conduite à tenir devant une AO thoracique dilatée

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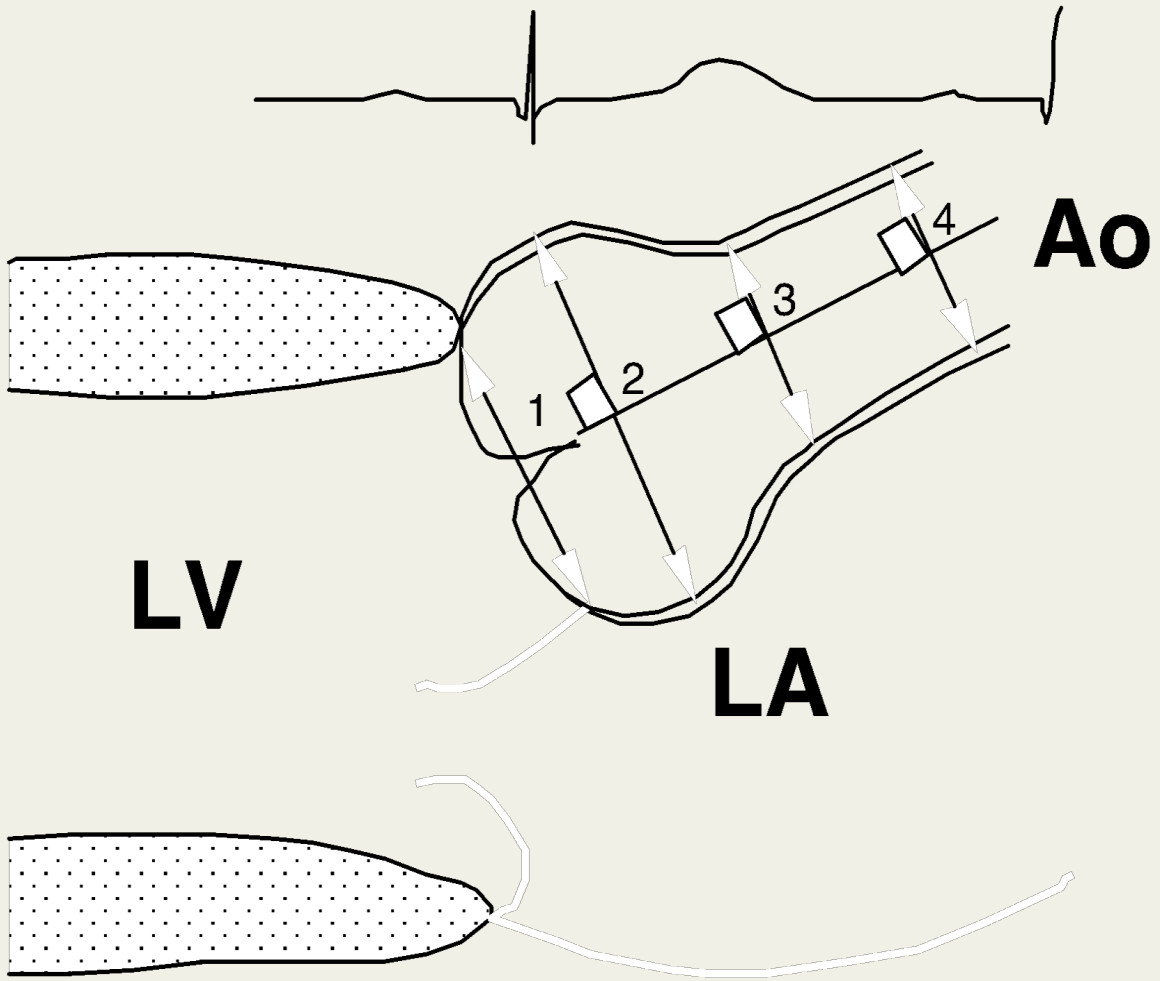
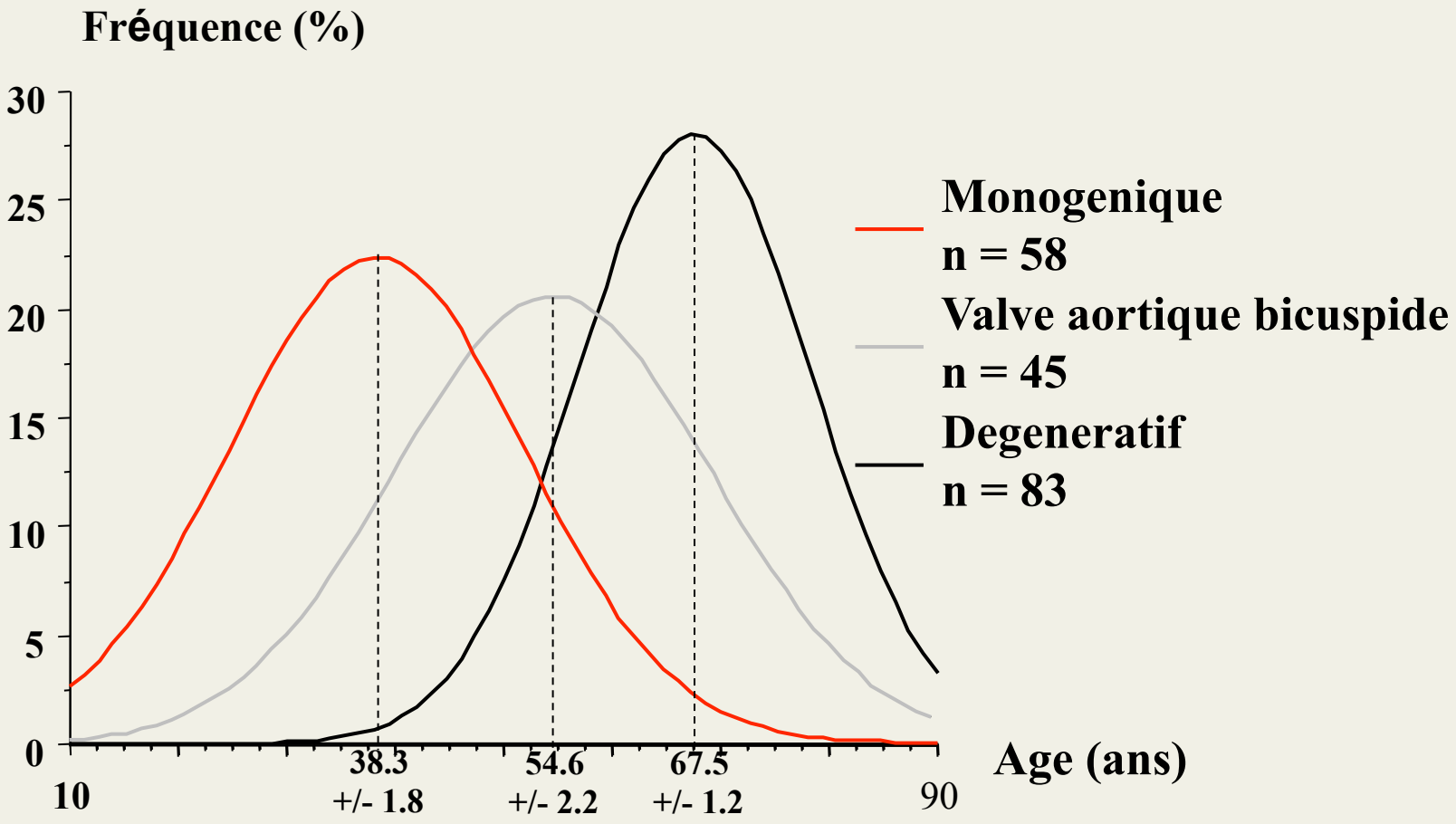
Les questions

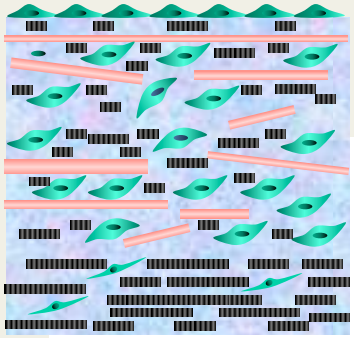
1) Quelle étiologie ?
    1) Type dilatation
    2) Signes extra-aortiques
    3) Enquête familiale
2) Quel traitement ? (quel risque)
    1) Mode de vie
    2) Médical
    3) Chirurgical
3) Quel suivi ?

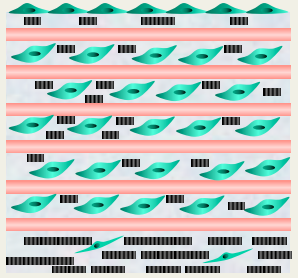
Diamètre aortique



Age lors de la chirurgie







MFS
(*FBN1*)

TAAD: familial Thoracic Aortic Aneurysm and Dissection

Revised Ghent criteria for Diagnosis of Marfan syndrome and Related conditions.

J Med Genet. 2010 Jul;47(7):476-85

En l'absence d'histoire familiale:

Ao (Z≥2) ET

FBN1 = MFS

EL = MFS

Syst (≥7pts) = MFS*

EL avec ou sans Syst ET

FBN1 avec Pb Ao = MFS

FBN1 sans Pb Ao connu ou pas *FBN1*= ELS

Ao (Z< 2) ET Syst (≥5) sans EL = MASS

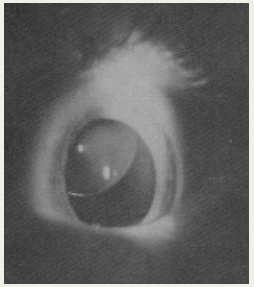
PVM ET Ao (Z<2) ET Syst (<5) sans EL = Syndrome PVM

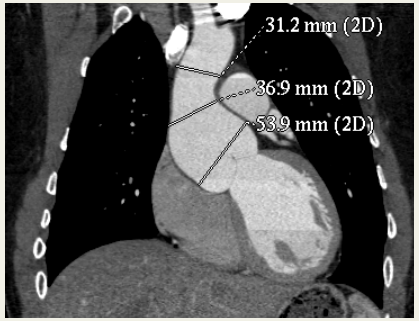
En cas d'histoire familiale:

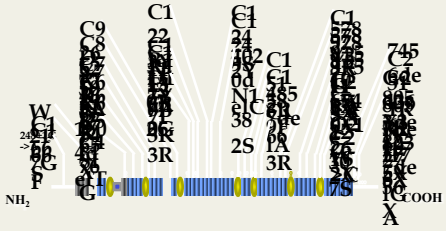
Ao (Z≥2 si + de 20 ans, ≥3 si – de 20 ans) = MFS

EL = MFS

Syst (≥7 pts) = MFS







Score systémique

Pectus

- Carinatum : **2**
- excavatum ou asymétrie thoracique : **1**

Signe du poignet ET du pouce : **3**

- Signe du poignet OU du pouce : **1**

Scoliose ou cyphose thoraco-lombaire : **1**

- Pas de scoliose sévère et ↓ US/LS et ↑ envergure / taille : **1**

Protrusio acetabuli : **2**

↓ extension coudes : 1

Anomalie arrière pied : **2**

- Pied plat : **1**

Anomalies faciales (3/5) : **1**

- dolichocephalie, enophtalmie, fentes palpébrales en bas et dehors, hypoplasie malaire, retrognathisme)

