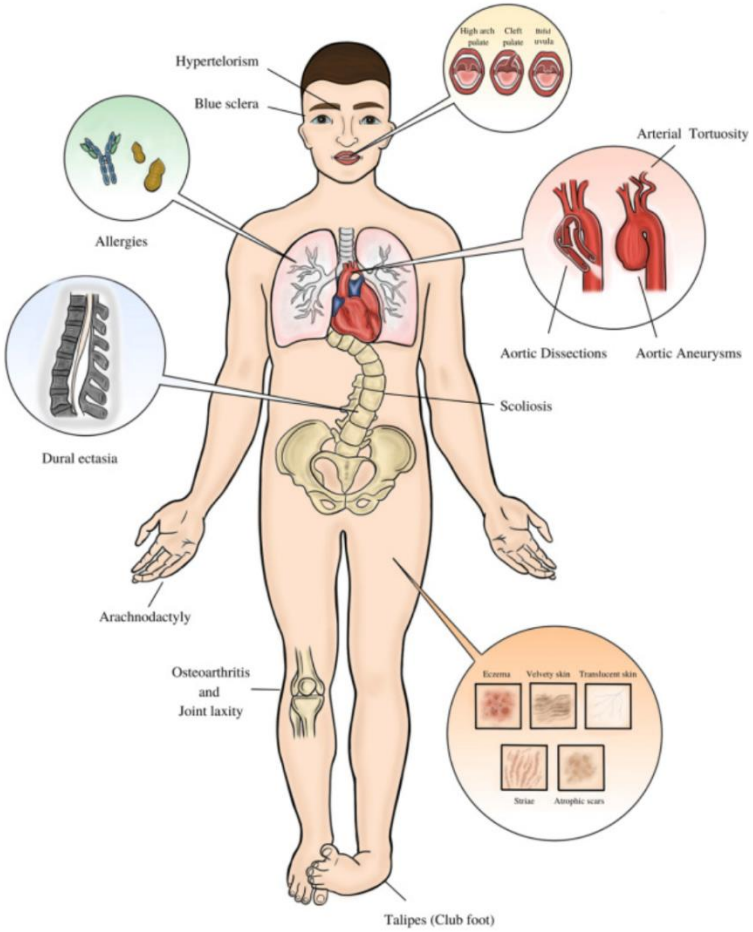
[a]Class of recommendation.
[b]Level of evidence.
[c]ESC guidelines for valvular heart disease are slightly more strict, recommending only one diameter (45 mm) regardless of other findings.
AR = aortic regurgitation; MR = mitral regurgitation.

