

Pediatric Issues in Marfan Syndrome

The Challenge:

Accommodating the issue of growth and the dynamic nature of the condition into diagnosis and management

General Diagnostic Issues

Cardiovascular Disease:

Aortic Disease:

Diagnosis

Worrisome Situations

Prevention

Mitral Valve Disease

Preventing Infection

Surgical Management

Growth

Performance

Communication

Diagnosis of Young Children

Diagnosis requires “major” involvement in 2 categories with “minor” involvement in a third.

Major:

- 4 of 8 selected skeletal features
- Aortic enlargement / dissection
- Lens dislocation
- Dural ectasia
- Family / genetic history

Minor:

- Specified skeletal features
- Specified eye findings
- Specified heart findings
- Lung involvement
- Skin involvement

Diagnostic Possibilities

Marfan Syndrome:

Meets diagnostic criteria on 1st visit

“Emerging” Marfan Syndrome:

Fails to meet criteria initially

Definitive at later visit

Other Disorders:

MASS phenotype:

Myopia, Mitral valve prolapse, borderline Aortic enlargement, Skin and Skeletal findings

Mitral Valve Prolapse Syndrome:

Mitral valve prolapse plus skeletal features

“Isolated Ectopia Lentis:

Lens dislocation plus skeletal features

Familial Aortic Aneurysm:

Aortic aneurysm with or without skeletal features

Diagnostic Possibilities

Scenario 1:

Diagnostic criteria not met at first visit but aortic enlargement is present

Plan: Start beta-blockers, exercise restriction
Close followup including echocardiogram

Scenario 2:

Suggestive features seen but criteria not met and no aortic enlargement

Plan: No medications or restrictions
Maintain follow-up at least until end of growth
(every year X 2, every other year X2...)

The “label” isn’t everything...deal with what you see

“Neonatal Marfan Syndrome” (NMS)

Some children are born with severe and rapidly progressive disease. Cardiovascular manifestations can be difficult to treat, causing early mortality in some cases.

Misconception:

NMS is a discrete clinical entity, allowing meaningful prognostication at the time of diagnosis.

Reality:

Marfan syndrome is a clinical continuum, even at the severe end of the spectrum.

Many patients with neonatal onset of severe and progressive disease can be adequately managed.

Diagnosis of MFS at birth does **NOT** necessarily predict severe disease.

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Aortic dilatation and dissection in childhood:

Current recommendation is for aortic root replacement at 5.0 cm in adults. This figure was determined by observing the size of the aorta at the time of dissection.

The reality is that aortic dissection is exceedingly rare during early childhood (preadolescent age group) irrespective of aortic root size. Aortic dissection manifests both aortic size and the duration of hemodynamic stress.