Ectasie durale : **2**

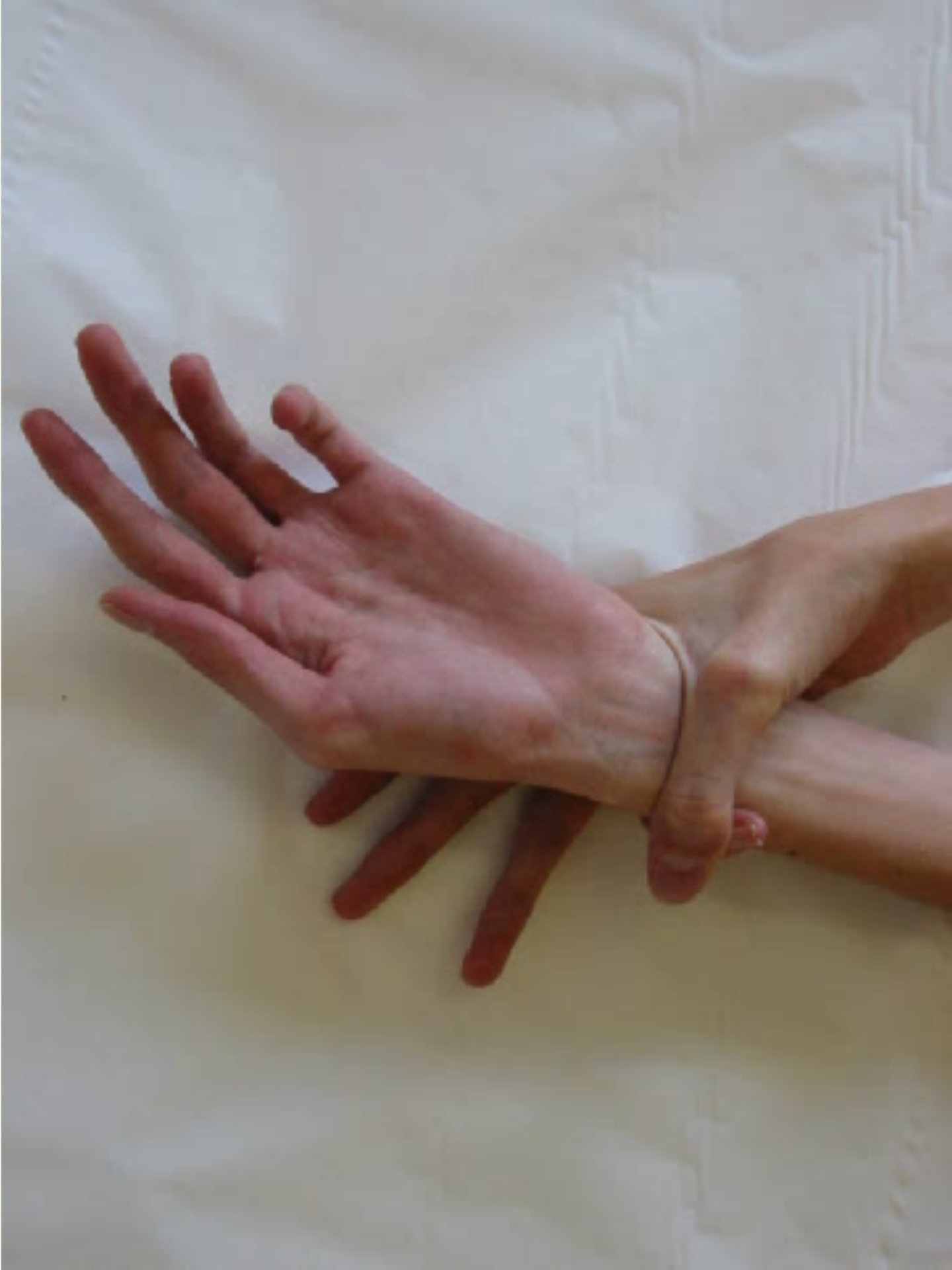
Pneumothorax : **2**

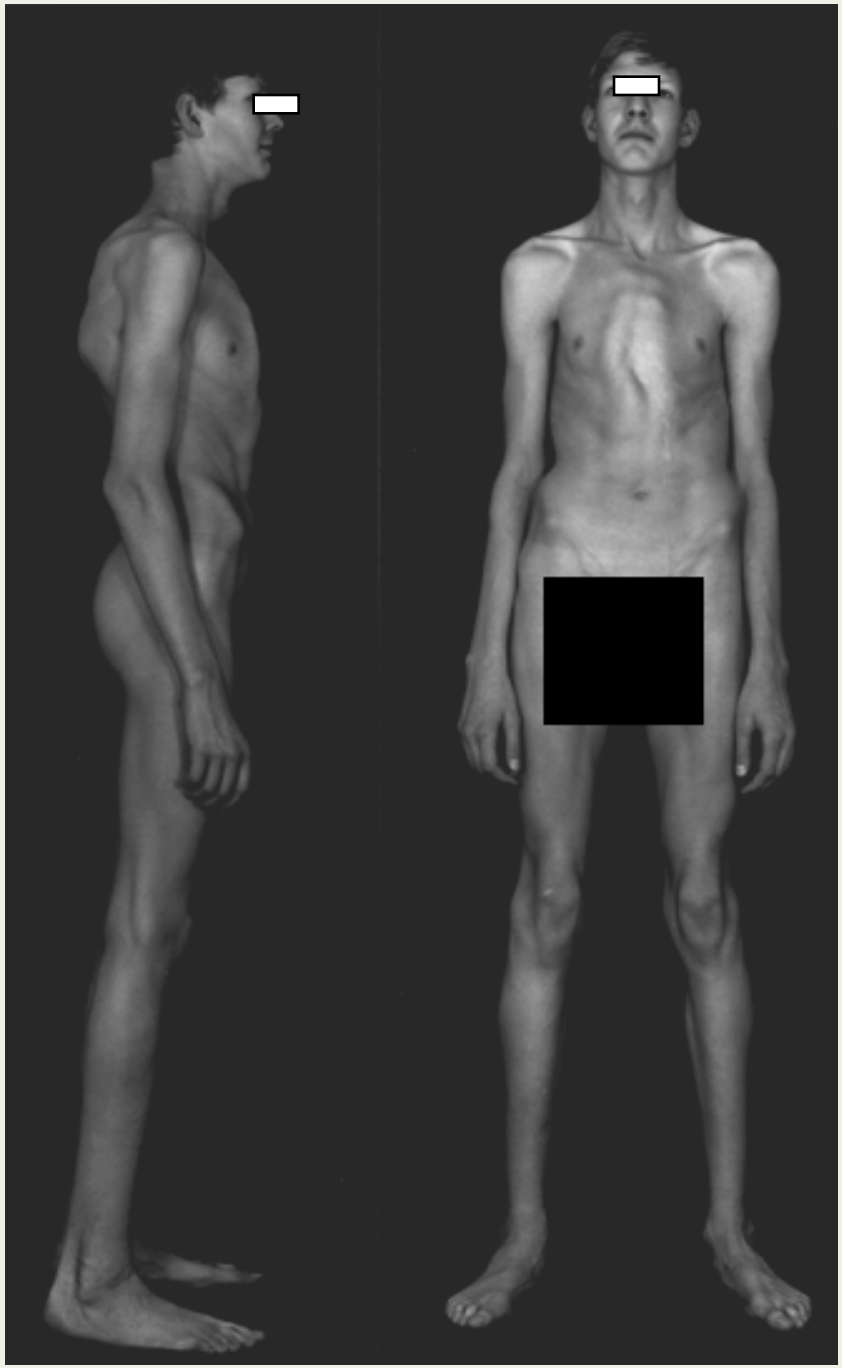
Vergetures: **1**

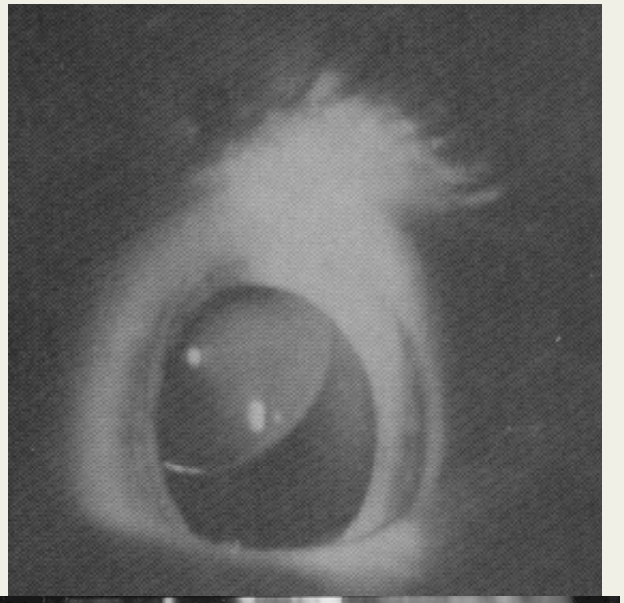
Myopie > 3 dioptries : **1**

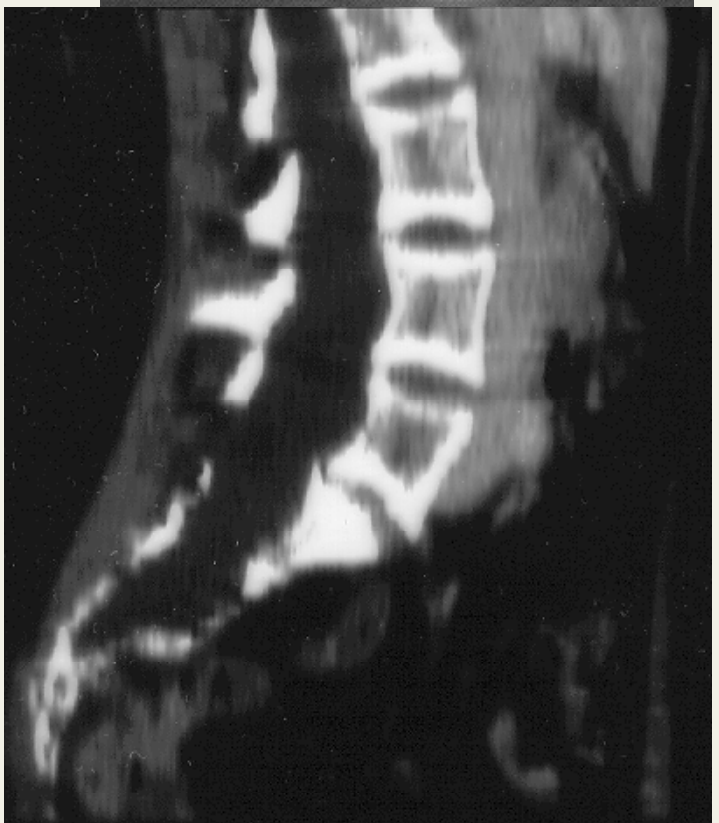
PVM : **1**

Total maximum : 20 points; score ≥7 indique une atteinte systémique

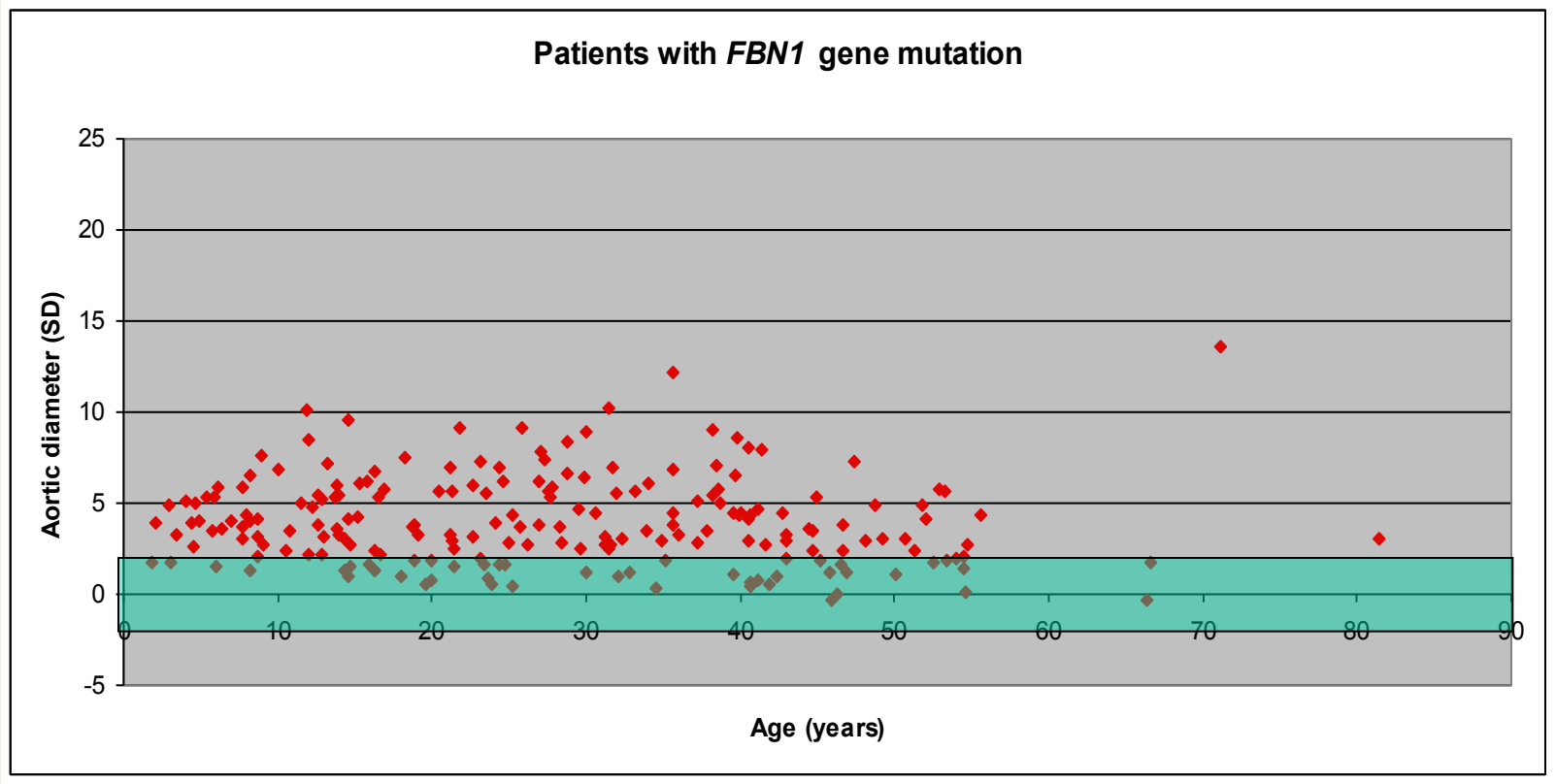






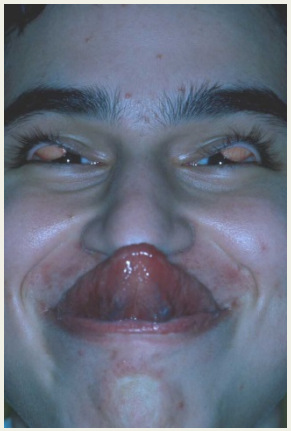


Difficile : grande variabilité

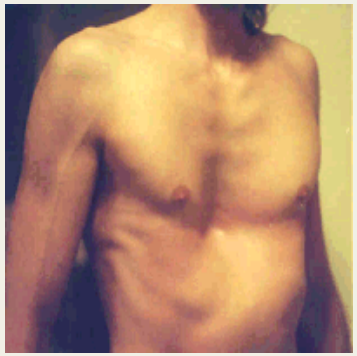


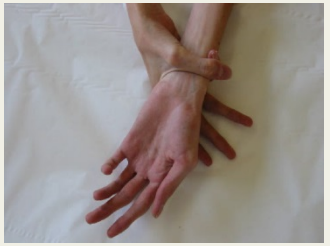
D Attias... G Jondeau (Circulation 2009;120:2541-2549)

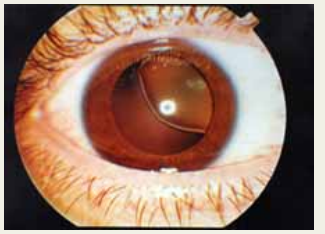
Syndromic forms of thoracic aortic aneurysm



EDS
(*COL3A1*)



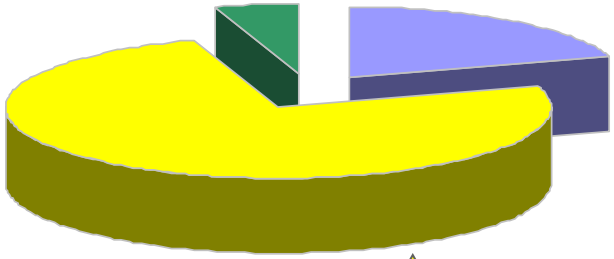




Familial cases

syndromic
<5%

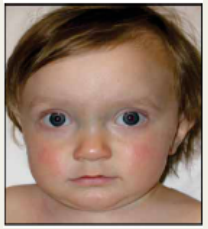
MFS
(*FBN1*)

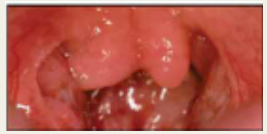


Sporadic cases
80%

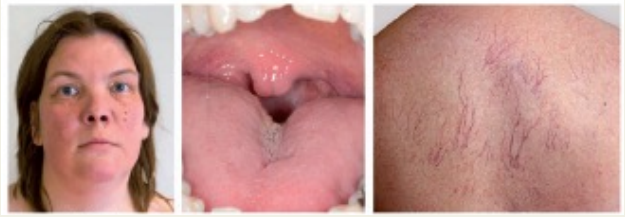
MFS2
(*TGFBR2*)

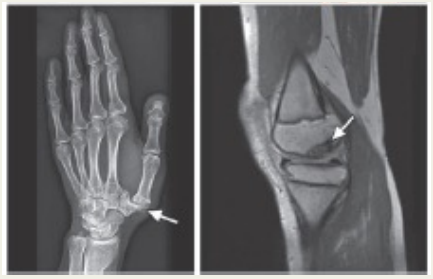
LDS
(*TGFBR1&2*)



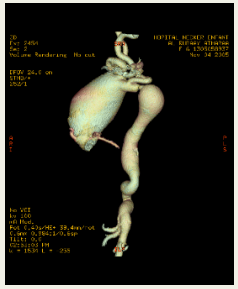


Aortic Osteoarthritis syndrome



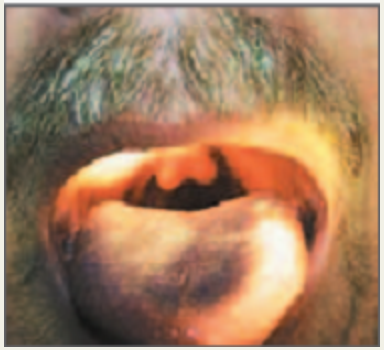


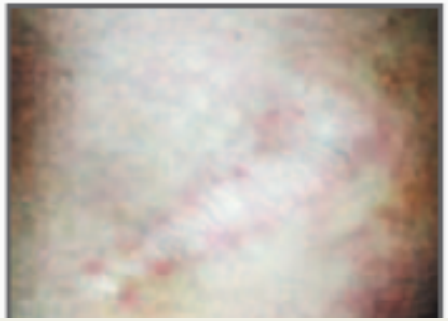
(*SMAD3*)

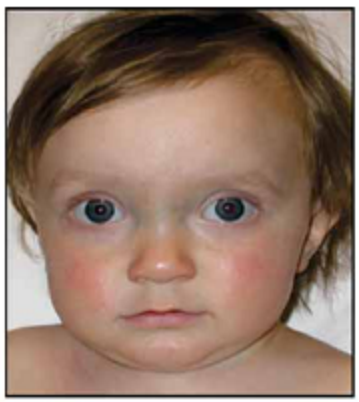


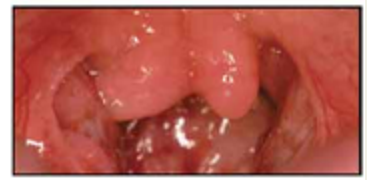
Van de Laar et al.,, 2011; Aubart et al., 2014