ESC Guidelines 2010

Marfan syndroom: beloop

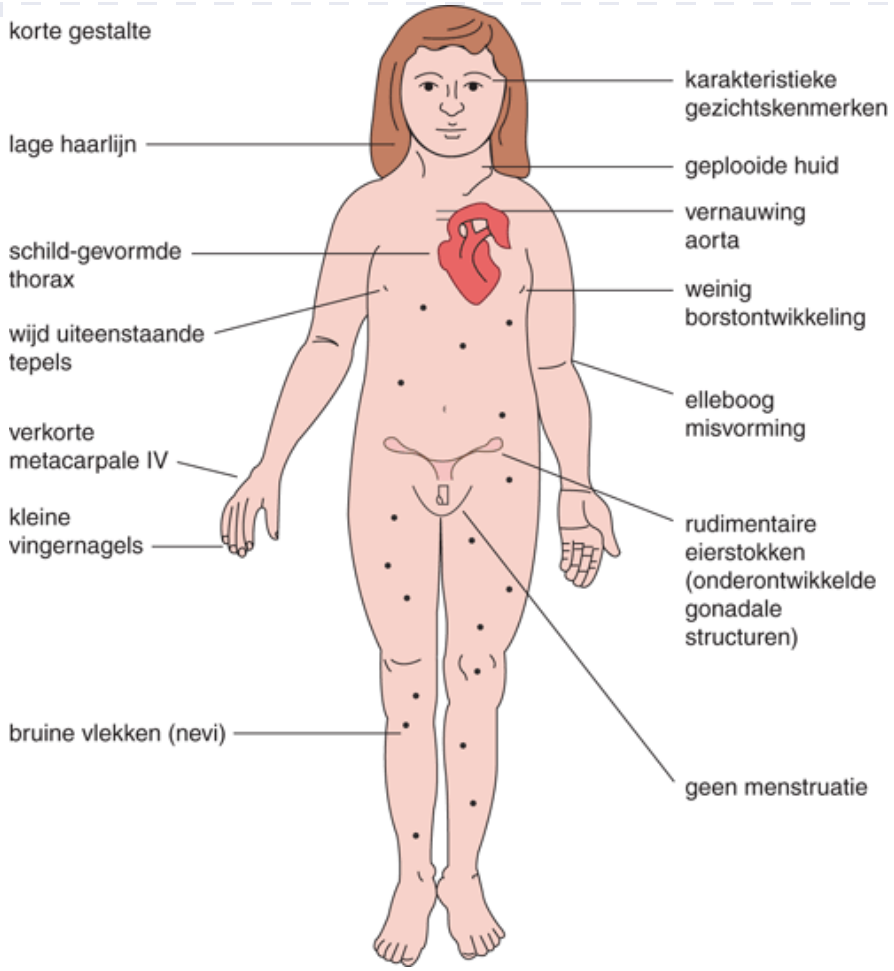
Loeys-Dietz syndroom

- Eerst beschreven in 2005
- Mutatie TGFBR1 of TGFBR2 gen, autosomaal dominant gedrag
- Arteriële turtuositeit en aneurysmata door de hele vaatboom
- Agressief gedragende arteriopathie
- Vroege interventie, >45 mm aorta ascendens



Syndroom van Turner

- Partieel of complete monosomie X chromosoom (45X0)
- Verminderde levensverwachting door spontane ruptuur viscerale organen of bloedvaten
- Cardiovasculair: bicuspide aortaklep, PAPVR en aorta dilatatie
- Vooral in ascendens en boog
- Aortic size index >2.5cm/m^2 risico voor dissectie

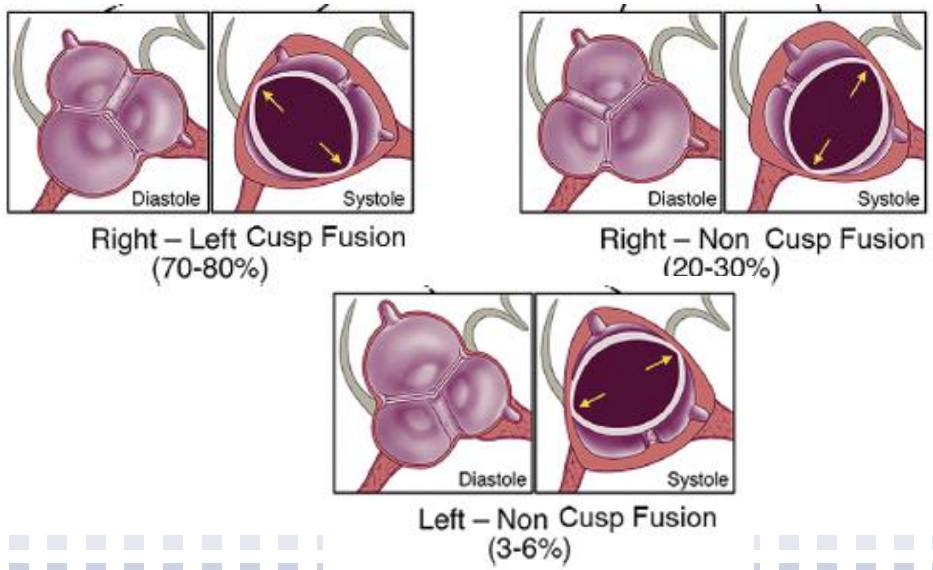


Bicuspide aortaklep

- Bicuspide aortaklep (BAV) meest voorkomende congenitale aandoening (0.5% tot 2%)
  - 3 tot 4 maal vaker bij mannen
- Vaak geïsoleerd maar ook frequent bijkomendecongenitale afwijkingen (15% volwassenen, 50% kinderen)
- Soms in kader van syndroom (Turner, Loeys-Dietz), vaak sporadisch
- Aorta afwijkingen frequent!

