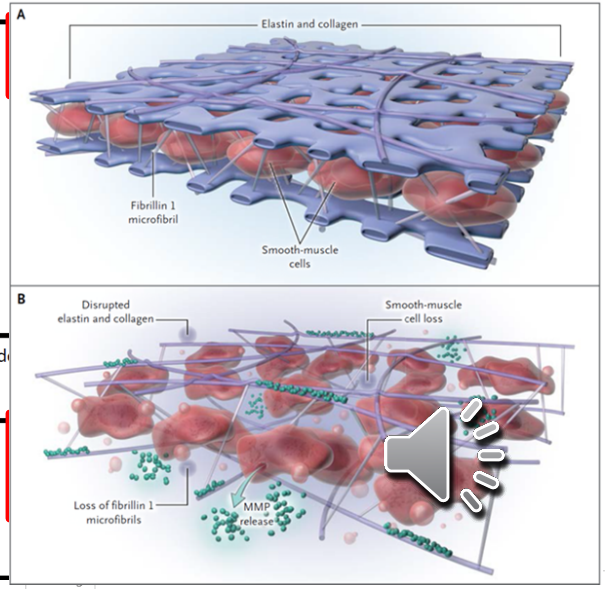
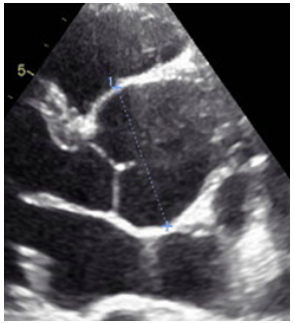
Syndrome de Marfan : atteinte aortique

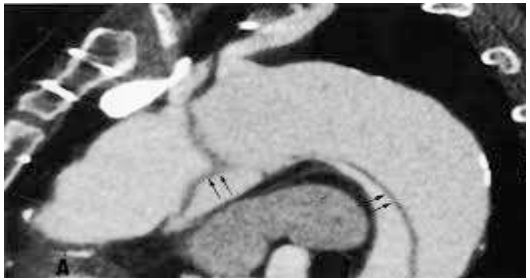
Absence d'histoire familiale de syndrome de Marfan

4 scénarios possibles pour faire le diagnostic de syndrome d

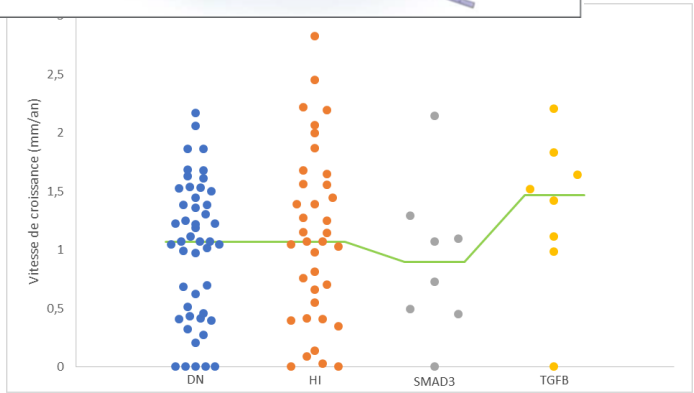
Histoire familiale de syndrome de Marfan (apparentés au 1er degré)
associée à un des critères suivants