NEJM 2006;355:8
Loeys Dietz Syndrome

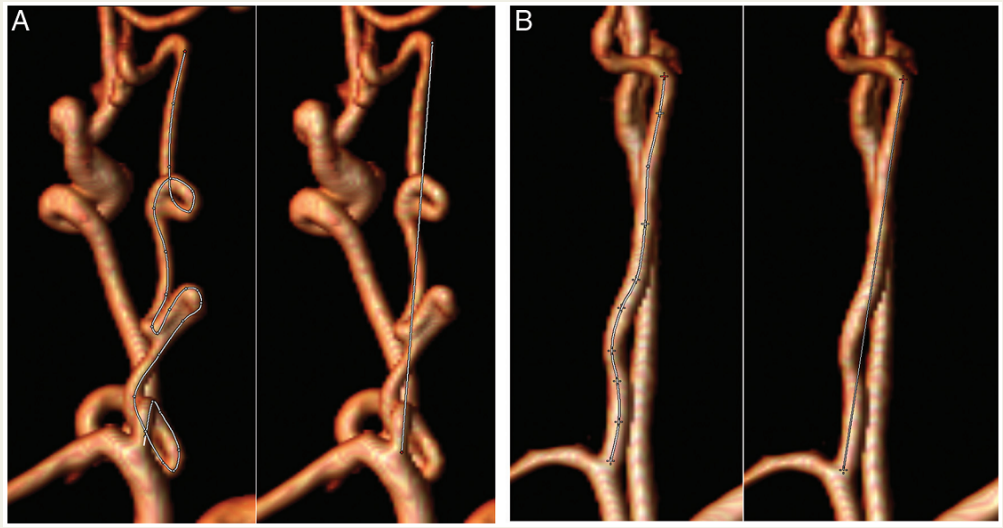


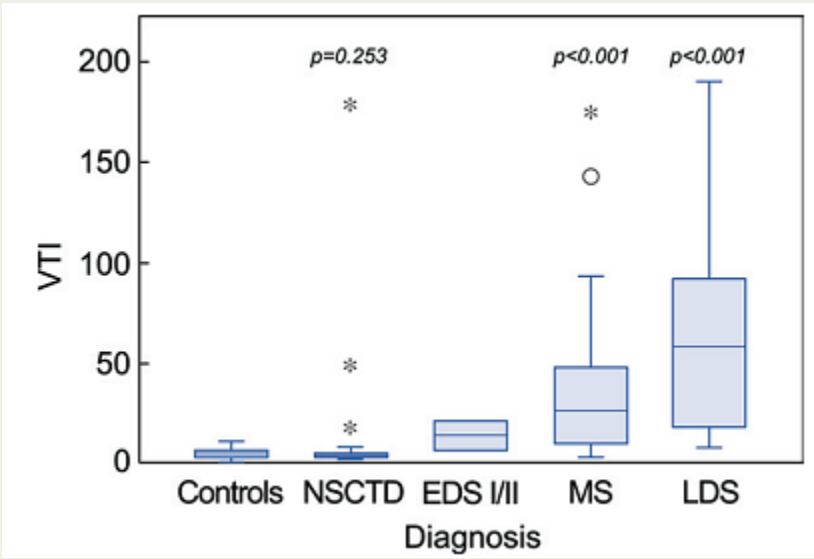


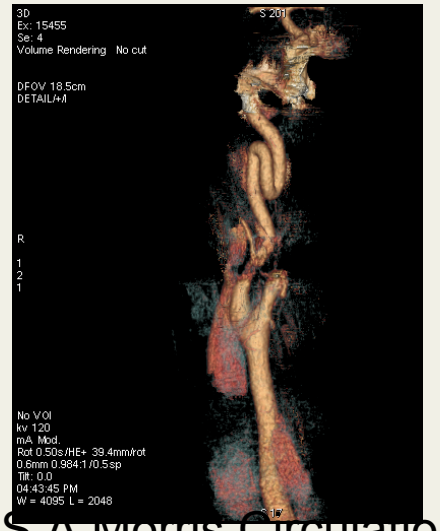


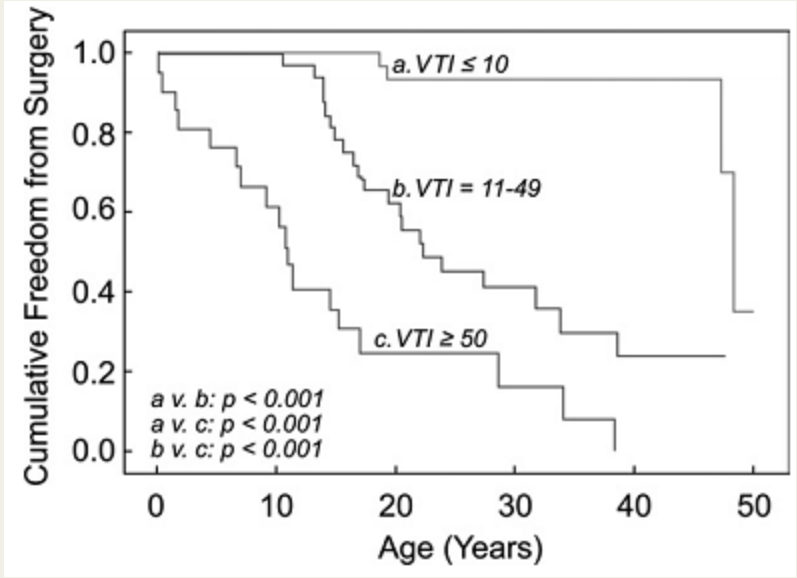


Tortuosité artérielle (vertébrale)

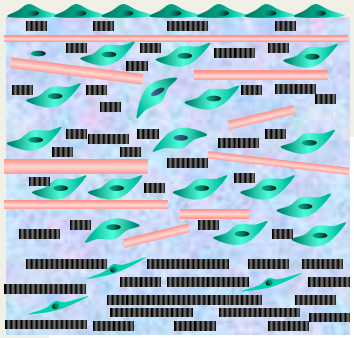


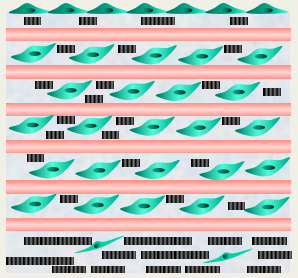






S.A.Morris Circulation.2011;124:388-396

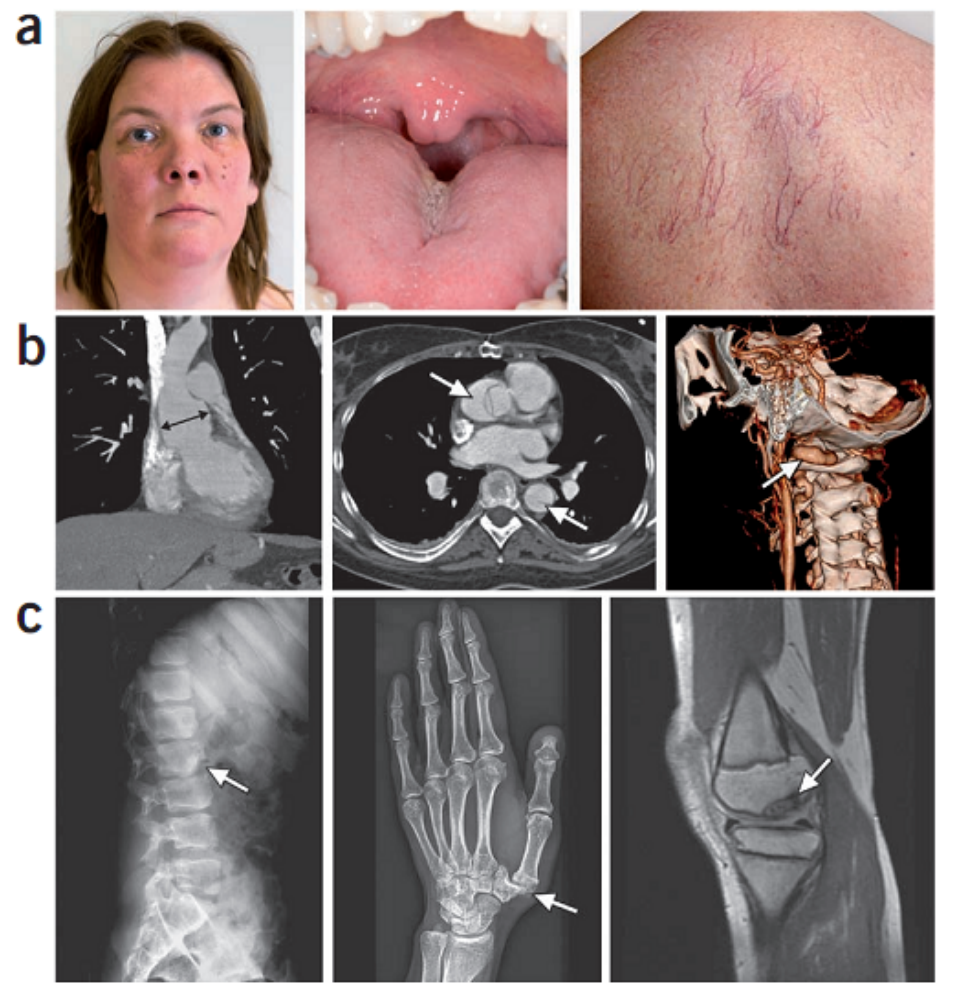




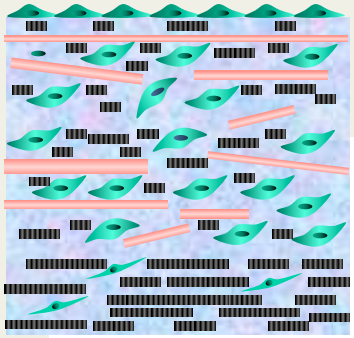
MFS
(*FBN1*)

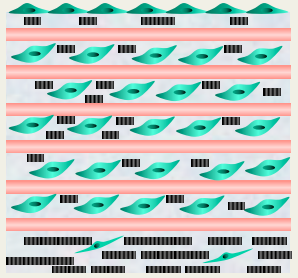
TAAD: familial Thoracic Aortic Aneurysm and Dissection

Smad 3



Ingrid M B H van de Laar Nature Genet 2011

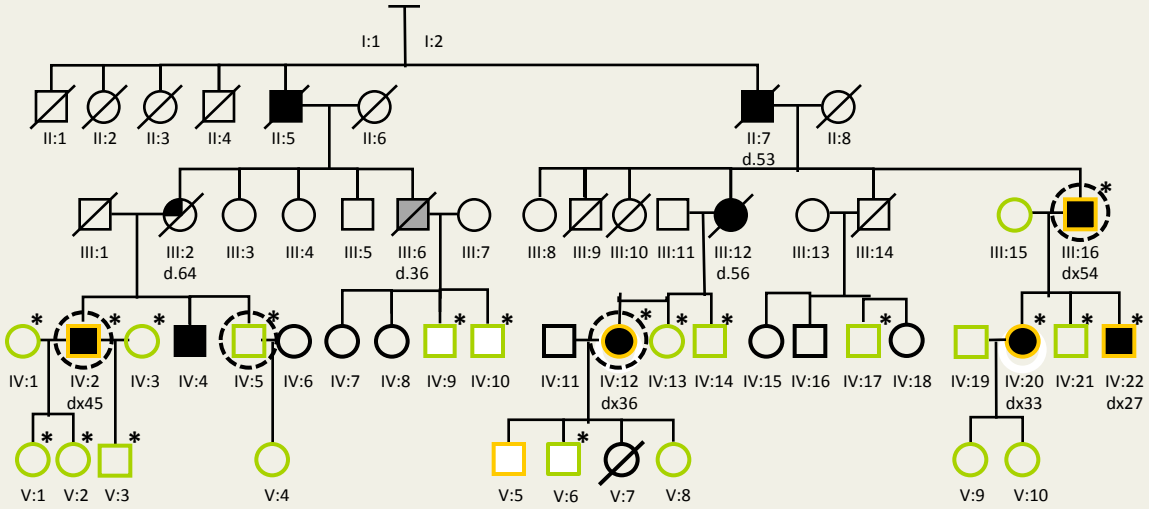




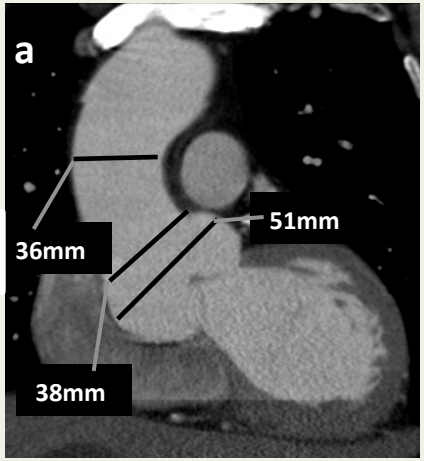
MFS
(*FBN1*)

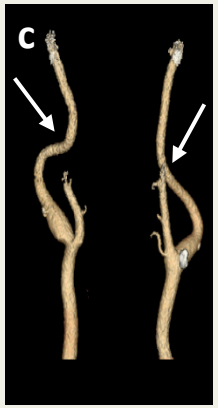
TAAD: familial Thoracic Aortic Aneurysm and Dissection

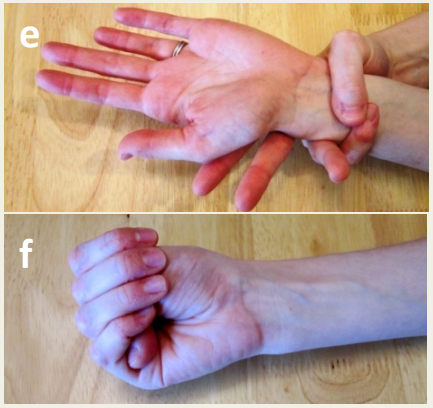
TGFB2 *Boileau, …, Jondeau*, Milewicz* Nature Genet 2012*

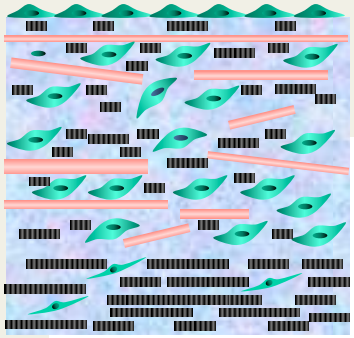


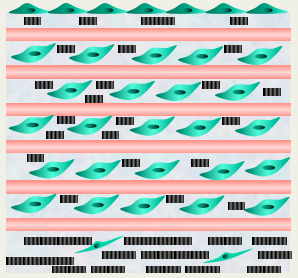
- Thoracic aortic aneurysm/dissection
- Sudden death of unknown cause
- Cerebrovascular disease
- Mutation present
- Mutation absent







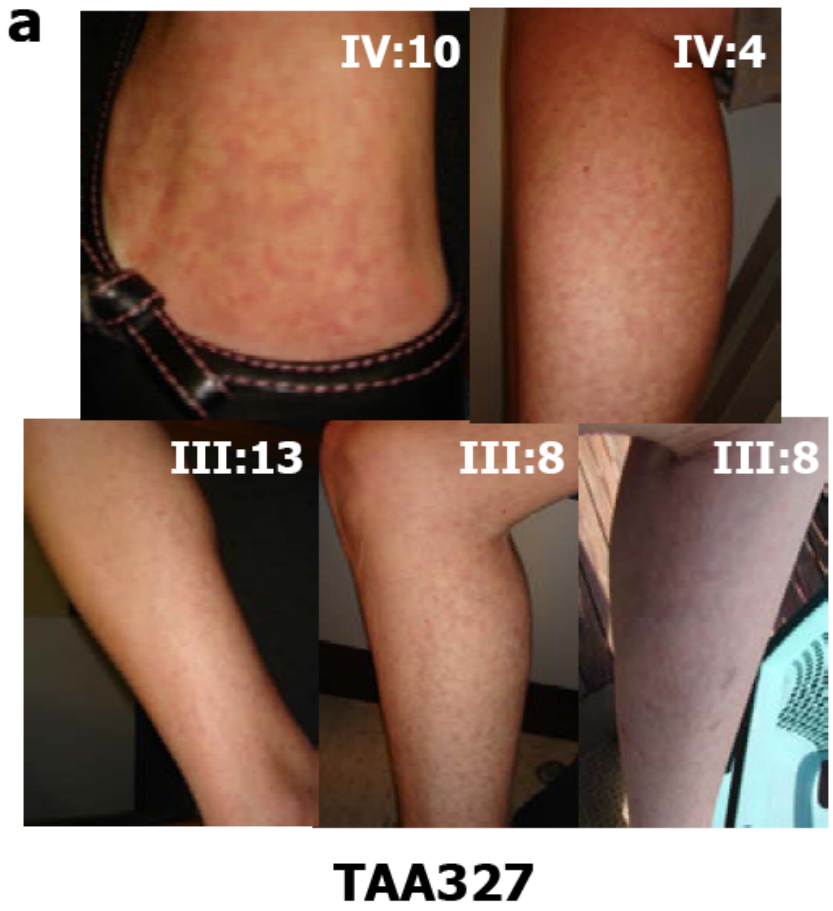


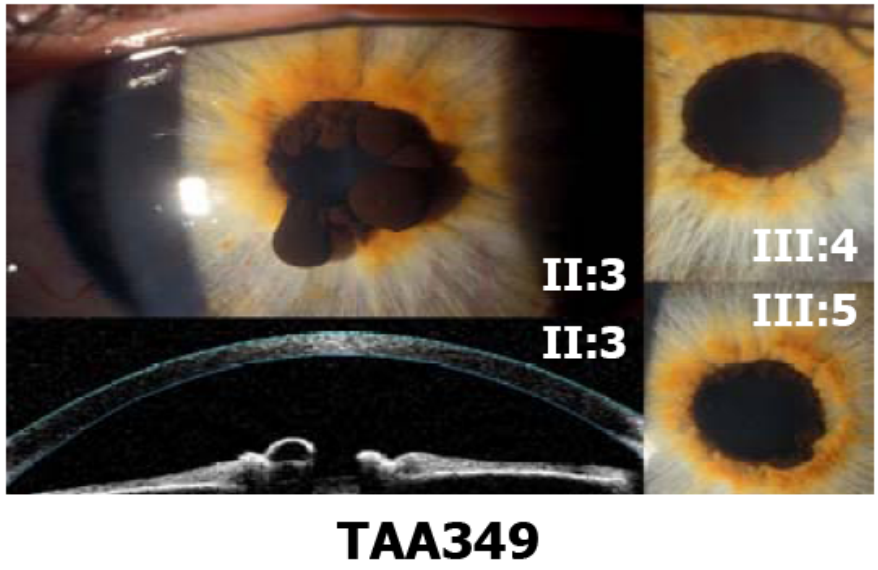


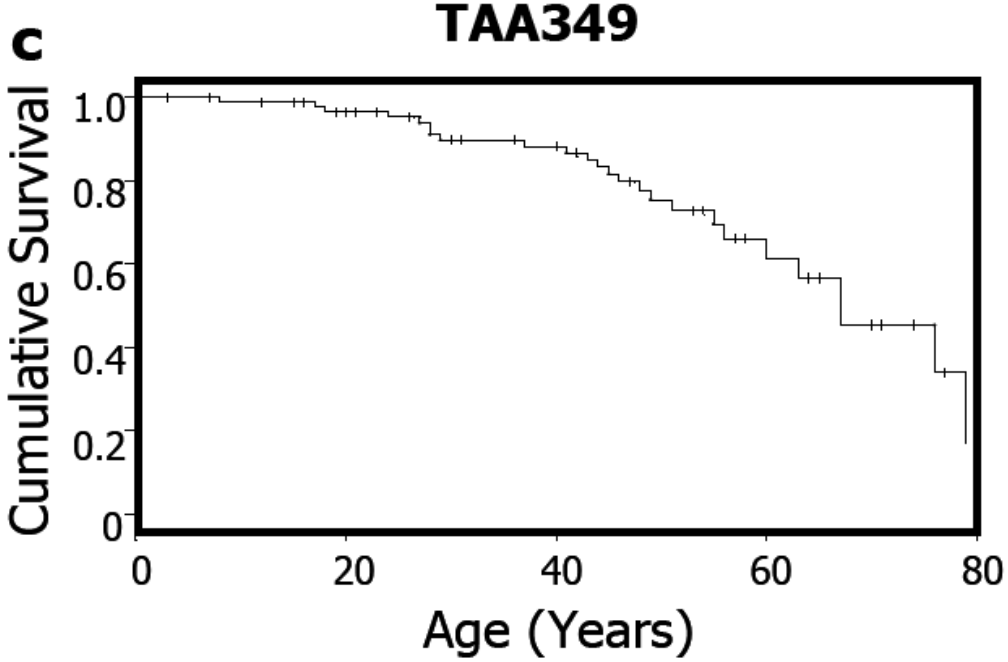
MFS
(*FBN1*)