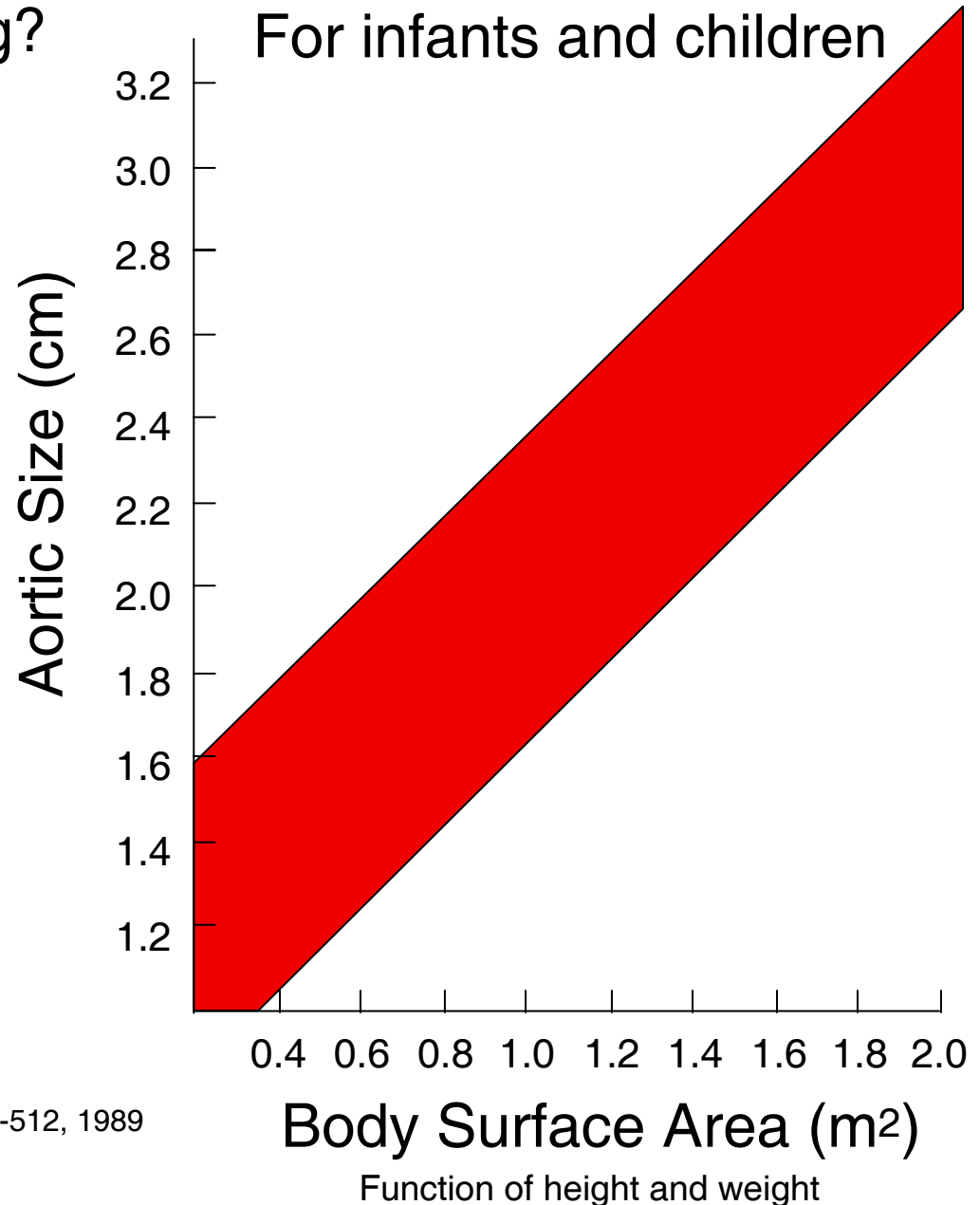
The good news: aortic dissection is exceedingly rare

The bad news: there have been too few events to develop meaningful guidelines for young children.

When is the Aorta Too Big?

The aorta normally enlarges with increase in age and body size.