Ghent 2010



Vitesse de croissance du sinus de Valsalva en fonction du génotype

R Vincent 2020

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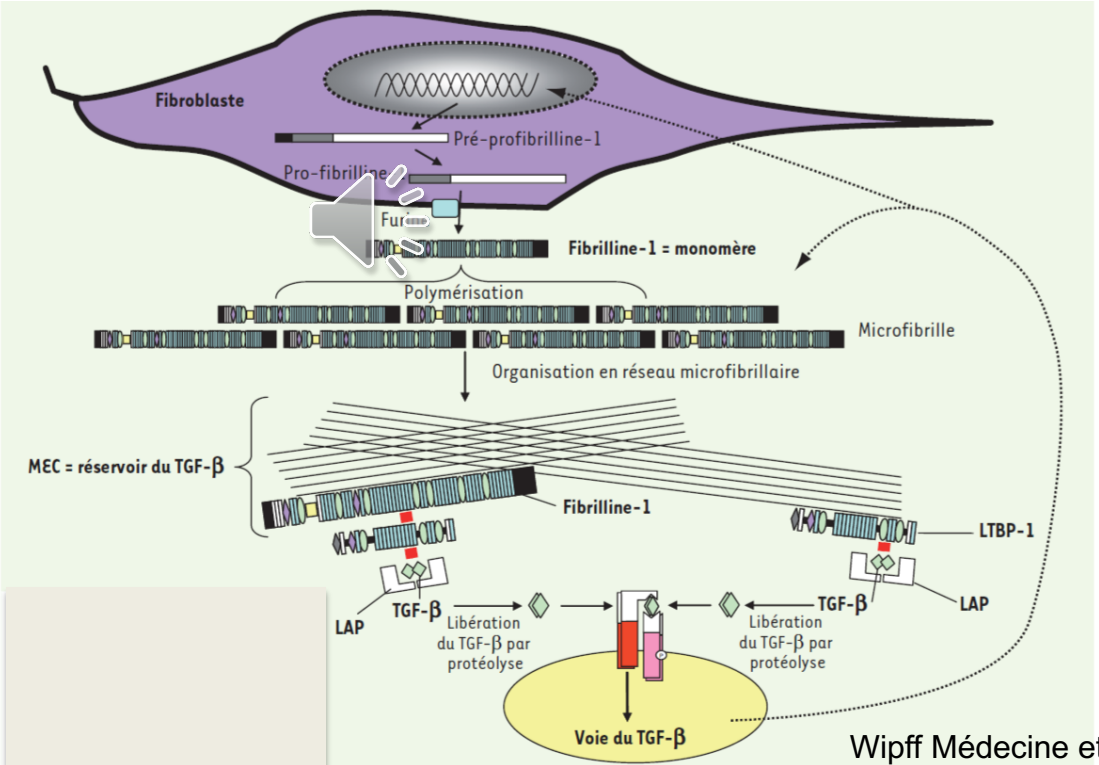
Physiopathologie des anévrysmes aortiques

Anomalie de la fibrilline 1 : constituant majeur des microfibrilles

- Intégrité homéostasie des tissus élastiques
- Interaction avec la voie du TGFβ

Gène FBN1 en 15q21.1



Wipff Médecine et science 2009

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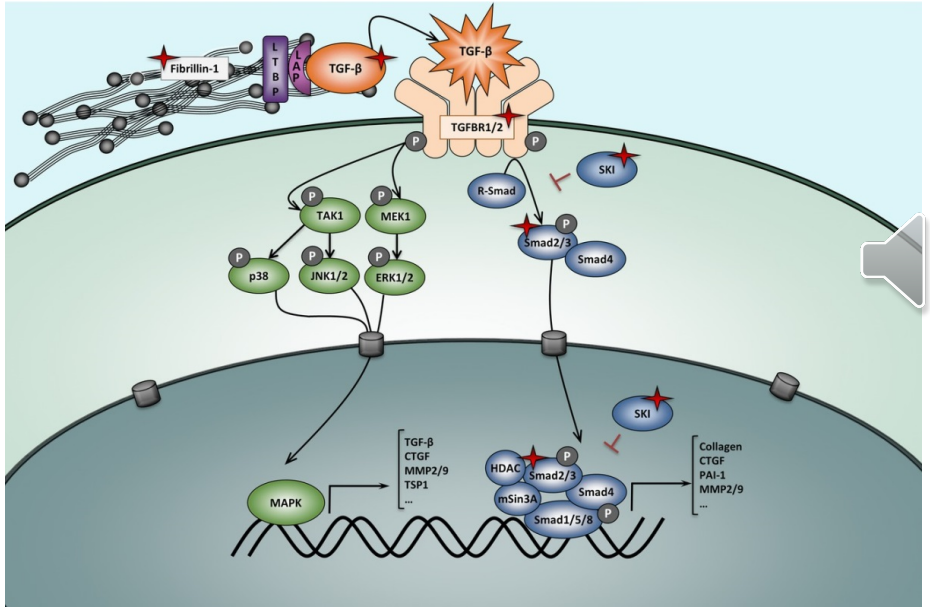
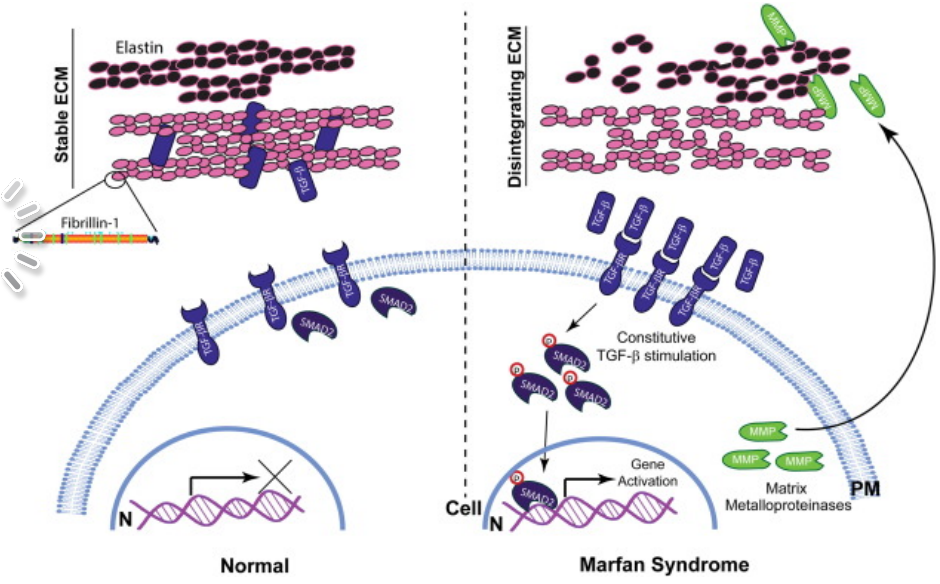
Physiopathologie des anévrysmes aortiques
interaction fibrilline 1/TGFβ