TAAD: familial Thoracic Aortic Aneurysm and Dissection

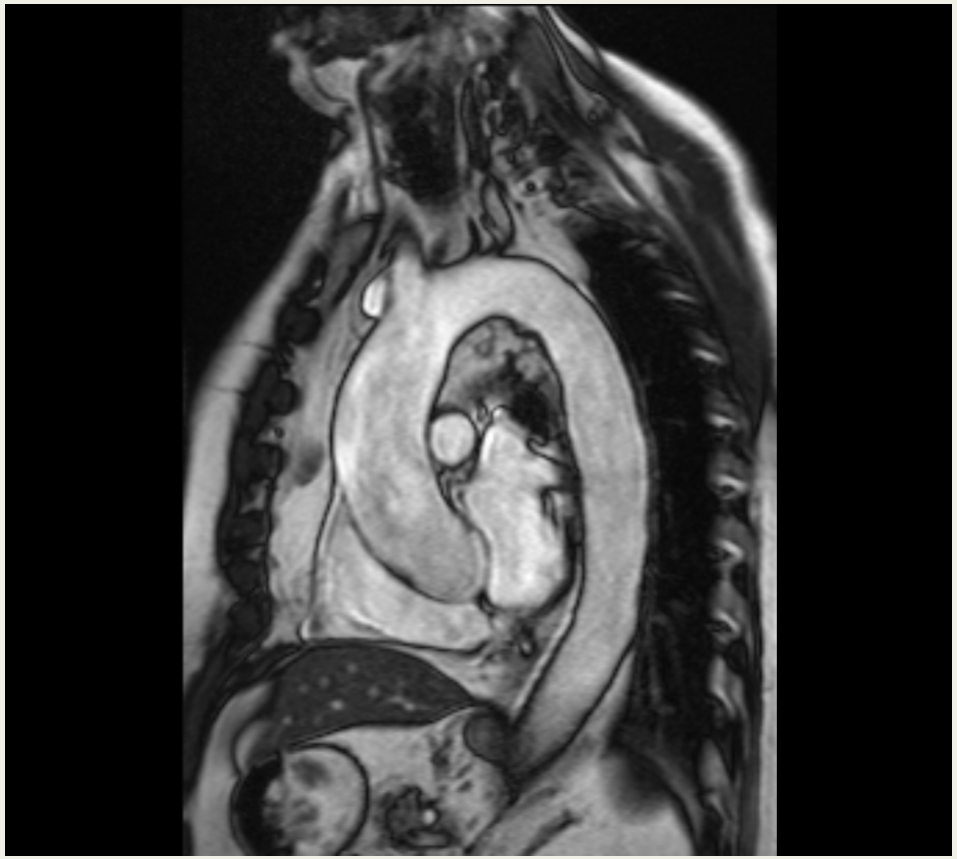
ACTA 2 Nature Genet 2007



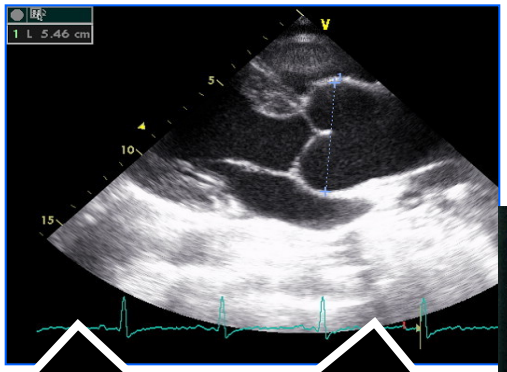




MYH11

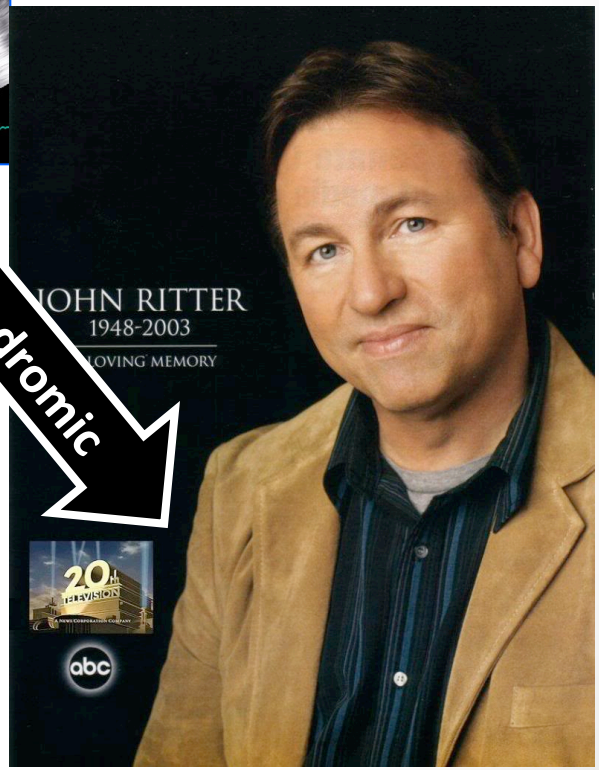


Heritable Thoracic Aortic Disorders
H-TAD



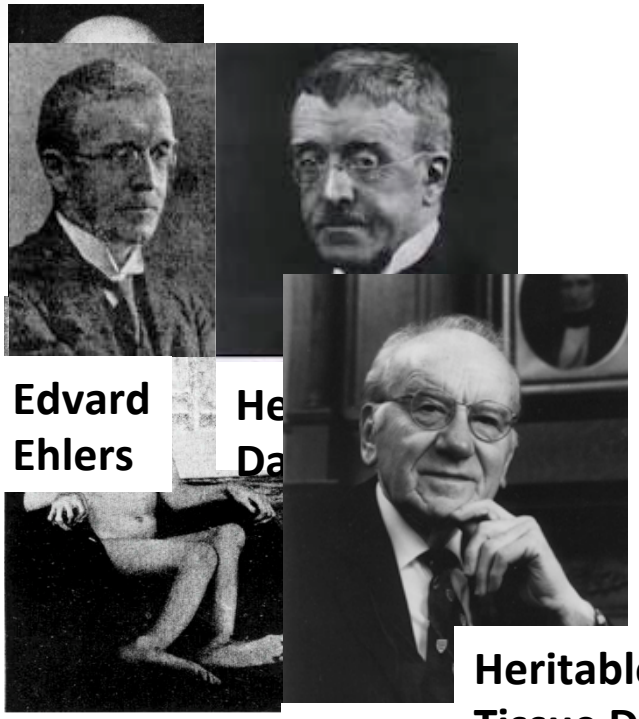
Syndromic

Nonsyndromic



Annual Symposium of the
Belgian Society for Cardiology
January 29-30 2015

H-TAD History



Edvard Ehlers

Antoine Marfa

Aortic I

Heritable Connective Tissue Disorders

1988

FBN1 M

Large coh
"muta
negati

COL3A1 mutation

Nonsyndromic TAD

FBN1
TGFBR1/2
TGFB2
TGFB3
SMAD3
ACTA2
MYH11
MYLK
PRKG1
MAT2A
MFAP5
....

Type 2
Marfan
TGFBR2

off
3

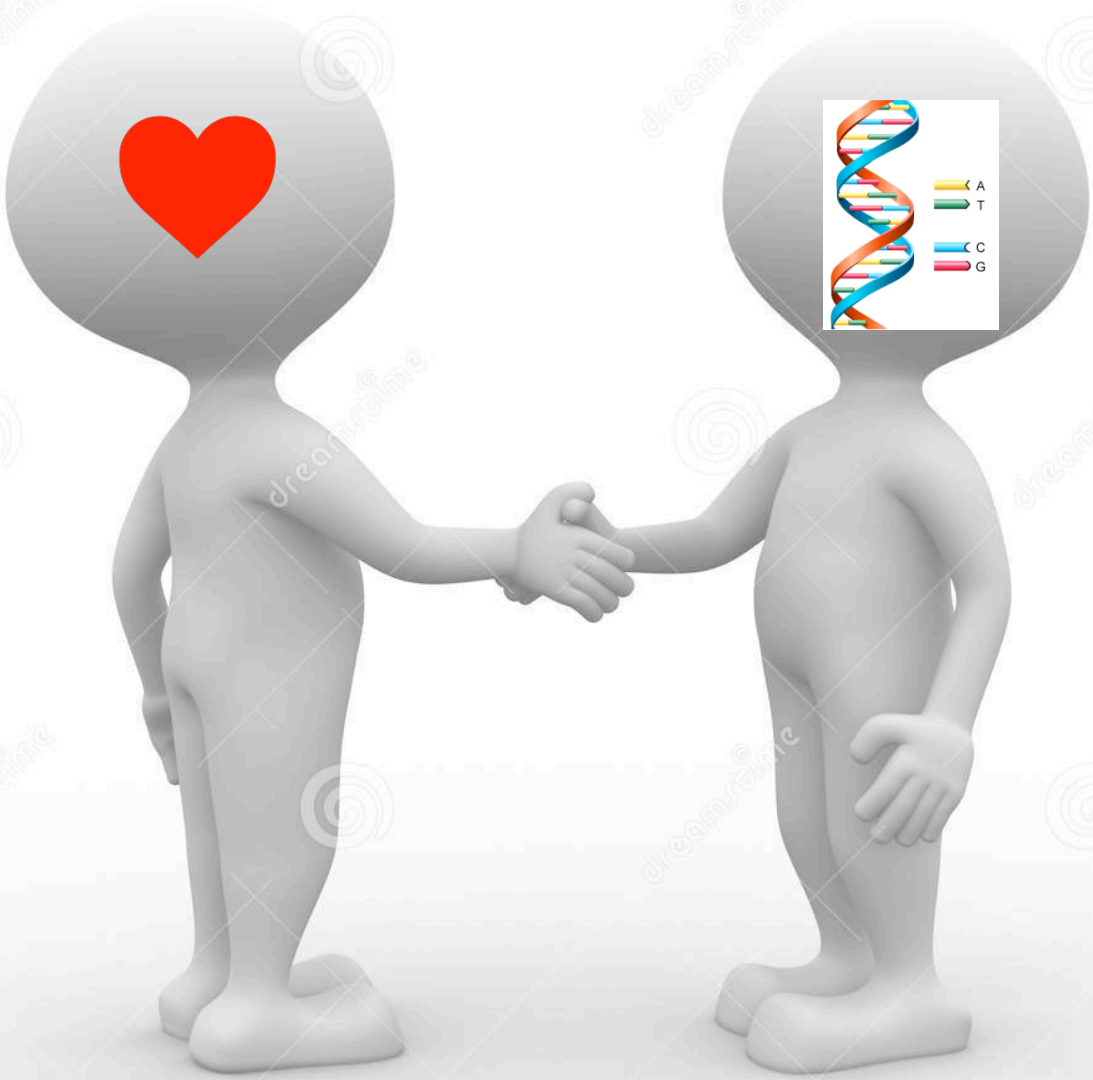
SMAD3

1895 1955 1972 1991 2004 2006 2012 2013

Annual Symposium of the Belgian Society for Cardiology January 29-30 2015

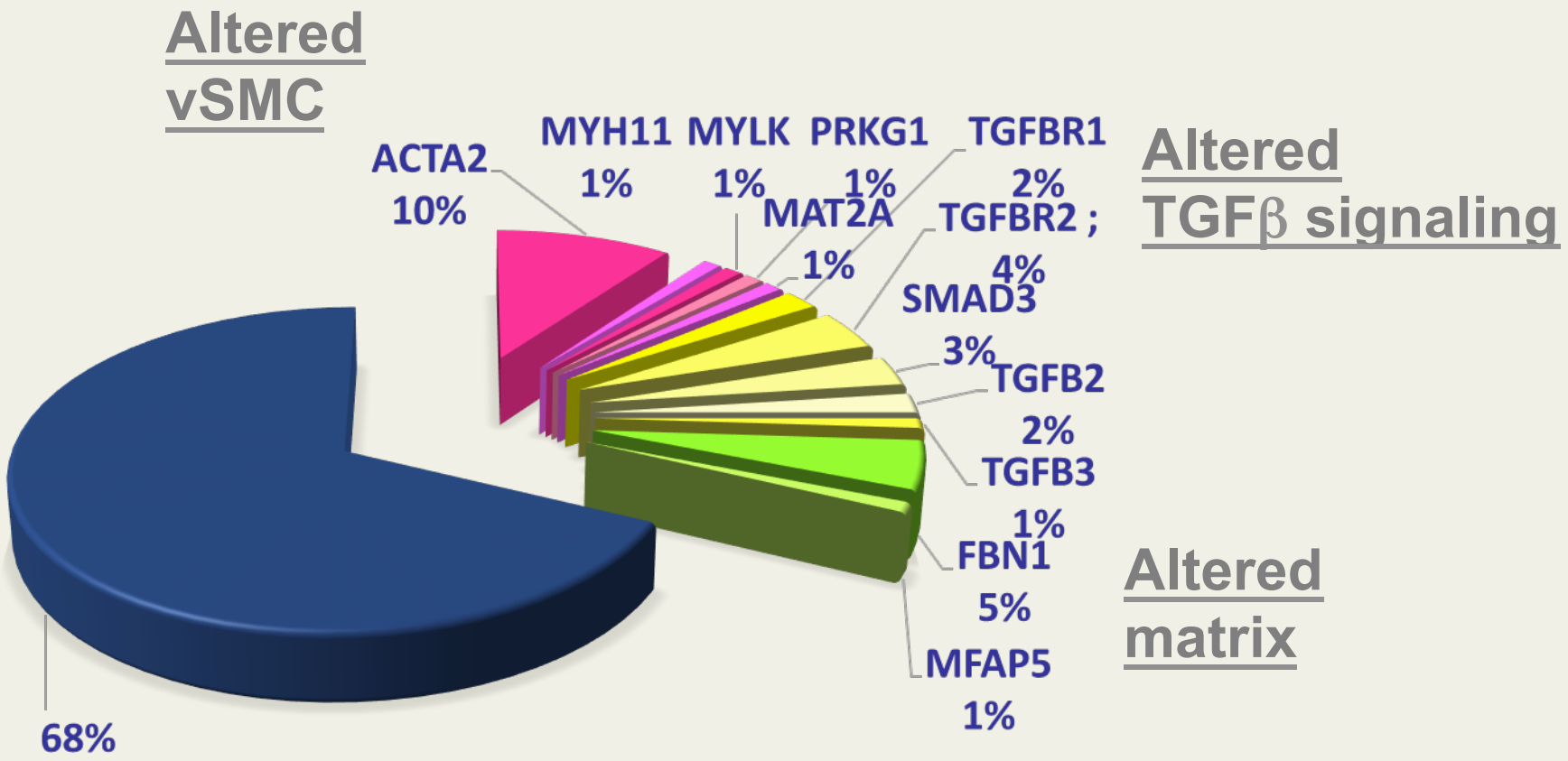
Universitair Ziekenhuis Gent

UNIVERSITEIT GENT


A
T
C
G

Take home message (2)