<30 jaar	30-39 jaar	40-49 jaar	50-60 jaar	>69 jaar
56%	74%	85%	91%	88%

Prevalentie dilatatie tubulaire deel aorta

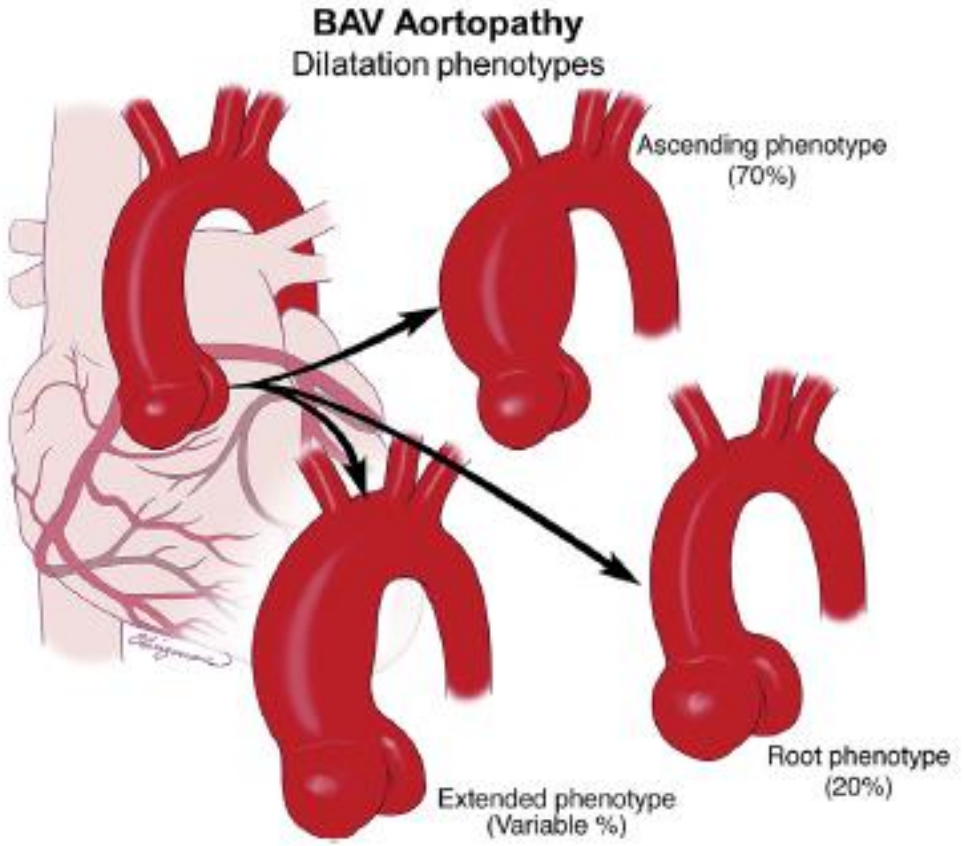


Verma et al, NEJM 2014

Bicuspide aortaklep

Twee belangrijke fenotypen dilatatie te onderscheiden:

- Ascendens fenotype (70%)
  - Ouder patiënten
  - Vaker aortaklep sclerose/stenose
  - Geen voorkeur man/vrouw
- Wortel fenotype (20%
  - Jongere patiënten
  - Vaker man
  - Vaker aortaklep insufficiëntie



Michelena et al, J Thorac Cardiovasc Surg 2021

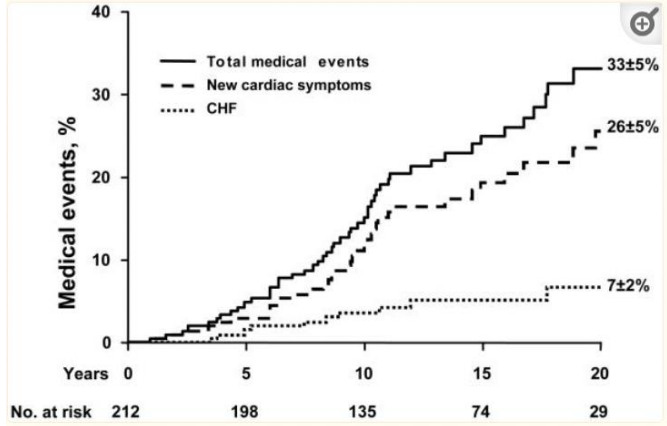
Bicuspide aortaklep

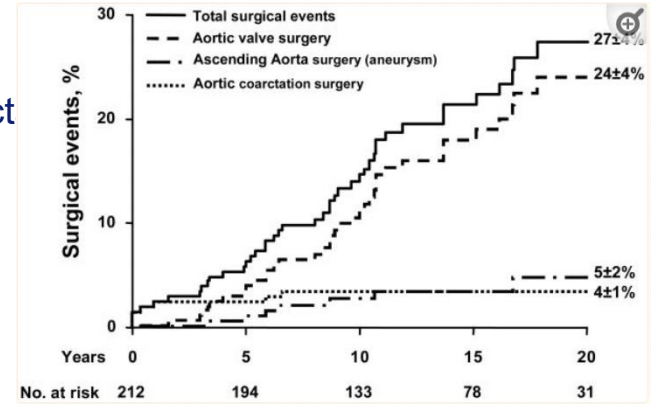
Lifetime risico chirurgie:

- >50% aortaklep vervanging
- >25% aorta chirurgie, vaak in combinatie met AoVR
- Vaak mechanoprothese, aortaklep repair of Ross in geselecteerde gevallen

Dissectie

- Risico relatief laag: 3.1 per 10.000 personen jaren, <1% lifetime risk
  - Echter, 8 maal hoger dan de normale populatie
  - 17.4 per 10.000 personen jaren >50 jaar
  - 44.9 per 10,000 personen jaren indien aneurysma bij baseline
- Risico factoren zijn: ‘root fenotype’, ernstige insufficiëntie, ongecontroleerde hypertensie, coarct en snelle groei (>3 mm per jaar)