Human Mutation, 2016,

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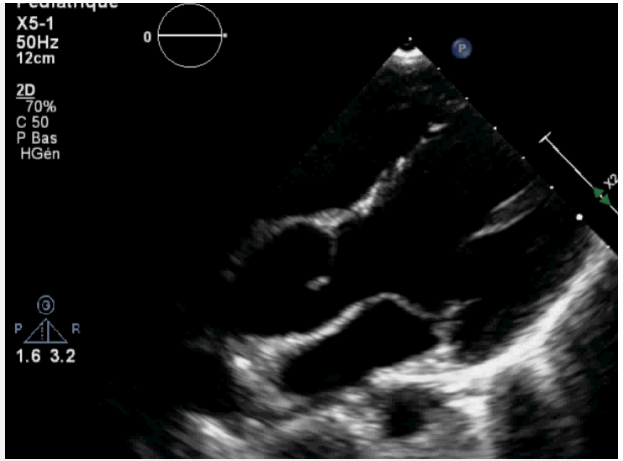
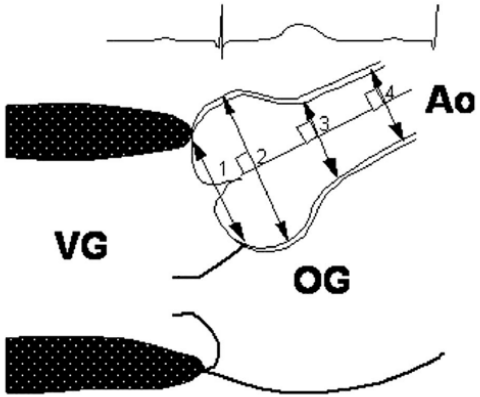
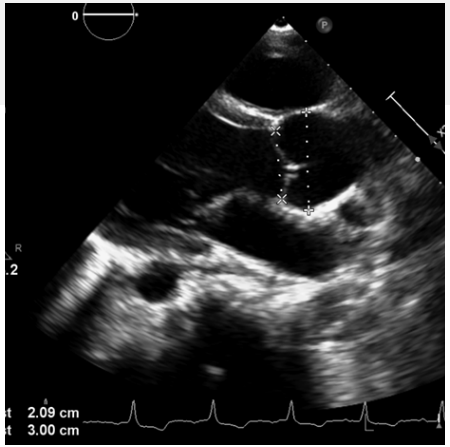
Diagnostic chez l'enfant