☑ **High level of genetic heterogeneity in familial TAA with many genes still to be identified.**



Take home message (1)

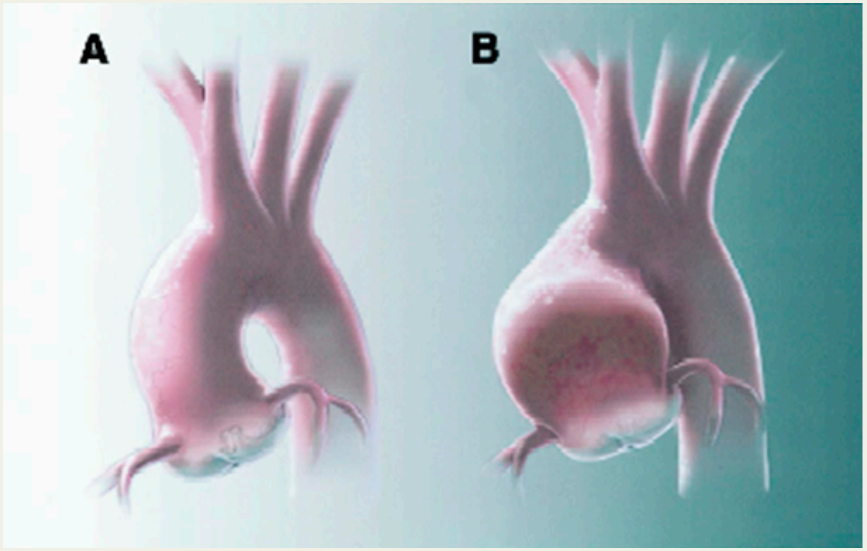
The Task Force for the Diagnosis and Treatment of Aortic Diseases of the European Society of Cardiology (ESC) Eur Heart J, 2014

Recommendations on genetic testing in aortic diseases

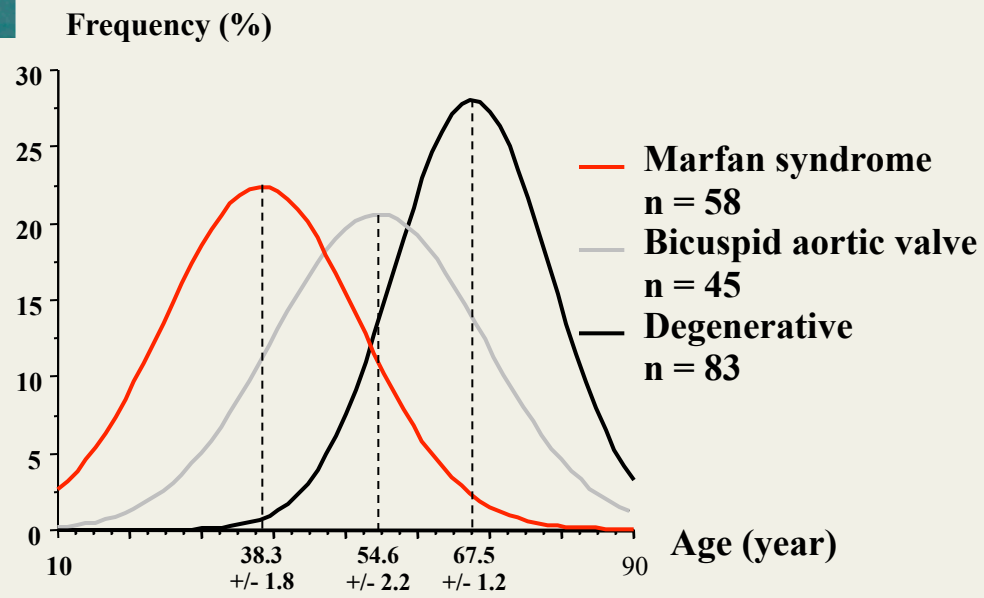
Recommendations	Class[a]	Level[b]
It is recommended to investigate first-degree relatives (siblings and parents) of a subject with TAAD to identify a familial form in which relatives all have a 50% chance of carrying the family mutation/disease.	I	C
Once a familial form of TAAD is highly suspected, it is recommended to refer the patient to a geneticist for family investigation and molecular testing.	I	C
Variability of age of onset warrants screening every 5 years of 'healthy' at-risk relatives until diagnosis (clinical or molecular) is established or ruled out.	I	C
In familial non-syndromic TAAD, screening for aneurysm should be considered, not only in the thoracic aorta, but also throughout the arterial tree (including cerebral arteries).	IIa	C

[a]Class of recommendation.
[b]Level of evidence.
TAAD = thoracic aortic aneurysms and dissection.

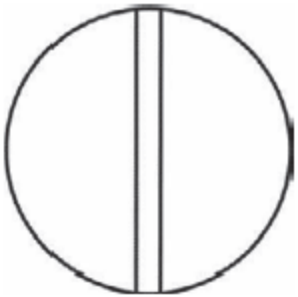
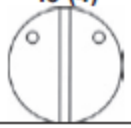
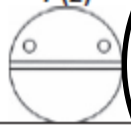
Aneurysm of the ascending aorta



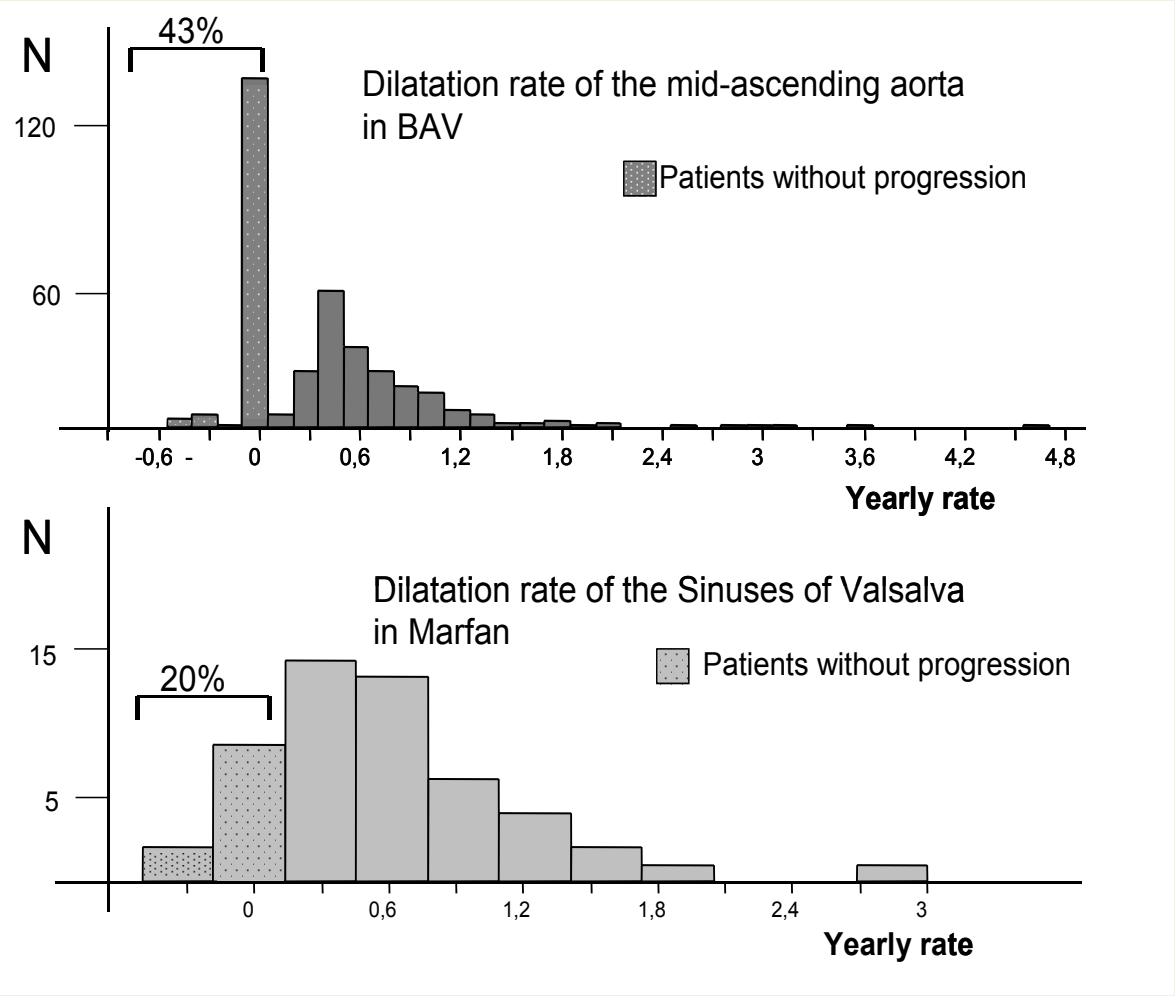
etiologic diversity



Hétérogénéité anatomique Classification Sievers JTCS 2007

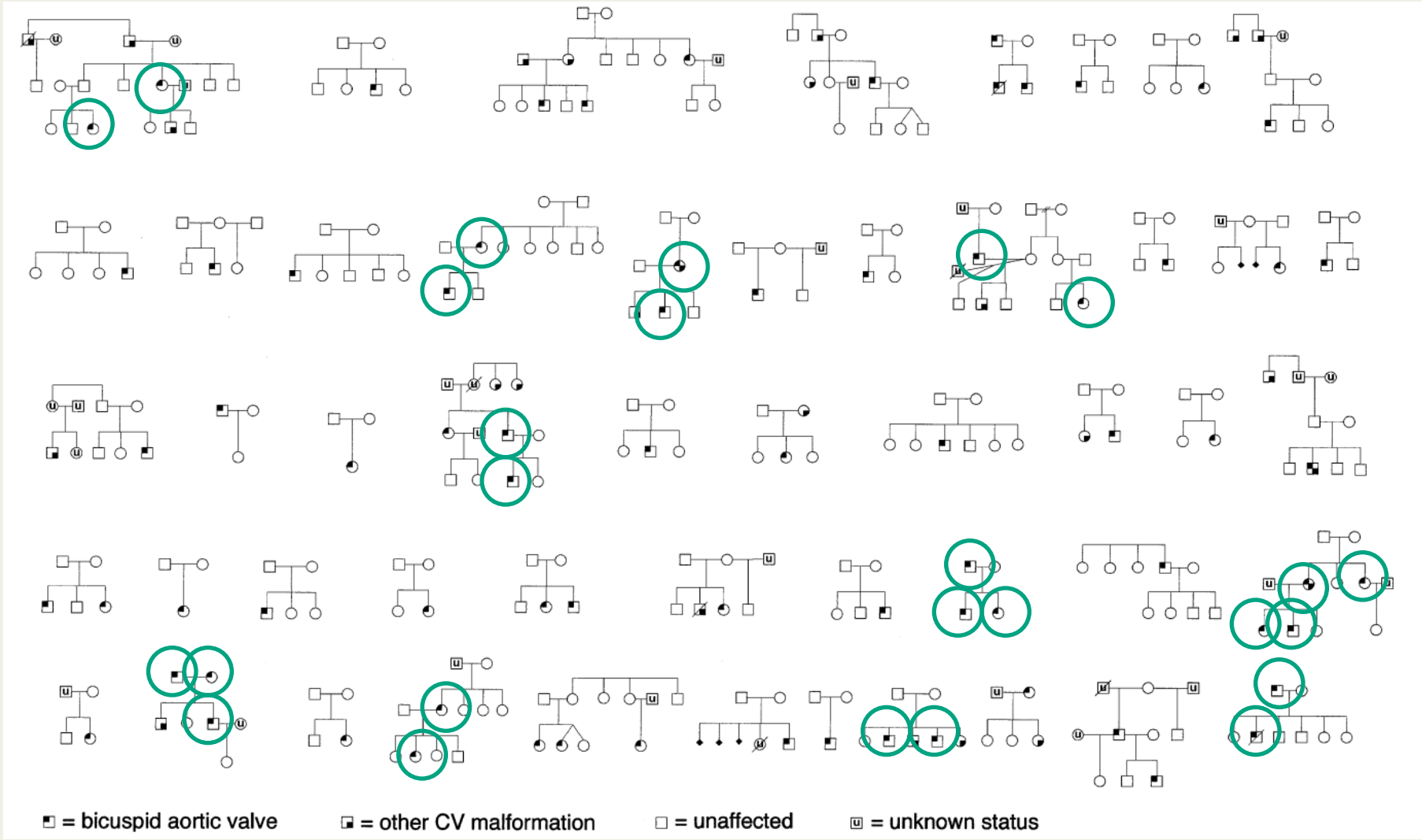
main category: number of raphes	0 raphe - Type 0  21 (7)		1 raphe - Type 1  269 (88)			2 raphes - Type 2  14 (5)
1. subcategory: spatial position of cusps in Type 0 and raphes in Types 1 and 2	lat 13 (4) 	ap 7 (2) 	L – R 216 (71) 	R – N 45 (15) 	N – L 8 (3) 	L – R / R – N 14 (5) 
2. subcategory:						
VALVULAR FUNCTION I	6 (2)	1 (0.3)	79 (26)	22 (7)	3 (1)	6 (2)
VALVULAR FUNCTION S	7 (2)	5 (2)	119 (39)	15 (5)	3 (1)	6 (2)
VALVULAR FUNCTION B (I + S)		1 (0.3)	15 (5)	7 (2)	2 (1)	2 (1)
VALVULAR FUNCTION No			3 (1)	1 (0.3)		

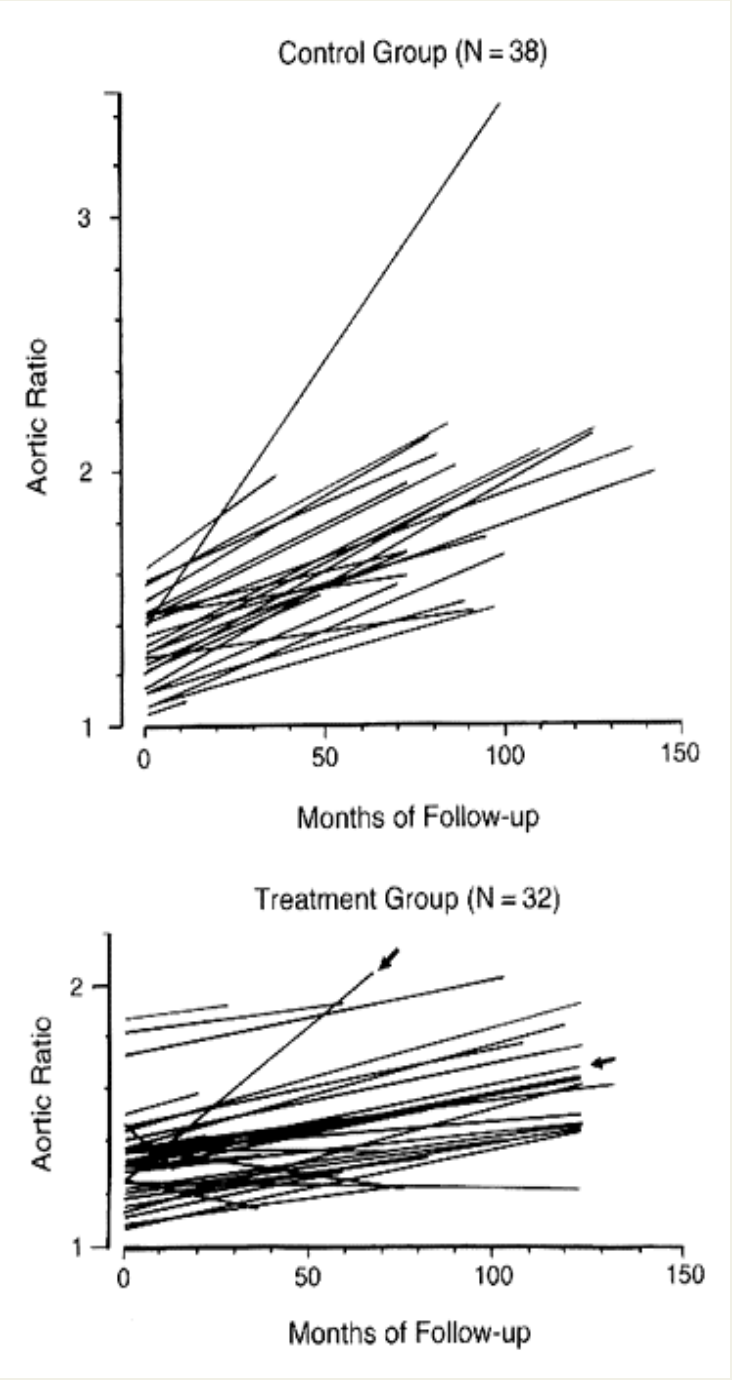
Vue du chrirurgien



D Detaint… G Jondeau, E Sarano Heart 2013

Cripe JACC 2004;44:138





ß blockade

> 11 y.o.
HR < 100 bpm exercise

	Control	ß-
death	2	0
Dissec	4	2
AR	2	2
>6cm	1	1
Total	9/38 (23%)	5/32 (16%)

Shores N Engl J Med 1994;330:1335-41

Qui traiter ?