Michelena et al, Circulation 2008
Rashed et al, Curr Cardiol Reports 2022

Familiaal non-syndromaal

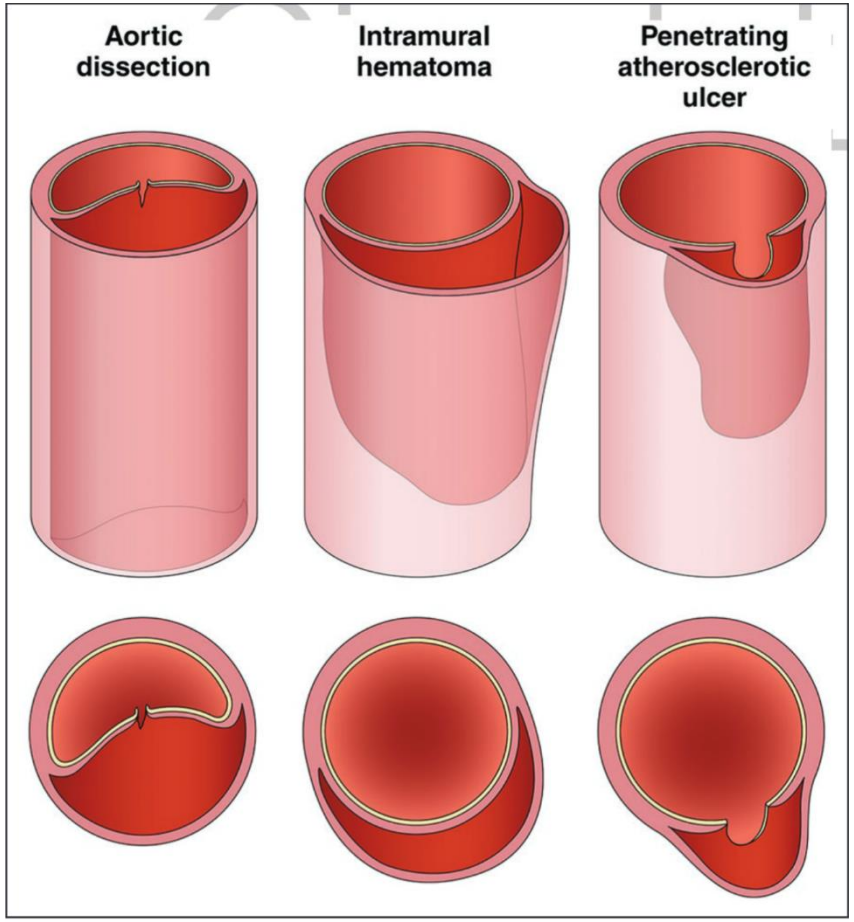
Familial Thoracic Aortic Aneurysm Disease (FTAAD)

- Meerdere personen aorta dilatatie of dissectie zonder onderliggende bindweefselaandoening
- Bij diagnose thoracaal aneurysma bedacht zijn op onderliggende genetische of familiale afwijkingen
  - Familiale TAAD mogelijk tot 20%
  - Jonge leeftijd ontwikkelen vaak aanwijzing hiervoor
  - Indien bij een of meer eerstegraads familie leden aanwezig, genetica overwegen
  - Cave ook vaatafwijkingen buiten de thoracale aorta

Nonsyndromic HTAD (Familial TAA)		
FTAA	*ACTA2*	TAA, aortic dissection, premature CAD and moyamoya-like cerebrovascular disease, livedo reticularis, iris flocculi
FTAA	*MYH11*	TAA, aortic dissection, PDA
FTAA	*MYLK*	Aortic dissection at relatively small aortic size
FTAA	*PRKG1*	Aortic dissection at young ages at small aortic sizes
FTAA	*MAT2A*	TAA, aortic dissection, BAV
FTAA	*MFAP5*	TAA, aortic dissection, skeletal features may be present
FTAA	*FOXE3*	TAA, aortic dissection
FTAA	*THSD4*	TAA, aortic dissection

Complicaties: acute aorta syndrome (AAS)

- AAS levensbedreigend: onderbreking vaatwand van de aorta
- Drie belangrijke vormen:
  - Aorta dissectie
  - Intramuraal hematoom
  - Penetrating ulcer