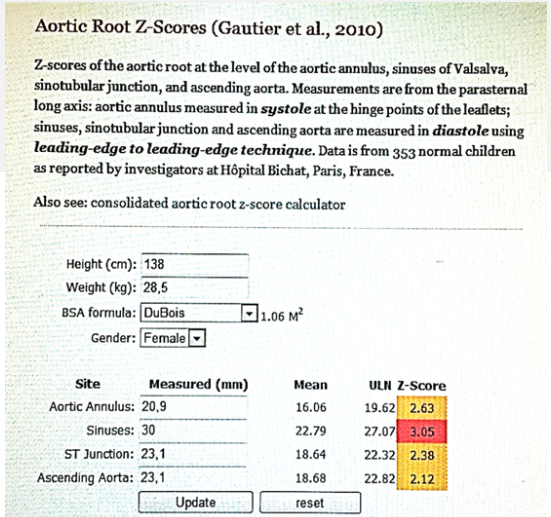
Elsa, 6 ans

- ✓ Souffle systolique
- ✓ Prolapsus valvulaire mitral familial
- ✓ Z-score taille +3
- ✓ Signe du poignet









Aortic Root Z-Scores (Gautier et al., 2010)

Z-scores of the aortic root at the level of the aortic annulus, sinuses of Valsalva, sinotubular junction, and ascending aorta. Measurements are from the parasternal long axis: aortic annulus measured in ***systole*** at the hinge points of the leaflets; sinuses, sinotubular junction and ascending aorta are measured in ***diastole*** using ***leading-edge to leading-edge technique***. Data is from 353 normal children as reported by investigators at Hôpital Bichat, Paris, France.

Also see: consolidated aortic root z-score calculator

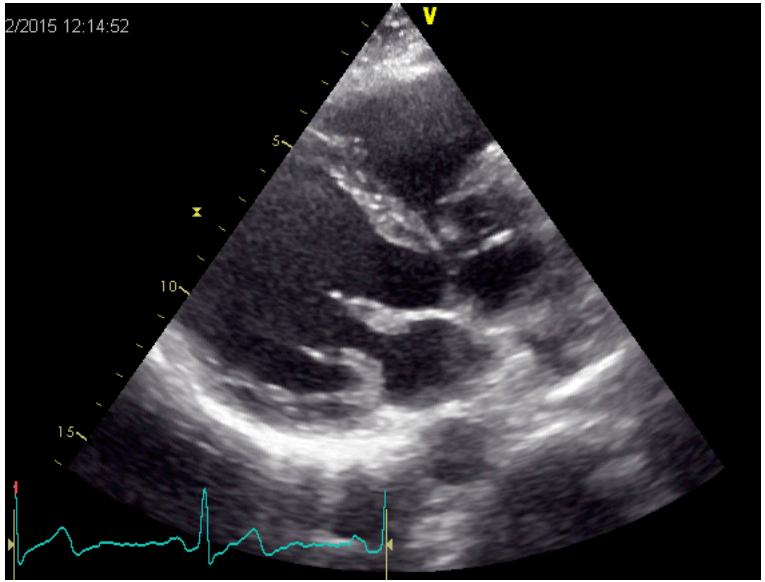
Height (cm): 138
Weight (kg): 28,5
BSA formula: DuBois 1.06 M^2
Gender: Female

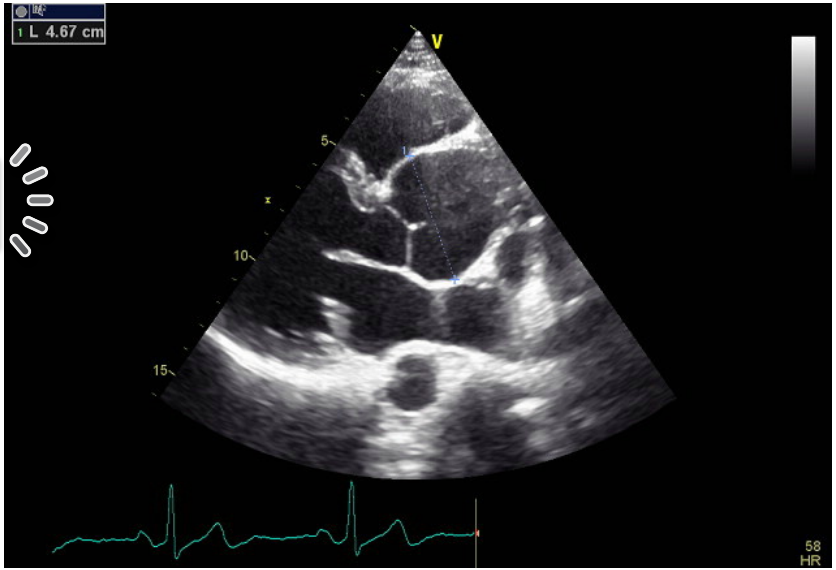
Site	Measured (mm)	Mean	ULN	Z-Score
Aortic Annulus:	20,9	16.06	19.62	2.63
Sinuses:	30	22.79	27.07	3.05
ST Junction:	23,1	18.64	22.32	2.38
Ascending Aorta:	23,1	18.68	22.82	2.12