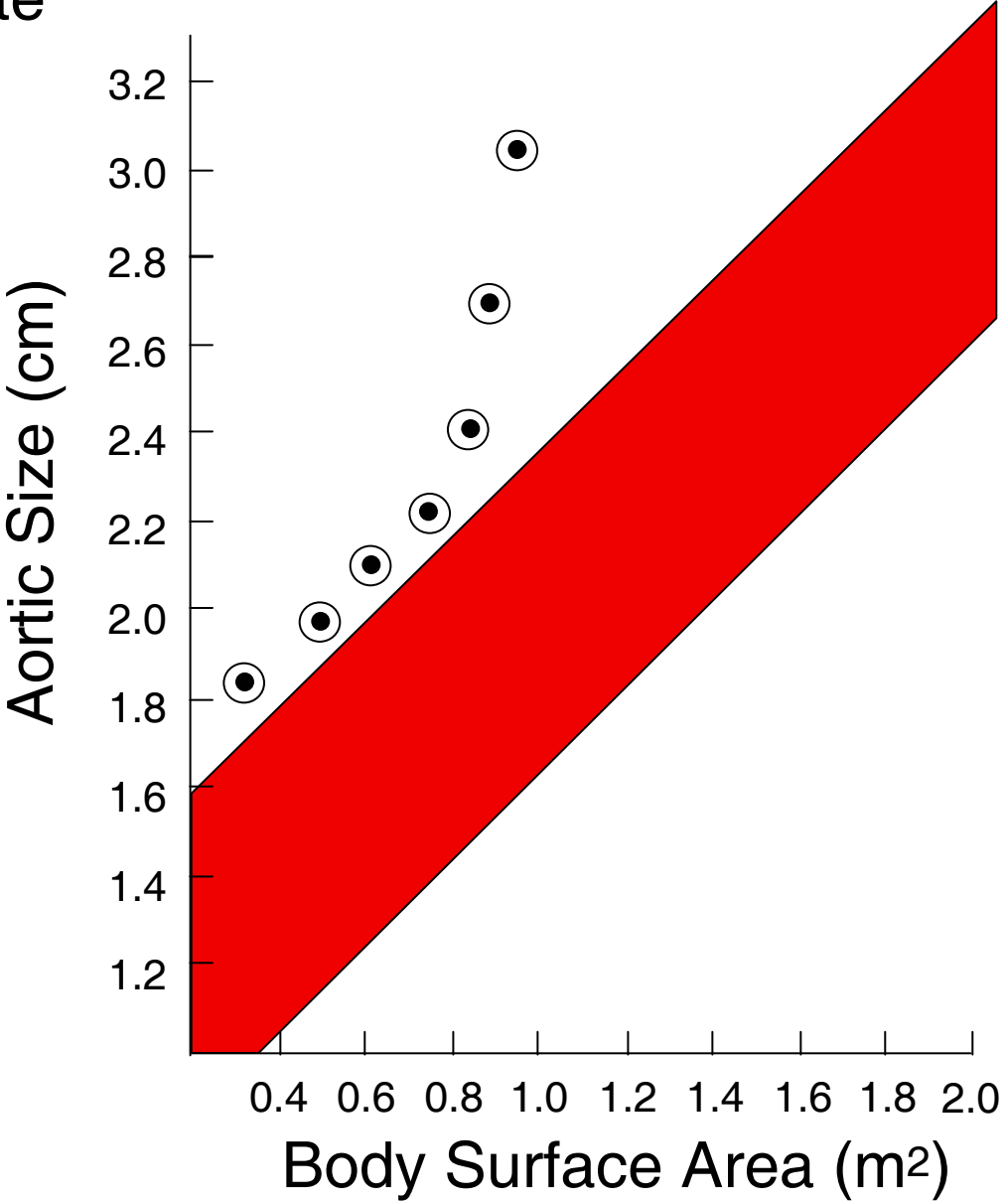
Values in children must be compared to “normals” for a given age and body size.