FBN1 mutation = aortic risk

L Faivre… G Jondeau J Med Genet. 2008;45:384

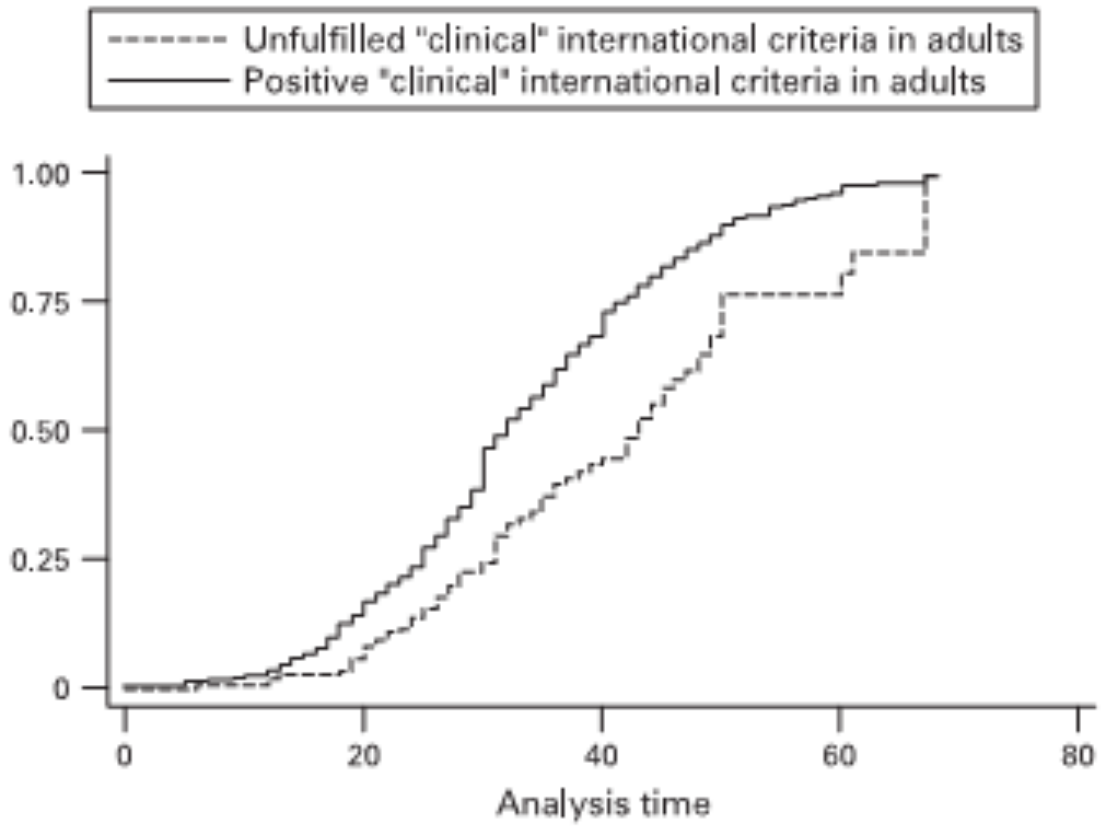
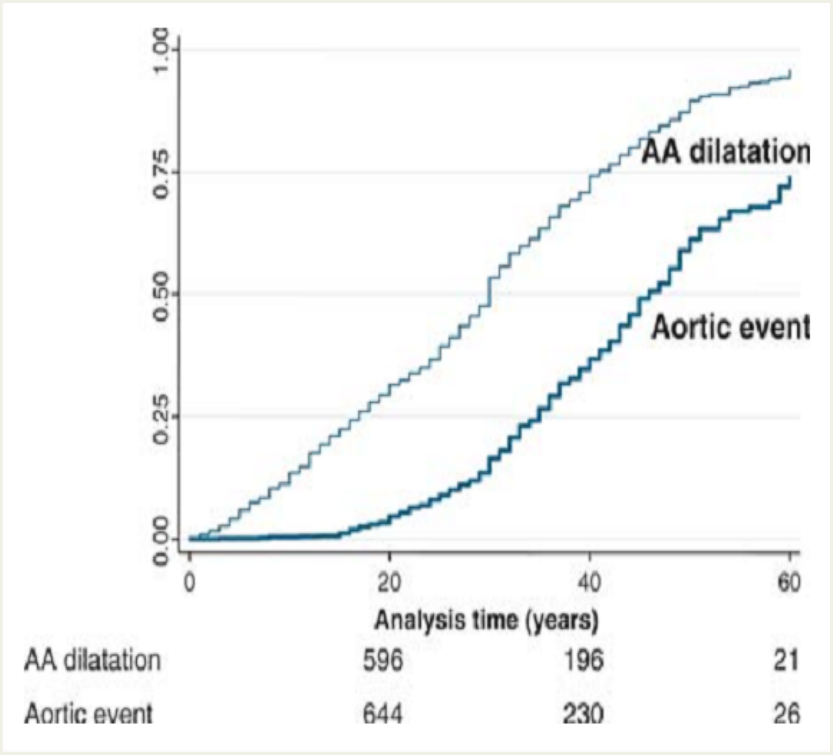
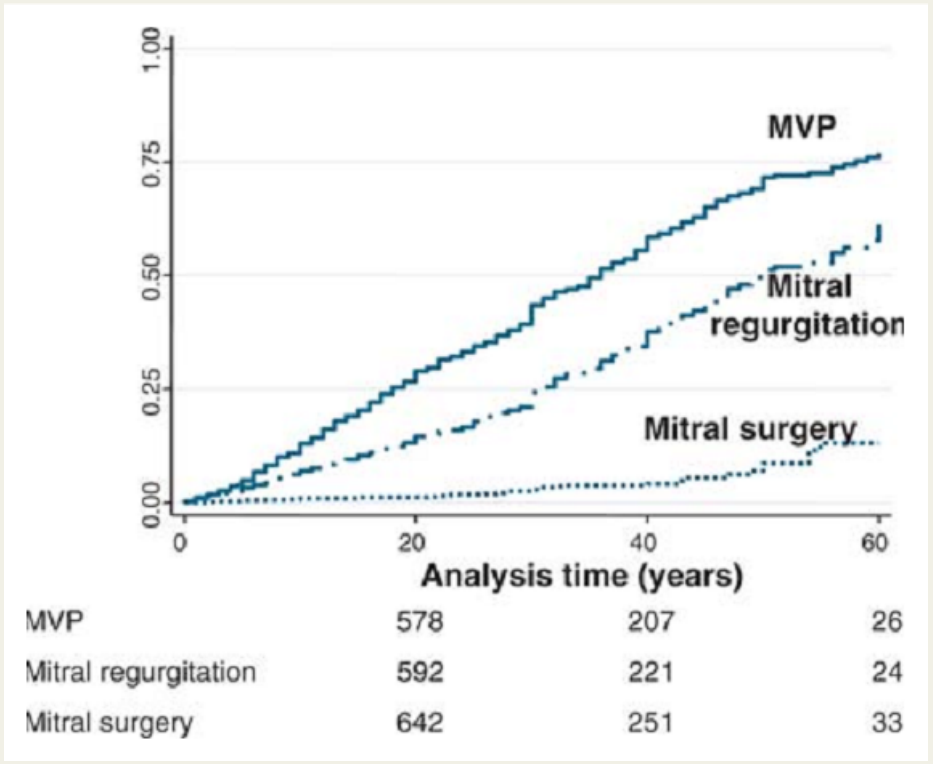


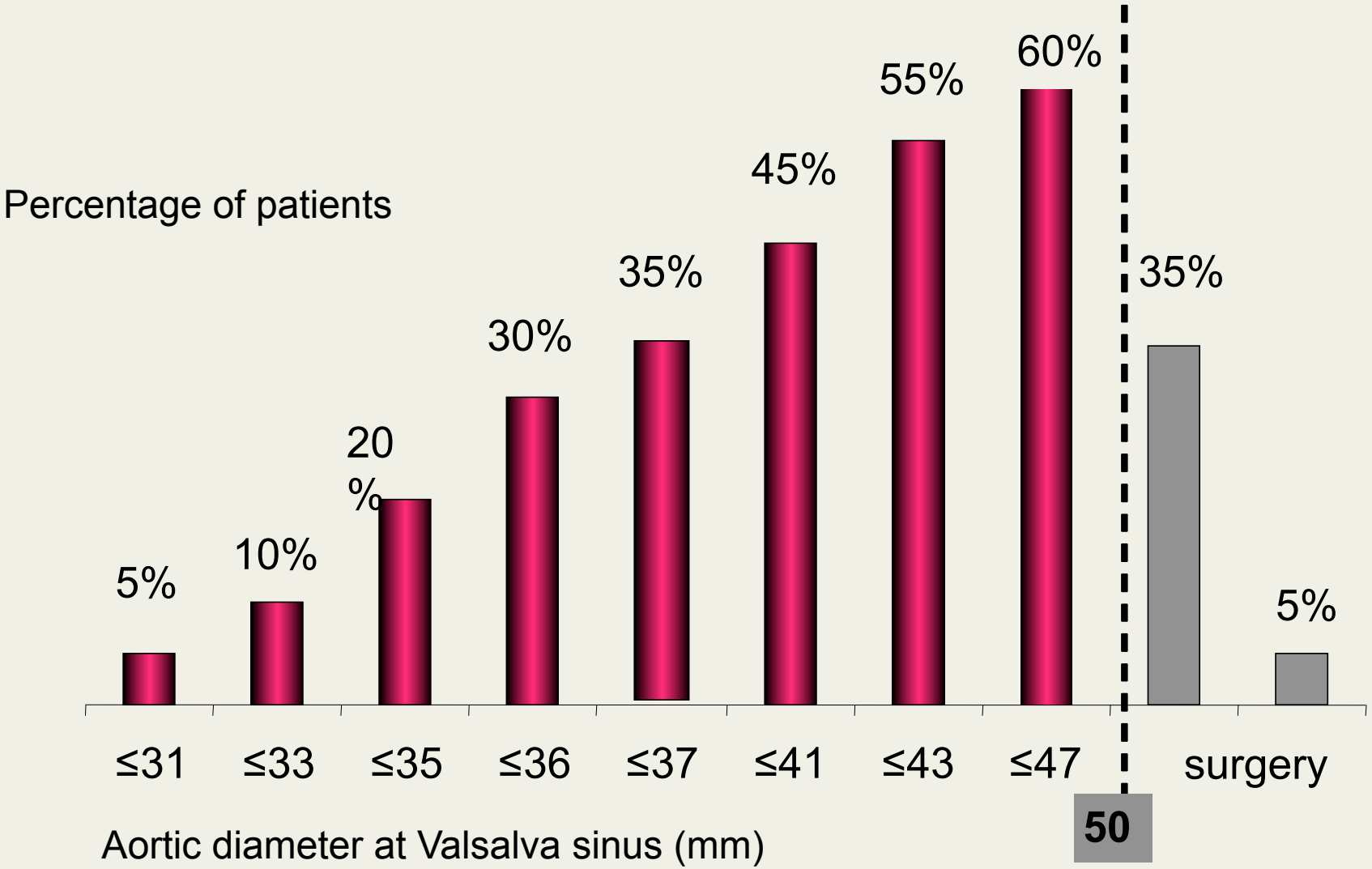
Figure 3 Cumulative probability of ascending aortic dilatation in adult probands* diagnosed on clinical grounds (n = 541, solid line) and in adult probands* with unfulfilled "clinical" international criteria (n = 146, broken line) (log rank test, $p<0.0001$). *≥18 years.





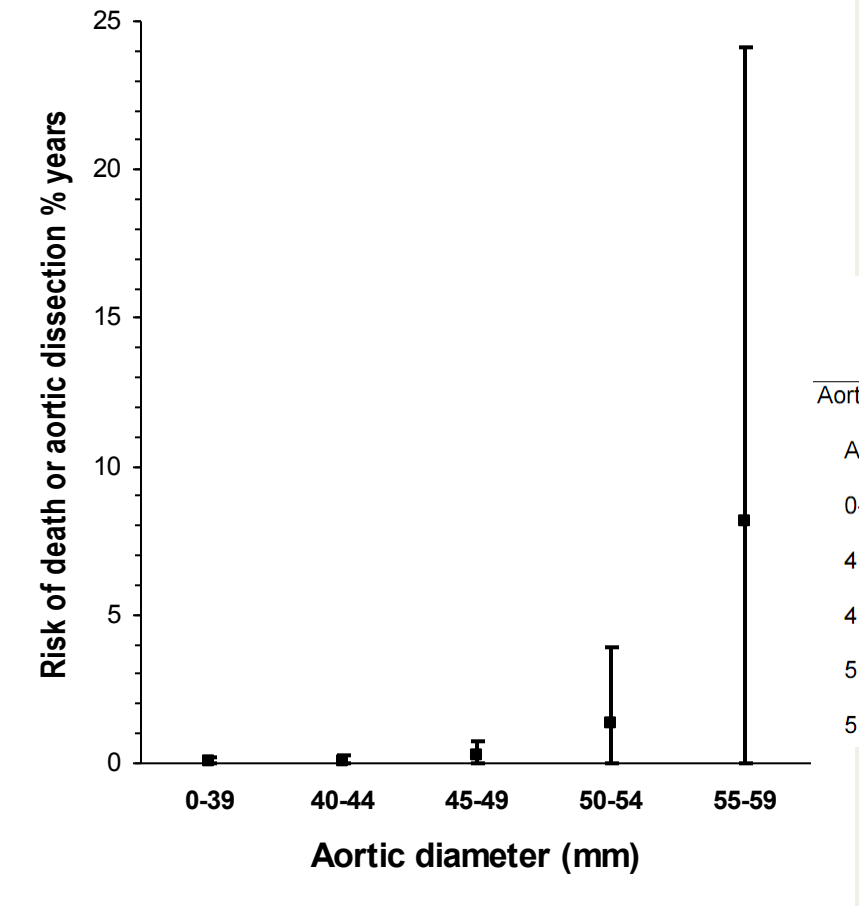
D Detaint... G Jondeau Eur Heart J 2010

Dissection descendante



L Mimoun…. G Jondeau; Eur Heart J 2011;32:443

Aortic risk as a function of maximal aortic diameter in Marfan Syndrome



	Patients (n)	Event number	Patient-years of follow-up	Annual risk (%) [CI 95%]
Aortic event without surgery				
Aortic diameter (mm)				
0–39	423	2	2353	0·09 [0·00–0·20]
40–44	219	1	995	0·10 [0·00–0·30]
45–49	157	2	675	0·30 [0·00–0·71]
50–54	54	1	75	1·33 [0·00–3·93]
55–59	14	1	12	8·14 [0·00–24·10]