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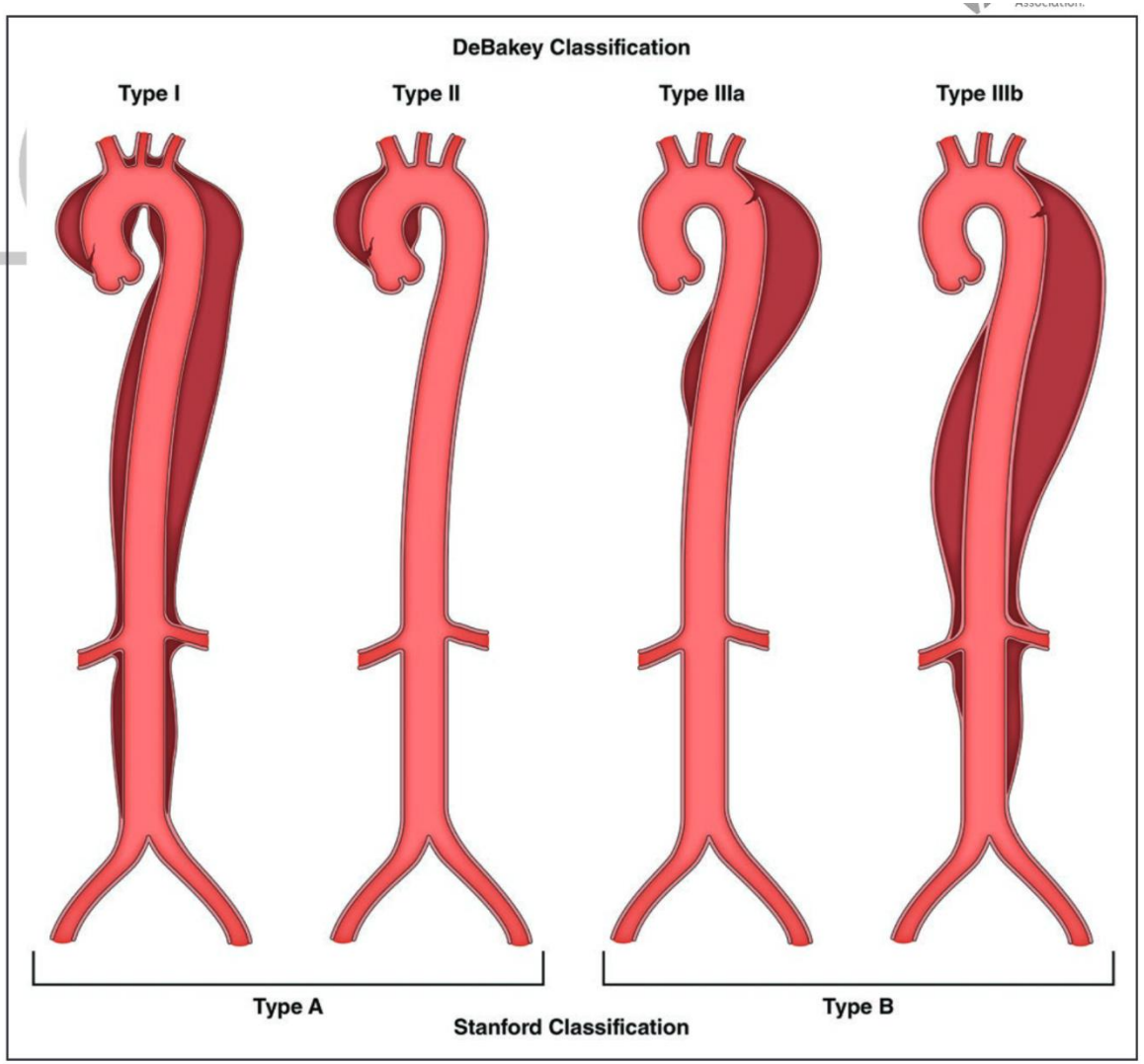
Complicaties: acute aorta syndrome (AAS)

Aorta dissectie

- Meest voorkomend AAS: 5-30 per miljoen
- Intima scheur, waardoor bloed de lamina media bereikt
  - Ware lumen en valse lumen
  - Antegrade of retrograde dissectie
- Hoge mortaliteit: 1% tot 2% per uur
- Uitkomsten chirurgie o.a. afhankelijk ervaring chirurg!



Zwangerschap en aorta afwijkingen

- Zwangerschap hoog risico voor patiënten met aorta aneurysma
  - Onderliggende diagnose van groot belang!
  - Tot 8.4 maal hoger risico op dissectie tijdens zwangerschap
    - Hoge mortaliteit
    - Vaak bij voorheen niet bekende aneurysmata
- Vooral in derde trimester (50%) en post-partum (33%)
  - 0.3% van alle dissecties
  - 1% alle dissecties bij vrouwen
- Counseling voorafgaand zwangerschap essentieel!

Zwangerschap en aorta afwijkingen

	mWHO II-III	mWHO III	mWHO IV
Diagnosis	1. Aorta <45 mm in BAV pathology 2. Marfan or other HTAD without aortic dilatation	Moderate aortic dilatation 1. 45–50 mm in BAV 2. 40-45mmm in Marfan or other HTAD 3. ASI 20-25 mm/m^2 in Turner 4. <50 mm in Fallot	Severe aortic dilatation: 1. >50 mm in BAV, 2. >45 mm in Marfan or other HTAD 3. ASI > 25 mm/m^2 in Turner 4. >50 mm in Fallot
Risk	Intermediate increased risk of maternal mortality or moderate to severe increase in morbidity	Significantly increased risk of maternal mortality or severe morbidity	Extremely high risk of maternal mortality or severe morbidity
Maternal cardiac event rate	10% – 19%	19% – 27%	40% – 100%
Care during pregnancy	Referral hospital	Expert centre for pregnancy and cardiac disease	Expert centre for pregnancy and cardiac disease
Minimal follow-up visits during pregnancy	Bimonthly	Monthly or bimonthly	Monthly
Location of delivery	Referral hospital	Expert centre for pregnancy and cardiac disease	Expert centre for pregnancy and cardiac disease