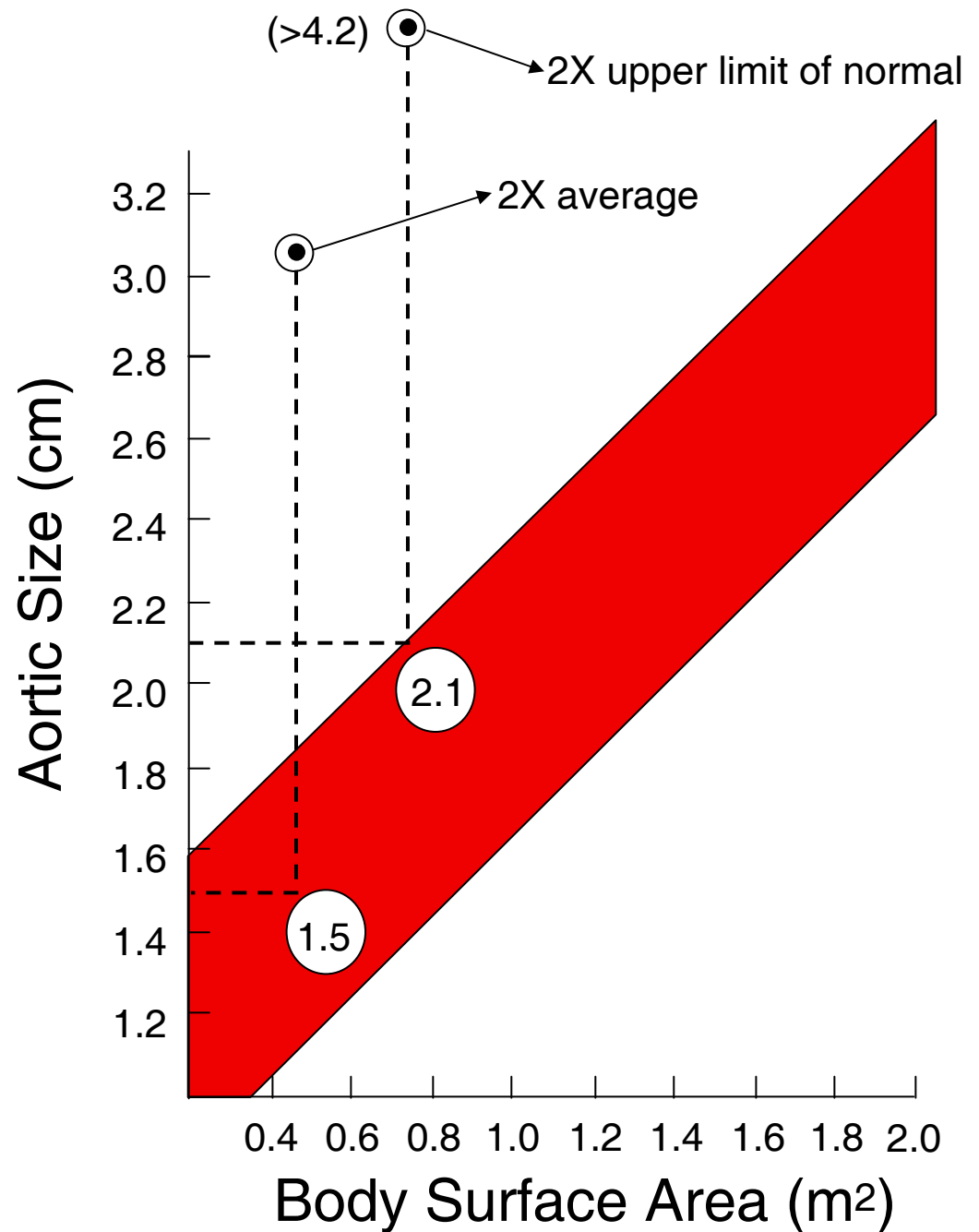
Adapted from Roman et al. Am J Cardiol 64:507-512, 1989