Update reset

Enquête familiale

Mère d'Elsa: 31 ans , « prolapsus mitral familial»,
Taille: 1,80 m, scoliose 20°, 24 SA de grossesse

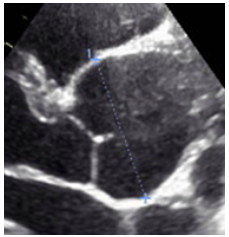


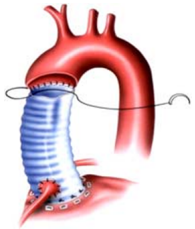


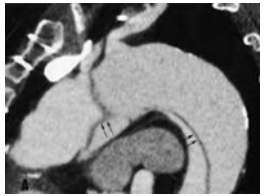
- Mutation FBN1+

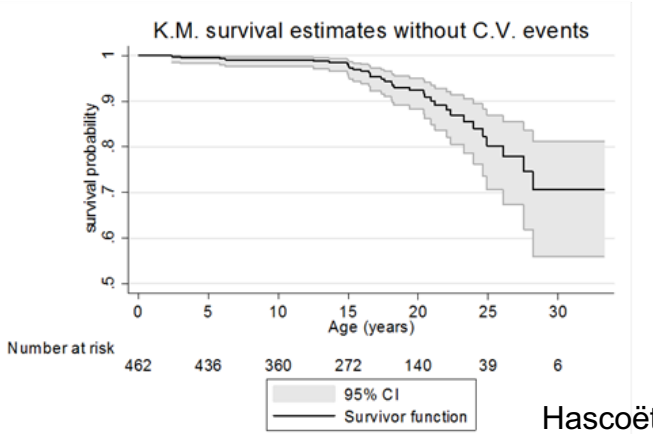
Manifestations cardiaques des enfants avec SM

- Dilatation aortique < 18 ans: 60-77%. z-score > +3:1/3
- Evolution du diamètre aortique: 0,5-1,4 mm/an (N: 0,1 mm/an)
- Prolapsus mitral: 30 % (IM significative 20 %). Chirurgies rares
- Prolapsus tricuspidien: 15 %
- Evénements aortiques: 8% <18 ans (2,4 % < 15 ans)
  - Chirurgies aortiques prophylactiques: 80 %
  - Dissections aortiques rares < 15 ans (1%)

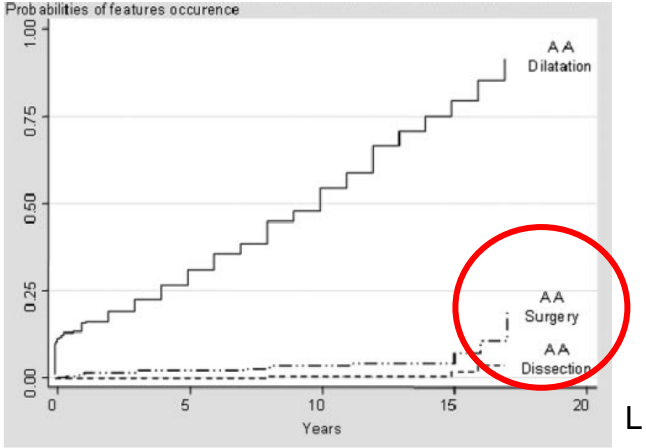








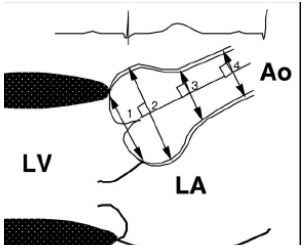
Hascoët ACVD 2019

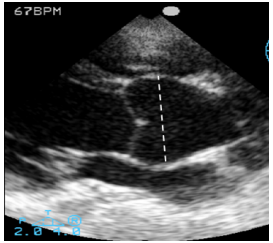


L Faivre. Pediatrics 2009

Quelle prévention cardiovasculaire chez l'enfant ?