Regitz-Zagrosek et al, EHJ 2018

Zwangerschap en aorta afwijkingen

Table 5 Aortic diseases

	Marfan[19,175]	Bicuspid aortic valve[176]	Loeys Dietz[182-184]	Turner[178,179]	Vascular Ehlers–Danlos[26]
Location of aneurysm/dissection	Everywhere (sinus of Valsalva)	Ascending aorta	Everywhere	Ascending aorta, arch and descending aorta	Everywhere
Risk of dissection	High: 1–10%	Low: <1%	High:1–10%	High: 1–10%	High: 1–10%
Comorbidity	Dural abnormalities Mitral regurgitation Heart failure Arrhythmias	Aortic stenosis or regurgitation	Dural abnormalities Mitral regurgitation	Low height Infertility Hypertension Diabetes Bicuspid aortic valve Coarctation	Dural abnormalities Uterine rupture
Advise not to become pregnant	Ascending aorta >45 mm (or >40 mm in family history of dissection or sudden death)	Ascending aorta >50 mm	Ascending aorta >45 mm (or >40 mm in family history of dissection or sudden death)	ASI >25 mm/m^2	All patients

ASI = aortic size index.

Regitz-Zagrosek et al, EHJ 2018

Zwangerschap en aorta afwijkingen: bevalling

It is recommended to deliver all women with aortic dilatation or (history of) aortic dissection in an experienced centre with a pregnancy heart team, where cardiothoracic surgery is available.	I	C
In patients with an ascending aorta <40 mm, vaginal delivery is recommended.[96]	I	C
In patients with an ascending aorta >45 mm, caesarean delivery should be considered.	IIa	C
In patients with (history of) aortic dissection, caesarean delivery should be considered.	IIa	C
Prophylactic surgery should be considered during pregnancy if the aorta diameter is >45 mm and increasing rapidly.	IIa	C
When the foetus is viable, delivery before necessary surgery should be considered.[96]	IIa	C
In patients with an aorta 40–45 mm, vaginal delivery with epidural anaesthesia and an expedited second stage should be considered.	IIa	C
In patients with an aorta 40–45 mm, caesarean section may be considered.	IIb	C

Regitz-Zagrosek et al, EHJ 2018

Sport en aorta afwijkingen

- Sporten belangrijk voor alle mensen met cardiovasculaire risico factoren!
- Aorta pathologie belangrijk bij sportadvies
  - Geen bewijs versnelde dilatatie aorta bij sporters
  - Aorta dilatatie (>40 mm) geen teken van sporthart
  - 4.1% dissecties gerelateerd aan sporten (32% golf)
- Let op bijkomende afwijkingen bij aorta pathologie, zoals bicuspide aortaklep!