G Jondeau et al. Circulation 2012

Seuil pour chirurgie aortique

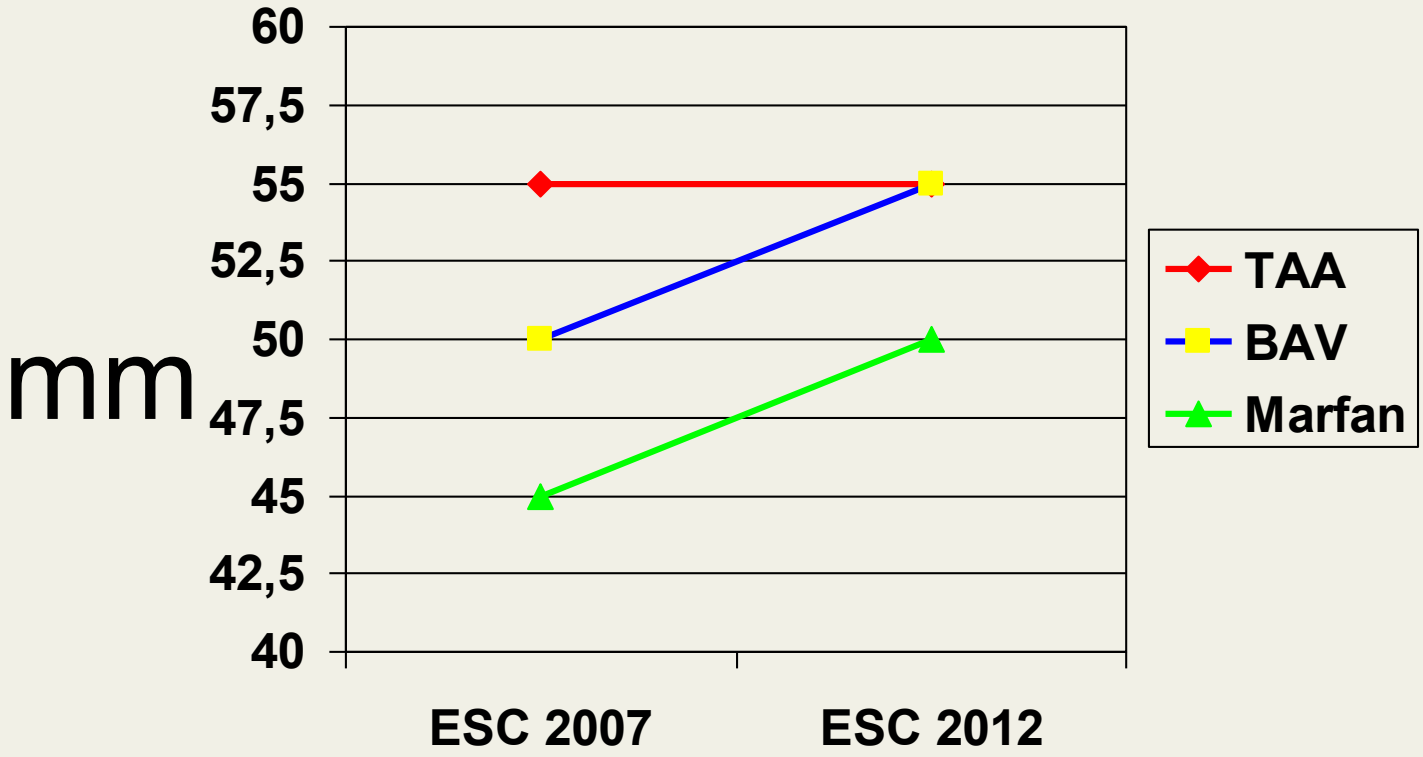


Table 8 **Indications for surgery in (A) severe aortic regurgitation and (B) aortic root disease (whatever the severity of aortic regurgitation)**

	Class [a]	Level [b]	Ref [c]
A. Indications for surgery in severe aortic regurgitation			
Surgery is indicated in symptomatic patients.	I	B	59
Surgery is indicated in asymptomatic patients with resting LVEF ≤50%.	I	B	71
Surgery is indicated in patients undergoing CABG or surgery of ascending aorta, or on another valve.	I	C	
Surgery should be considered in asymptomatic patients with resting EF >50% with severe LV dilatation: LVEDD >70 mm, or LVESD >50 mm or LVESD >25 mm/m^2 BSA.[d]	IIa	C	
B. Indications for surgery in aortic root disease (whatever the severity of AR)			
Surgery is indicated in patients who have aortic root disease with maximal ascending aortic diameter[e] ≥50 mm for patients with Marfan syndrome.	I	C	
Surgery should be considered in patients who have aortic root disease with maximal ascending aortic diameter: ≥45 mm for patients with Marfan syndrome with risk factors[f] ≥50 mm for patients with bicuspid valve with risk factors[g] ≥55 mm for other patients	IIa	C	

AR = aortic regurgitation; BSA = body surface area; CABG = coronary artery bypass grafting; EF = ejection fraction; LV = left ventricular; LVEDD = left ventricular end-diastolic diameter; LVESD = left ventricular end-systolic diameter.

[a]Class of recommendation.

[b]Level of evidence.

[c]Reference(s) supporting class I (A + B) and IIa + IIb (A + B) recommendations.

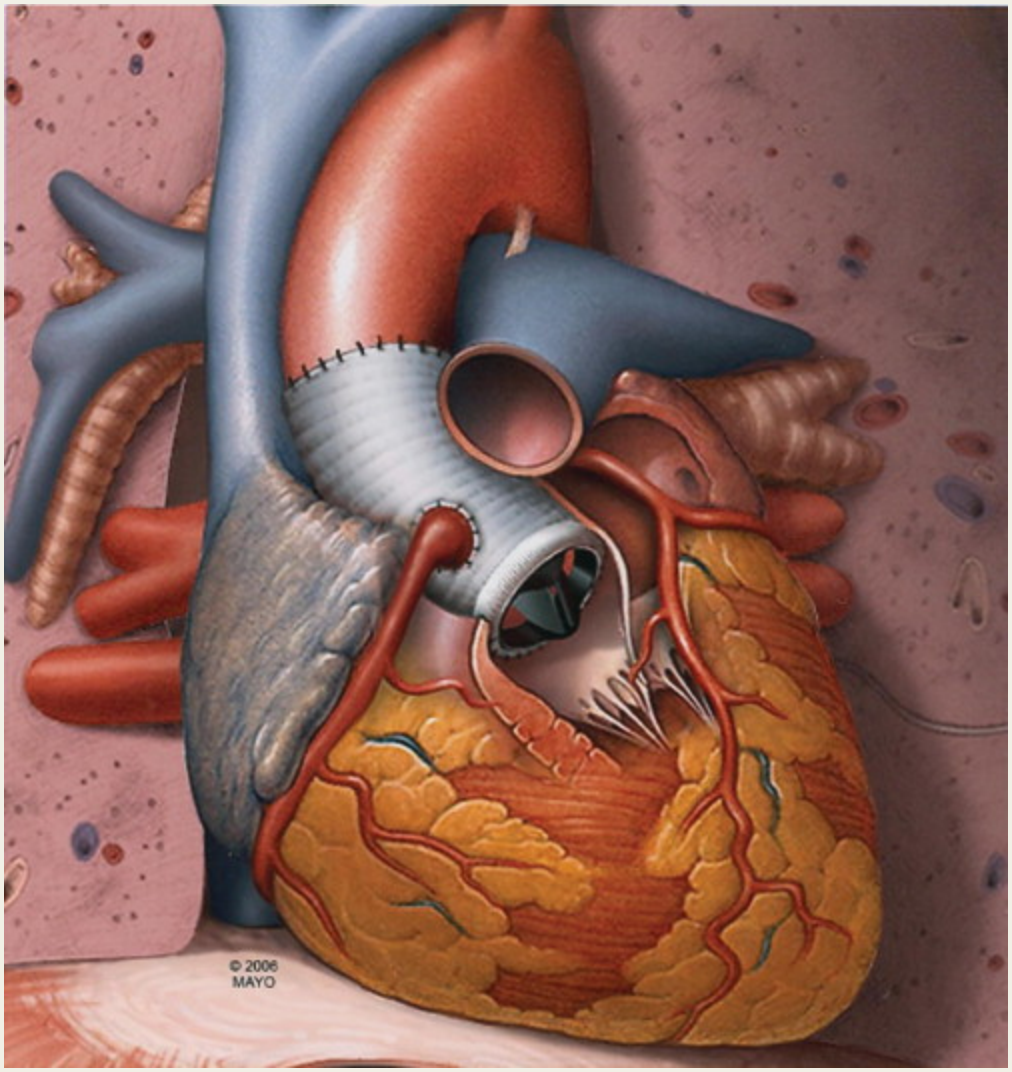
[d]Changes in sequential measurements should be taken into account.

[e]Decision should also take into account the shape of the different parts of the aorta. Lower thresholds can be used for combining surgery on the ascending aorta for patients who have an indication for surgery on the aortic valve.

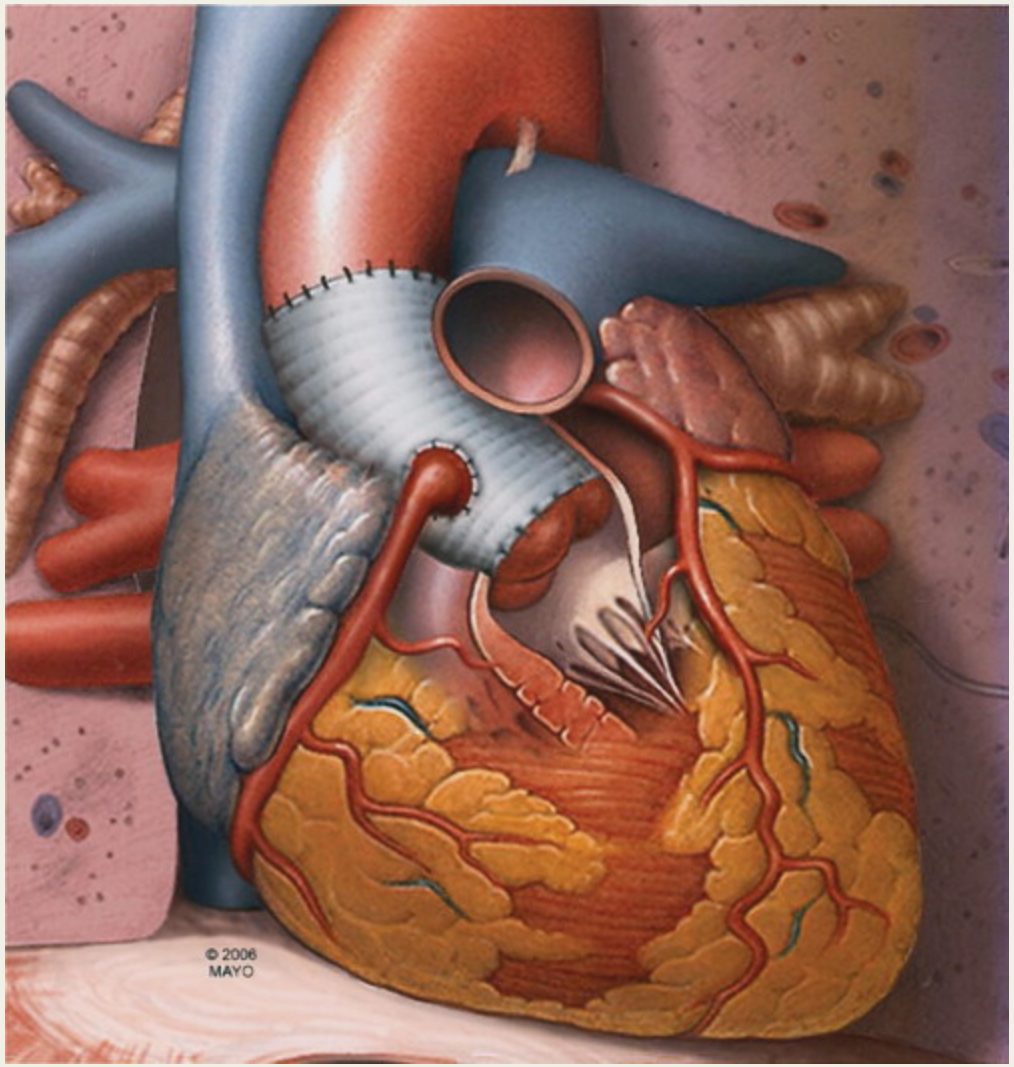
[f]Family history of aortic dissection and/or aortic size increase >2 mm/year (on repeated measurements using the same imaging technique, measured at the same aorta level with side-by-side comparison and confirmed by another technique), severe AR or mitral regurgitation, desire of pregnancy.

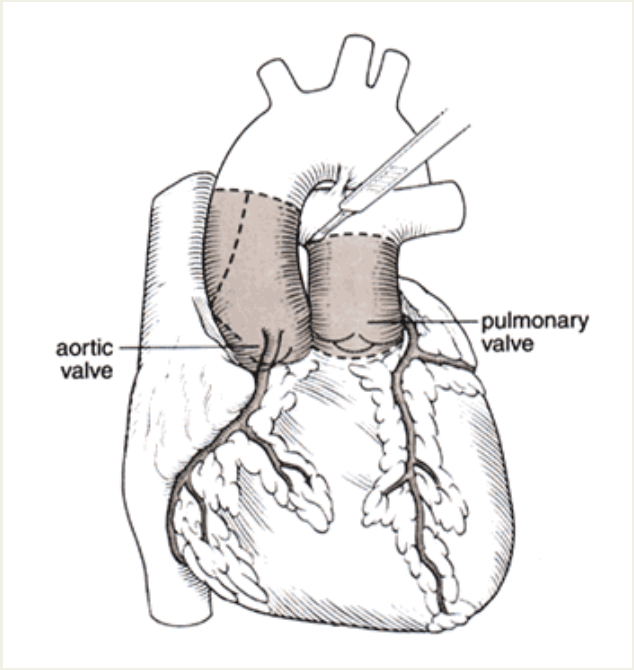
[g]Coarctation of the aorta, systemic hypertension, family history of dissection or increase in aortic diameter >2 mm/year (on repeated measurements using the same imaging technique, measured at the same aorta level with side-by-side comparison and confirmed by another technique).