Rapid Increase in Rate
Of Aortic Growth

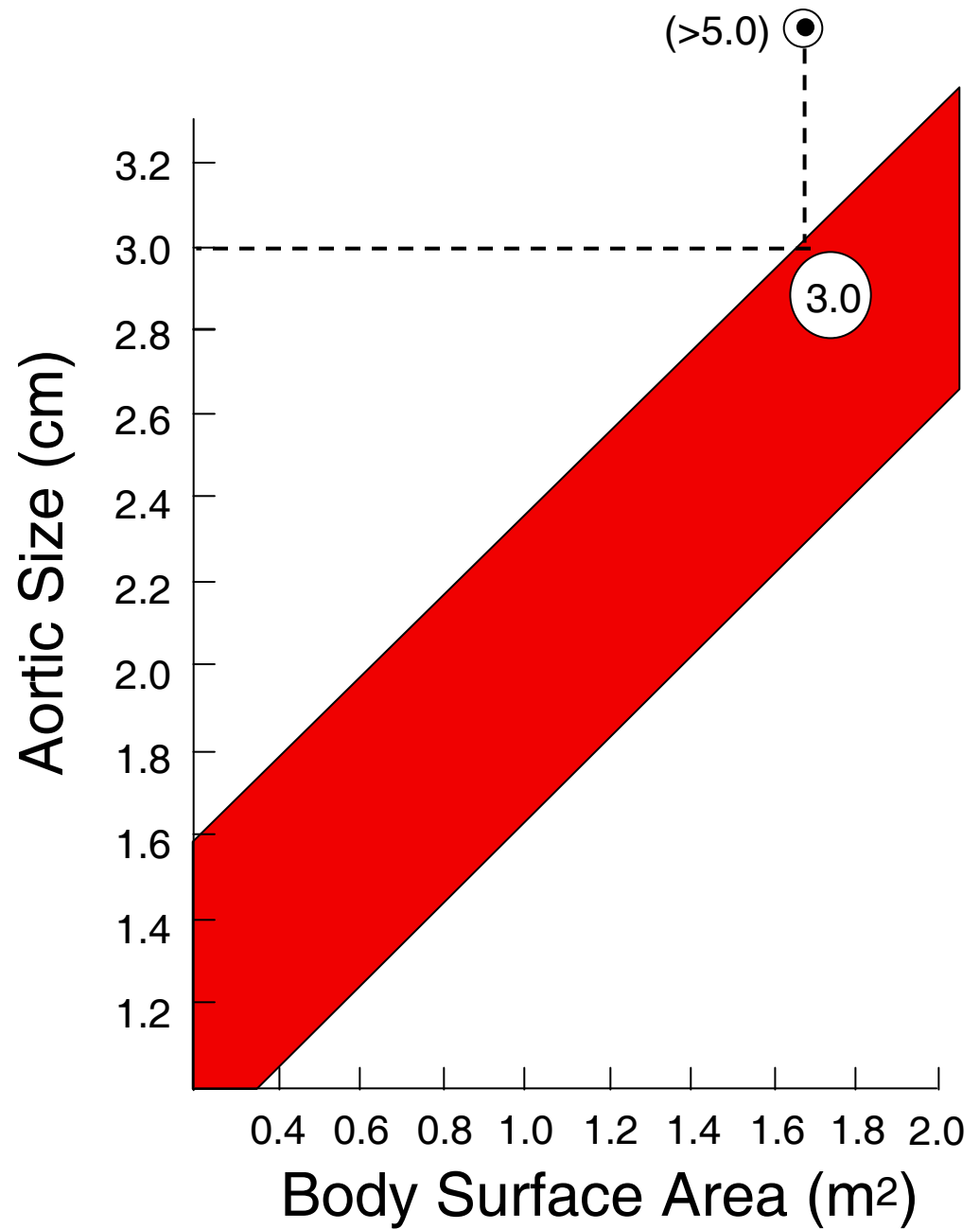
> 1cm / year