- ✓ **Dépister : famille**

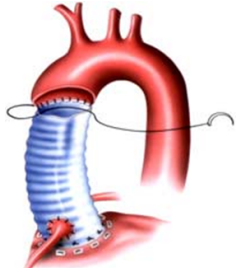




- ✓ **Surveiller:** écho annuelle, IRM/TDM

- ✓ **Prévenir** : sport, bétabloquants

- ✓ **Chirurgie prophylactique**



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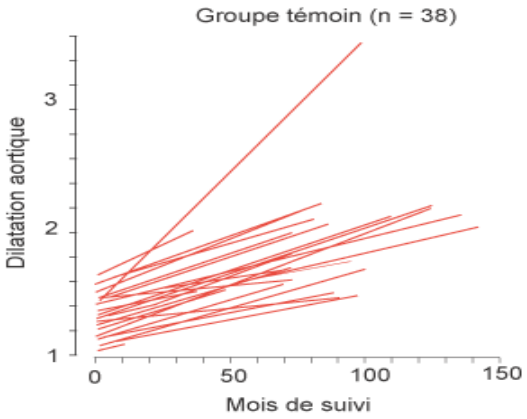
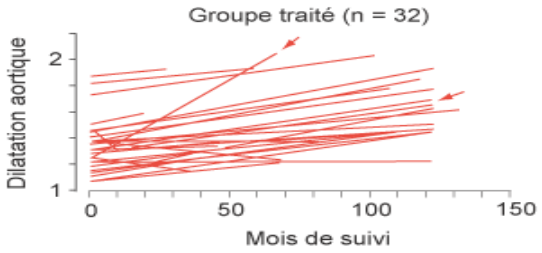
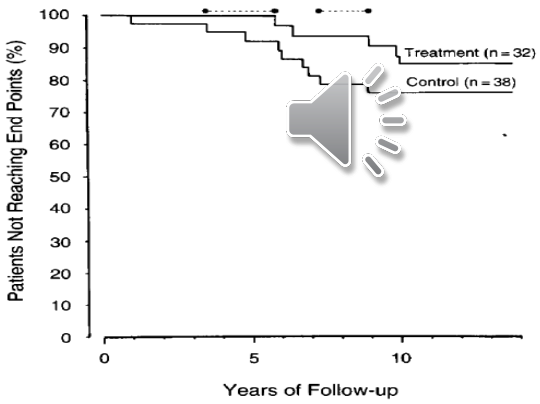
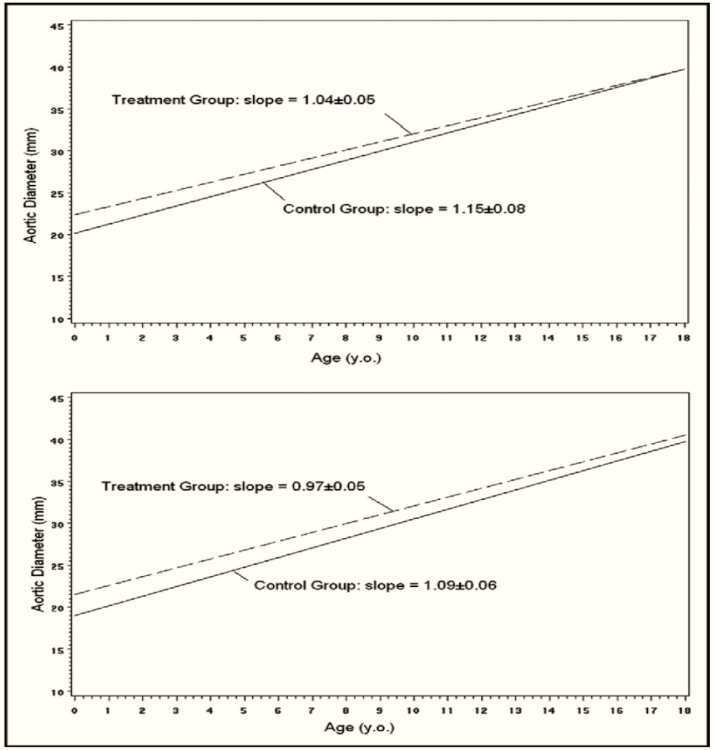
Traitement bétabloquant chez l'enfant avec SM



Shores . N Engl J Med 1994







Bétabloquants lors du diagnostic (après 4-5 ans):

- Bisoprolol en débutant à 1,25 mg/ jour
- Contre indications (asthme sévère): verapamil , sartans?

155 enfants < 12 ans (6,1 ans)
M Ladouceur . Am J Cardiol. 2007

PNDS Marfan 2018