Pellicia et al, EHJ 2020

Sport en aorta afwijkingen

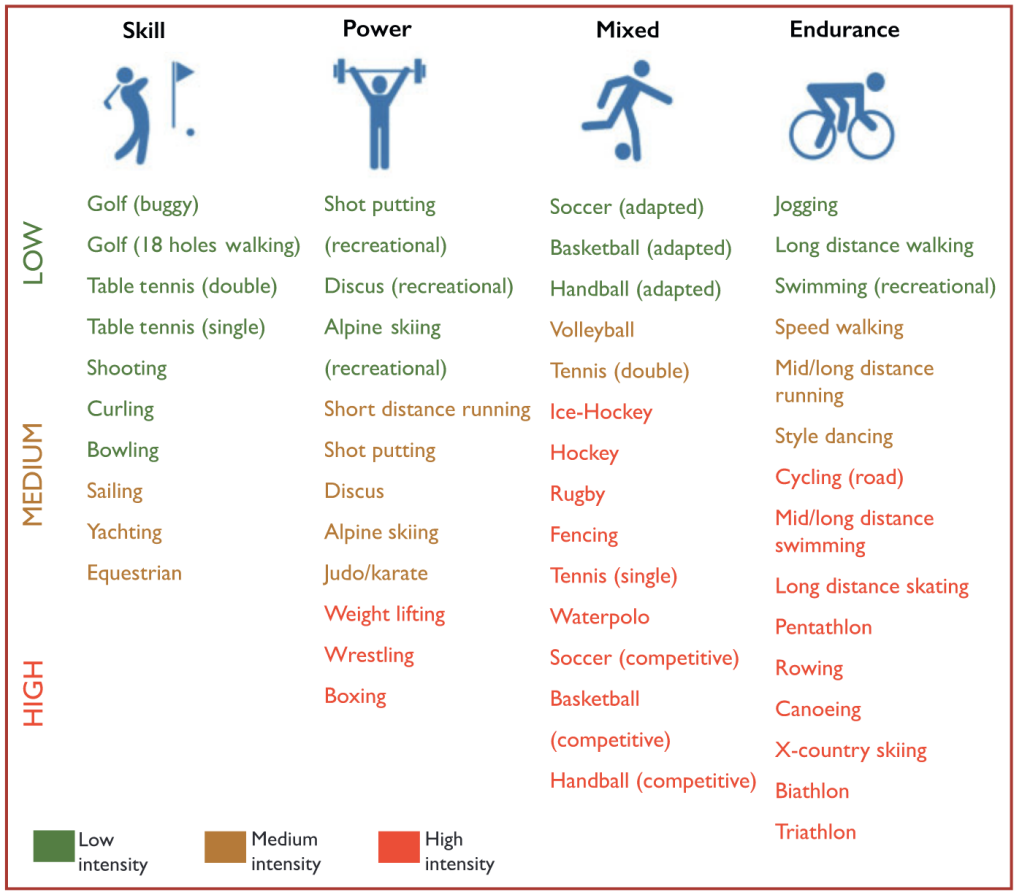


Table 14 Classification of risk to perform sports in patients with aortic pathology

	Low risk	Low-intermediate risk	Intermediate risk	High risk
Diagnosis	• Aorta <40 mm in BAV or tricuspid valve • Turner syndrome without aortic dilatation	• MFS or other HTAD syndrome without aortic dilatation • Aorta 40–45 mm in BAV or tricuspid valve • After successful thoracic aorta surgery for BAV or other low risk situation	• Moderate aortic dilatation (40–45 mm in MFS or other HTAD; 45–50 mm in BAV or tricuspid valve, Turner syndrome ASI 20–25 mm/m², tetralogy of Fallot <50 mm) • After successful thoracic aorta surgery for MFS or HTAD	• Severe aortic dilatation (>45 mm in MFS or other HTAD, >50 mm in BAV or tricuspid valve, Turner syndrome ASI >25 mm/m², tetralogy of Fallot >50 mm) • After surgery with sequelae
Advice	• All sports permitted with preference for endurance over power sports	• Avoid high and very high intensity exercise, contact, and power-sports. • Preference for endurance over power sports	• Only skill sports or mixed or endurance sports at low intensity	• Sports are (temporarily) contra-indicated
Follow-up	Every 2–3 years	Every 1–2 years	Every 6 months to 1 year	Re-evaluation after treatment

Pellicia et al, EHJ 2020

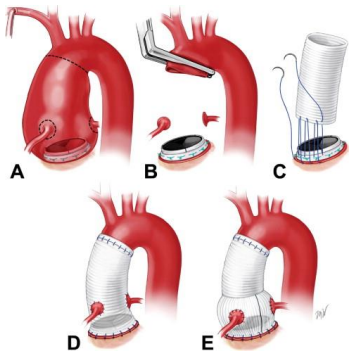
Behandeling van aorta afwijkingen

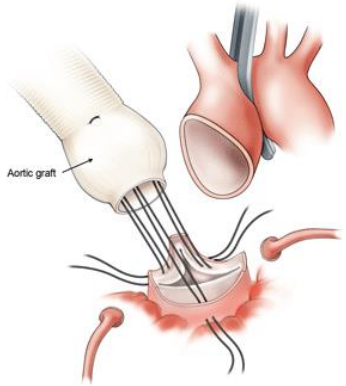
- Algemene behandeling
  - Optimale behandeling van de cardiovasculaire risicofactoren
  - Medicamenteuze behandeling: reductie van de shear stress
    - Streef bloeddruk <140/90 mmHg
    - Specifieke medicamenteuze overwegingen bij Marfan of LD syndroom
      - Geen bewijs in degeneratief aorta lijden
    - Beweegadvies voor alle patiënten

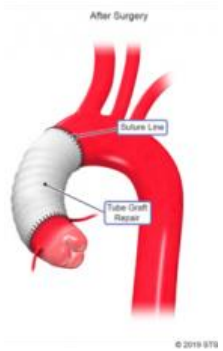
Behandeling van aorta afwijkingen

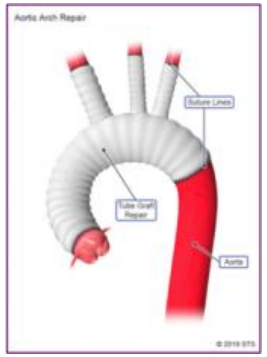
Chirurgische behandeling

- Indicatie aorta ascendens vervanging: 55 mm in afwezigheid van risicofactoren
  - Mortaliteit electieve chirurgie aorta ascendens/boog 2.4%-3.0%
  - <55 jaar mortaliteit 1.2% met stroke rate 0.6%-1.2%









Erbel et al, EHJ 2014
Isselbacher et al, Circulation 2022

6.5.1. Surgery for Sporadic Aneurysms of the Aortic Root and Ascending Aorta

Recommendations for Surgery for Sporadic Aneurysms of the Aortic Root and Ascending Aorta
Referenced studies that support the recommendations are summarized in the Online Data Supplement.