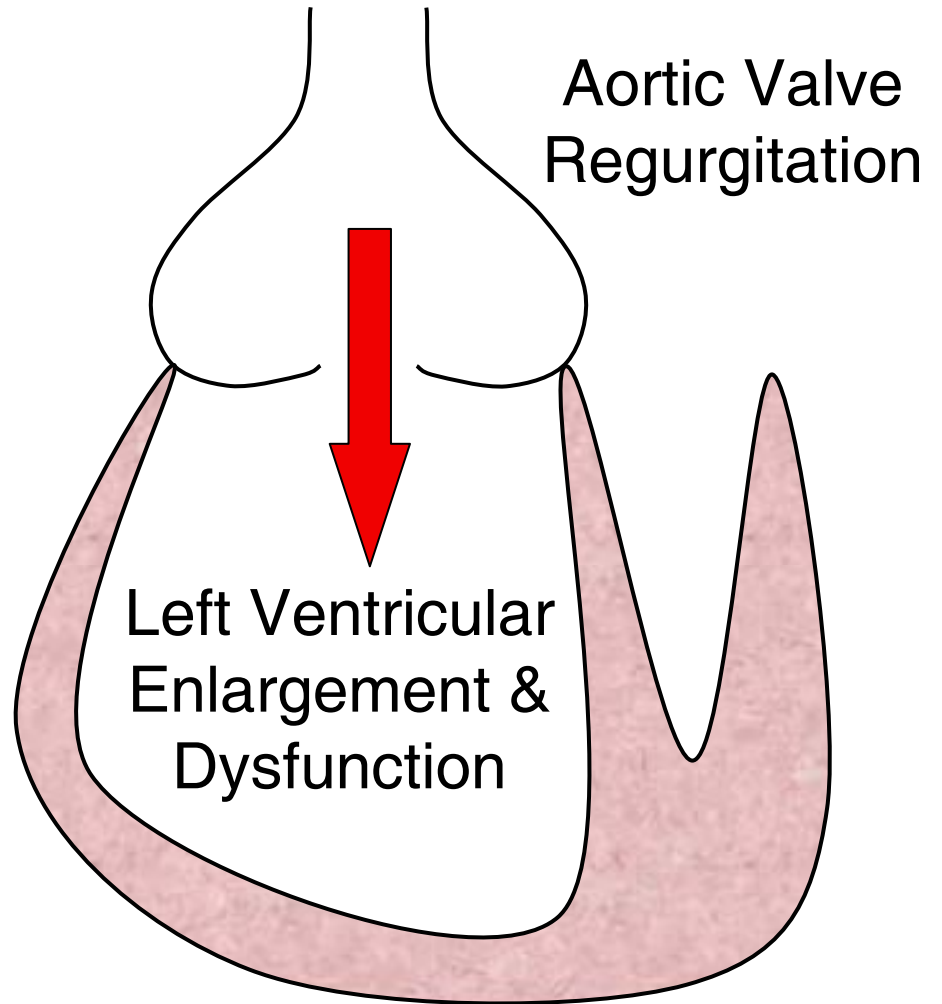
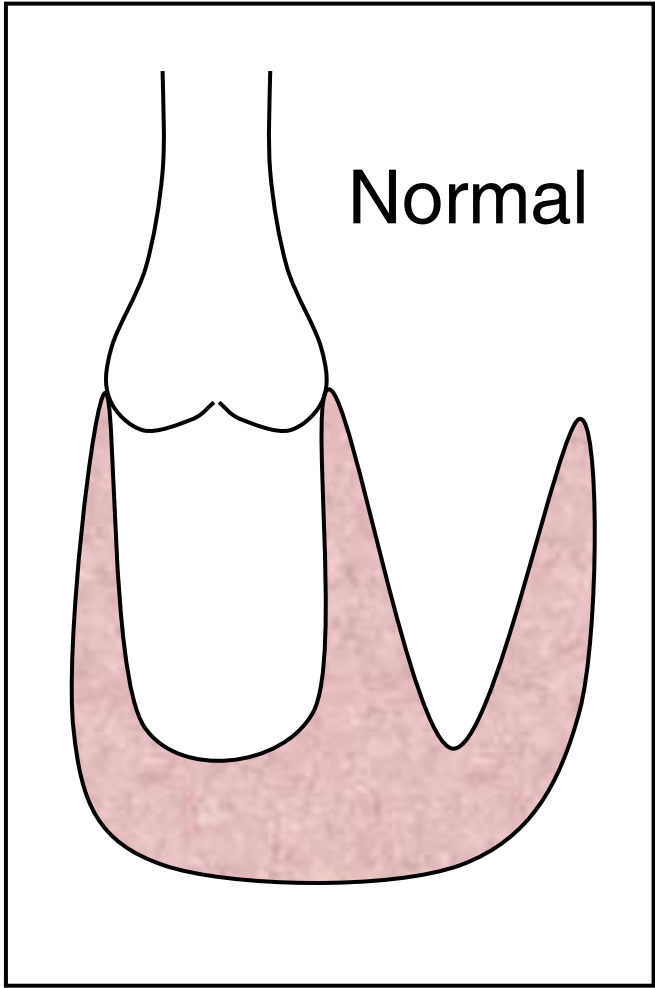