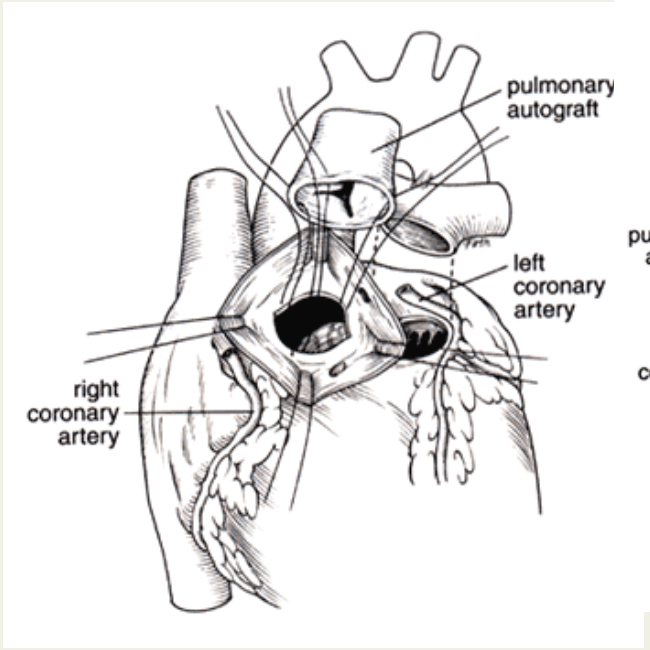
Bentall

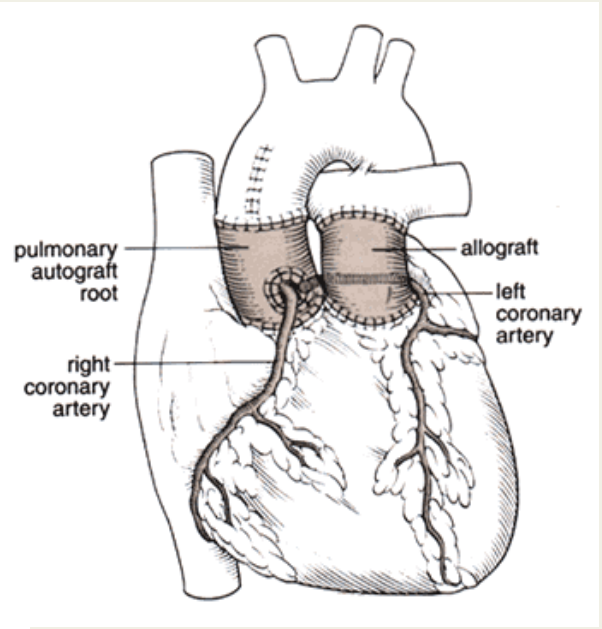


Valve sparing

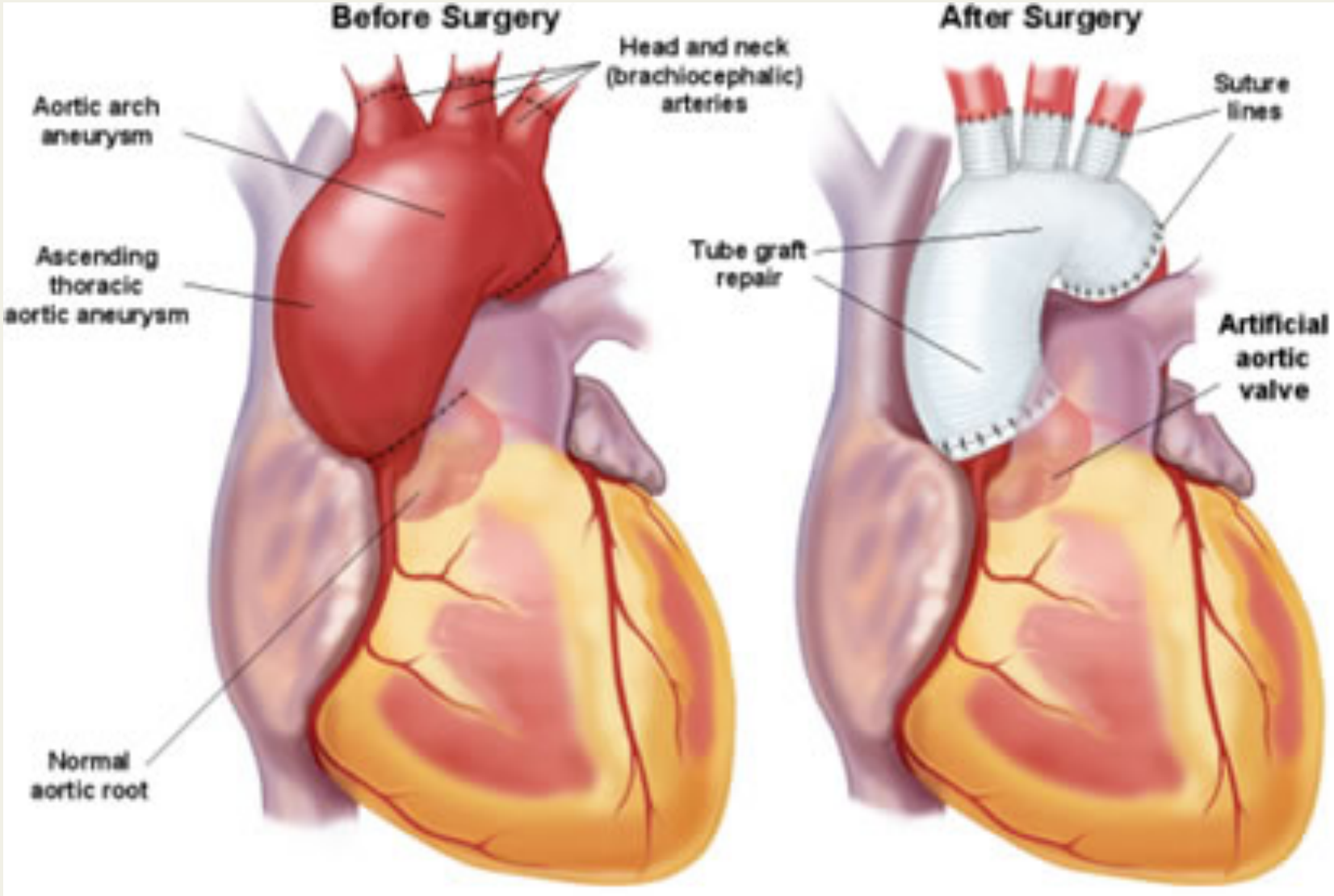



aortic valve
pulmonary valve


pulmonary autograft
left coronary artery
right coronary artery

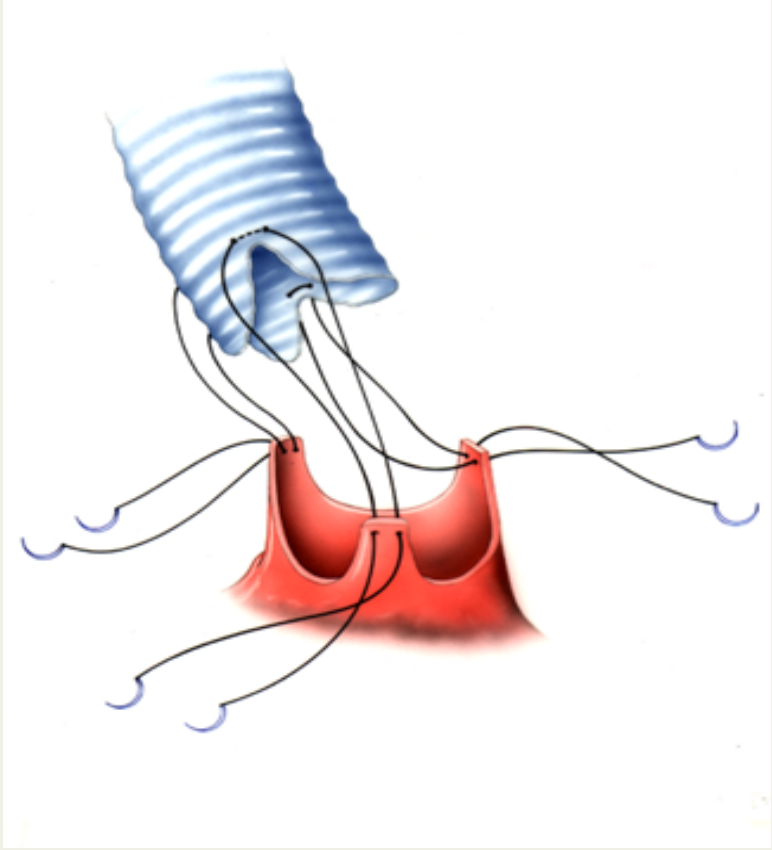

pulmonary autograft root
allograft
left coronary artery
right coronary artery

Sus-coronary, arch



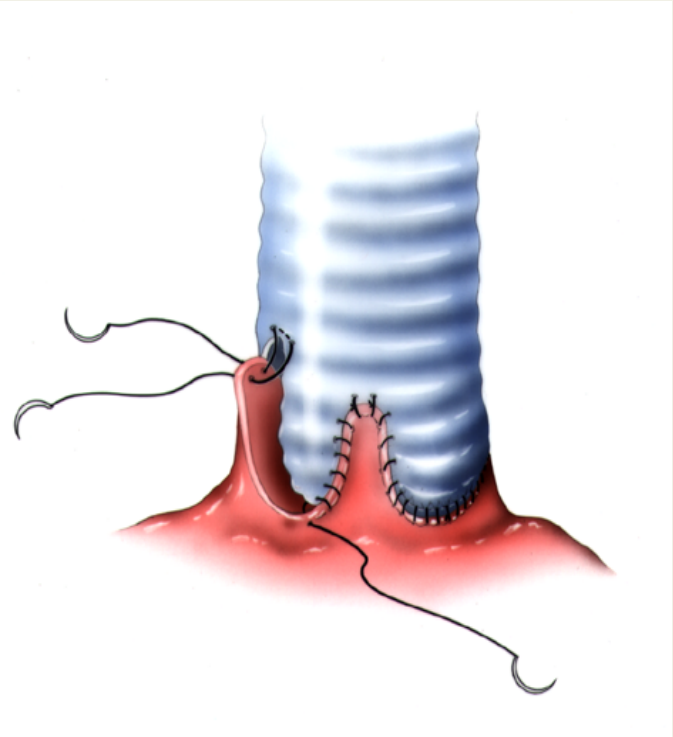
LA TECHNIQUE DE REMODELAGE DE M. YACOUB ET T. DAVID

- La suture du tube et de l'orifice aortique commence par le sommet des com-missures .



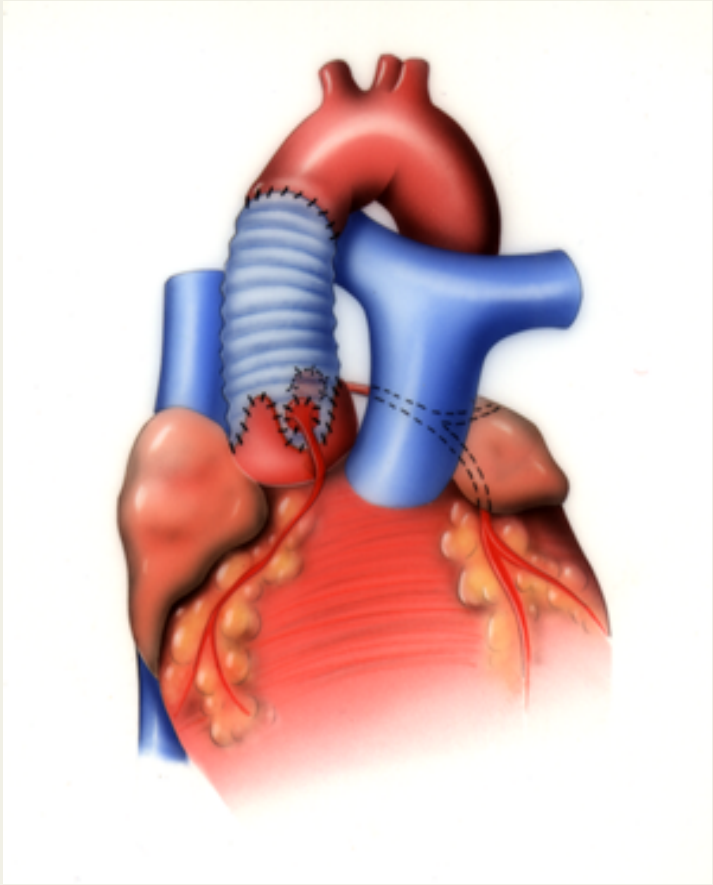
LA TECHNIQUE DE REMODELAGE DE M. YACOUB ET T. DAVID

- La fin du surjet s ’effectue au fond des trois cuspides avec des points de plus en plus serrés: suture « bouffante »



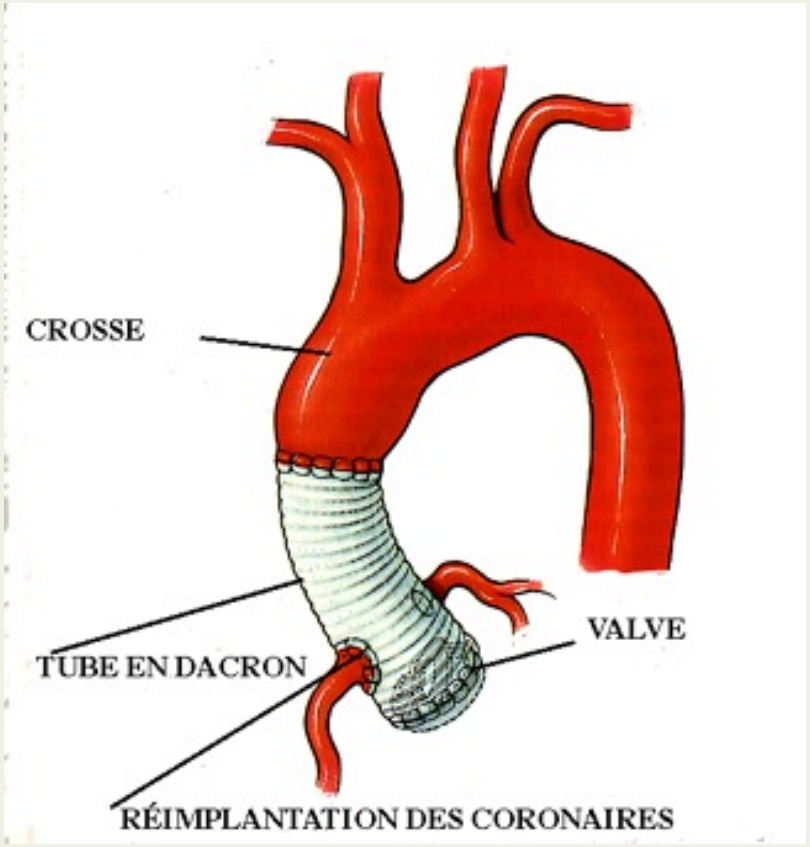
LA TECHNIQUE DE REMODELAGE DE M. YACOUB ET T. DAVID

■L ’intervention est terminée par la réimplantation des collerettes ostiales, comme dans l ’opération de Bentall .



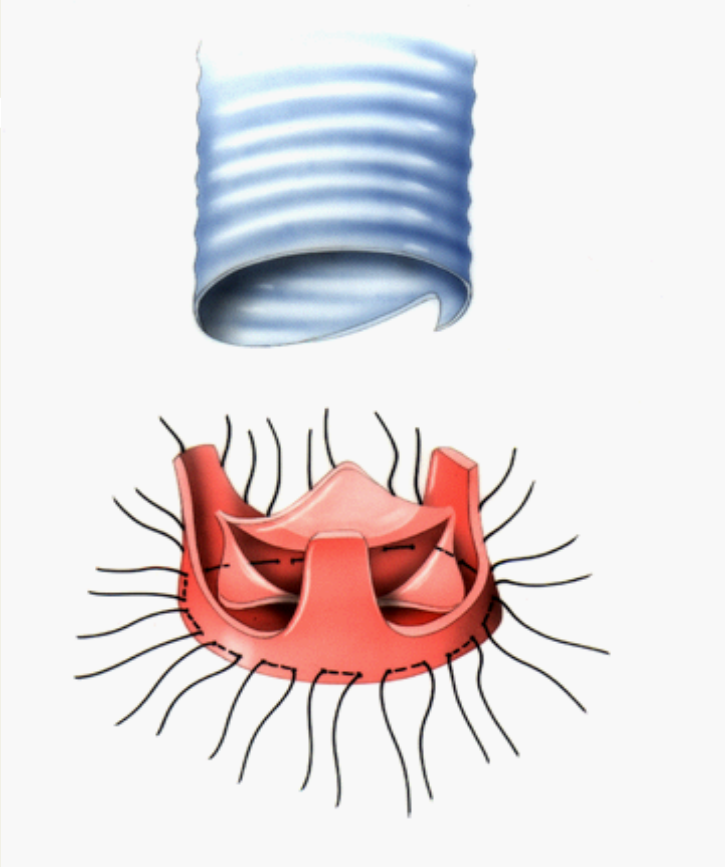
ANNULO-ECTASIE
L ’OPÉRATION DE BENTALL

- **Tube valvé, implanté sur l ’anneau.**
- **Réimplantation directe des ostia coronaires avec une collerette aortique.**
- **Mortalité: 3% pour les interventions programmées (199 P.)**
- **Résultats durables**



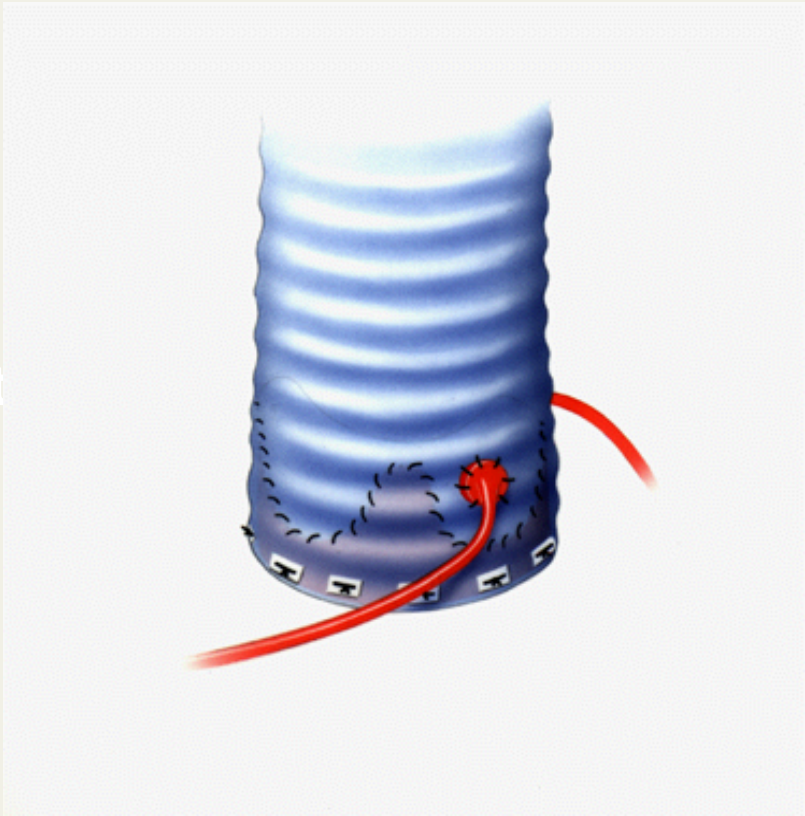
TECHNIQUE D ’INCLUSION DE TIRONE DAVID (1992)

- Exérèse complète des sinu de valsalva
- Points de fixation en U passés dans l ’anneau sous les sigmoïdes
- Prothèse échancrée



LA TECHNIQUE D ’INCLUSION DE TIRONE DAVID

- La prothèse est descendue autour de l ’appareil sigmoïdien.
- La découpe aortique est fixée à l ’intérieur du tube par un surjet.
- Les ostia coronaires sont réimplantés.



Les questions

1) Quelle étiologie ?
    1) Type dilatation
    2) Signes extra-aortiques
    3) Enquête familiale
2) Quel traitement ? (quel risque)
    1) Mode de vie
    2) Médical
    3) Chirurgical
3) Quel suivi ?
    1) Quelle fréquence
    2) Pour qui ?