COR	LOE	Recommendations
1	C-LD	1. In patients with aneurysms of the aortic root and ascending aorta who have symptoms attributable to the aneurysm, surgery is indicated.[1,2]
1	B-NR	2. In asymptomatic patients with aneurysms of the aortic root or ascending aorta who have a maximum diameter of ≥5.5 cm, surgery is indicated.[3-9]
1	C-LD	3. In patients with an aneurysm of the aortic root or ascending aorta of <5.5 cm, whose growth rate confirmed by tomographic imaging is ≥0.3 cm/y in 2 consecutive years, or ≥0.5 cm in 1 year, surgery is indicated.[10-13]
2a	B-NR	4. In asymptomatic patients with aneurysms of the aortic root or ascending aorta who have a maximum diameter of ≥5.0 cm, surgery is reasonable when performed by experienced surgeons in a Multidisciplinary Aortic Team.[14-17]

Behandeling van aorta afwijkingen

Specifieke overwegingen bij syndromale aorta afwijkingen

Recommendations	Class[a]	Level[b]
Surgery is indicated in patients who have aortic root aneurysm, with maximal aortic diameter[c] ≥50 mm for patients with Marfan syndrome.	I	C
Surgery should be considered in patients who have aortic root aneurysm, with maximal ascending aortic diameters: • ≥45 mm for patients with Marfan syndrome with risk factors.[d] • ≥50 mm for patients with bicuspid valve with risk factors.[e,f] • ≥55 mm for other patients with no elastopathy.[g,h]	IIa	C
Lower thresholds for intervention may be considered according to body surface area in patients of small stature or in the case of rapid progression, aortic valve regurgitation, planned pregnancy, and patient's preference.	IIb	C

Erbel et al, EHJ 2014
Isselbacher et al, Circulation 2022

Tabel 8. Criteria voor het overwegen van een preventieve aorta ascendens vervanging.

Kind < 16 jaar	Volwassene > 16 jaar
z-score >3*	ASI >2.5 cm/m²***
z-score toename >1/jaar	Toename absolute diameter >3mm/jaar
Aanwezigheid van risicofactoren** (in combinatie met aortadilatatie)	Aanwezigheid van risicofactoren** (in combinatie met aortadilatatie)

Turner

Recommendations for Replacement of the Aorta in Patients With Loeys-Dietz Syndrome

COR	LOE	Recommendations
1	C-LD	1. In patients with Loeys-Dietz syndrome and aortic dilation, the surgical threshold for prophylactic aortic root and ascending aortic replacement should be informed by the specific genetic variant, aortic diameter, aortic growth rate, extra-aortic features, family history, patient age and sex, and physician and patient preferences (Table 11).[1-9]
2b	C-EO	2. In patients with Loeys-Dietz syndrome attributable to a pathogenic variant in *TGFBR1*, *TGFBR2*, or *SMAD3*, surgery to replace the intact aortic arch, descending aorta, or abdominal aorta at a diameter of ≥4.5 cm may be considered, with the specific genetic variant, patient age, aortic growth rate, family history, presence of high-risk features (Table 11), and surgical risk informing the decision.

Table 11. Surgical Thresholds for Prophylactic Aortic Root and Ascending Aortic Replacement in Loeys-Dietz Syndrome Based on Genetic Variant

COR	LOE (references)	Genetic Variant	Presence of High-Risk Features*	Aortic Diameter (cm)
1	C-LD[2]	*TGFBR1*	No	≥4.5
1	C-LD[2]	*TGFBR2*	No	≥4.5
2b	C-EO[7]	*TGFBR1*	Yes	≥4.0
2a	C-LD[1,2]	*TGFBR2*	Yes	≥4.0
2a	C-EO[13,16]	*SMAD3*	–	≥4.5†
2b	C-EO[5-7]	*TGFB2*‡	–	≥4.5†
2b	C-EO[8,23]	*TGFB3*	–	≥5.0†

Conclusie

- Thoracaal aorta aneurysma relatief zeldzaam
  - Aorta dimensies nemen toe met de leeftijd en hoe groter de diameter, hoe sneller de groei
  - Risico op scheuren neemt dramatisch toe boven diameter van 6 cm
- Behandeling patiënten met aorta aneurysma is levensloop zorg: specifieke aandacht voor zwangerschap en sport
- Profylactische chirurgie verdient de voorkeur! Cut-off niet syndromale patiënten >55 mm, lagere cut-offs voor syndromale aorta afwijkingen

ACAHA
Academic Centre for Congenital Heart Disease

Erasmus MC