Value Twice Expected
For Body Size and Age

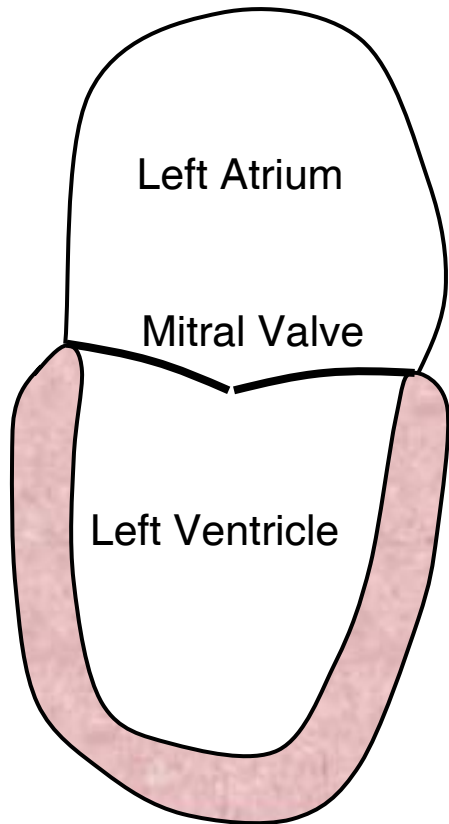


Value Has Reached
Adult Limit of ~5.0cm

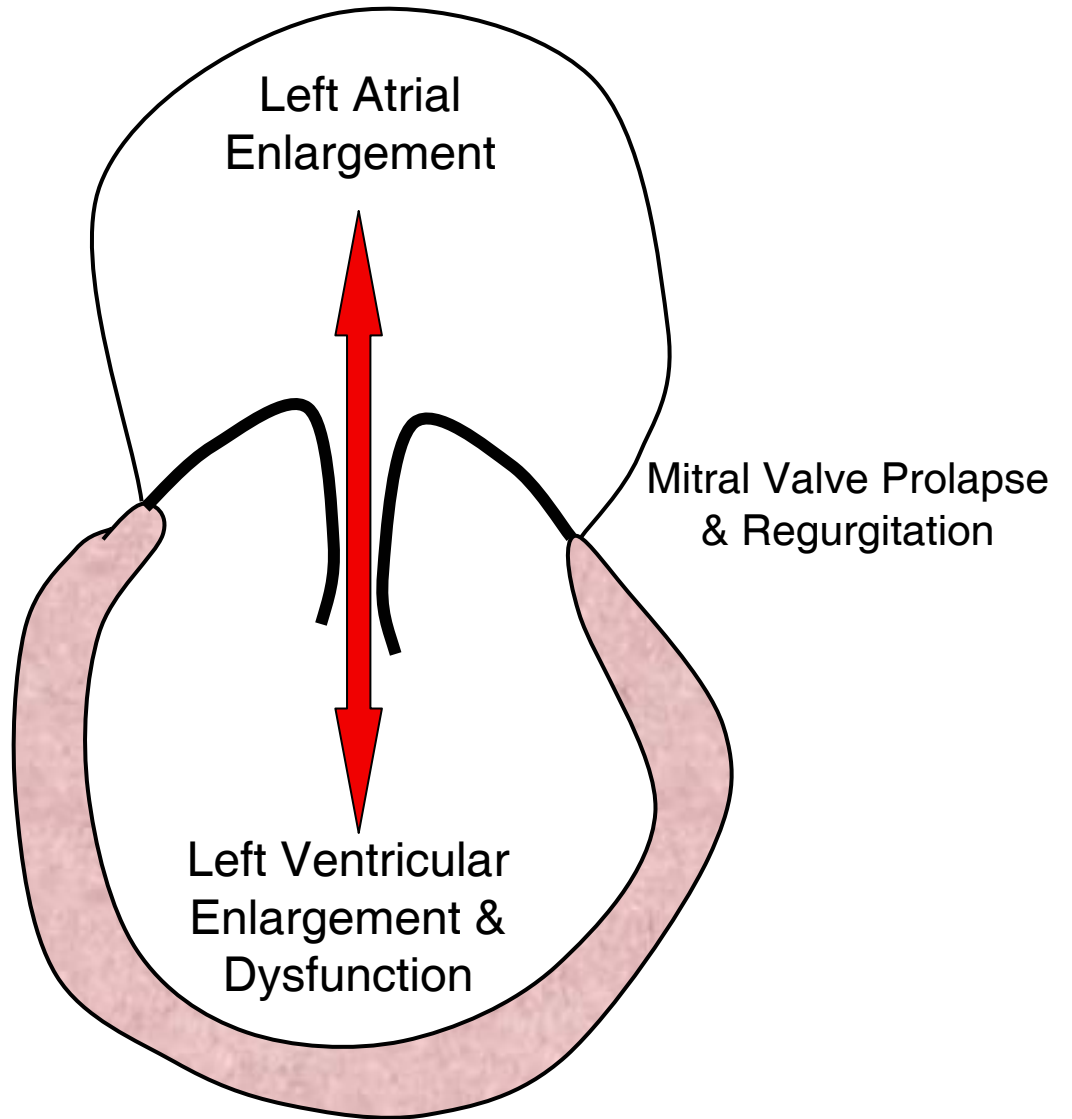




Normal



Left Atrial
Enlargement

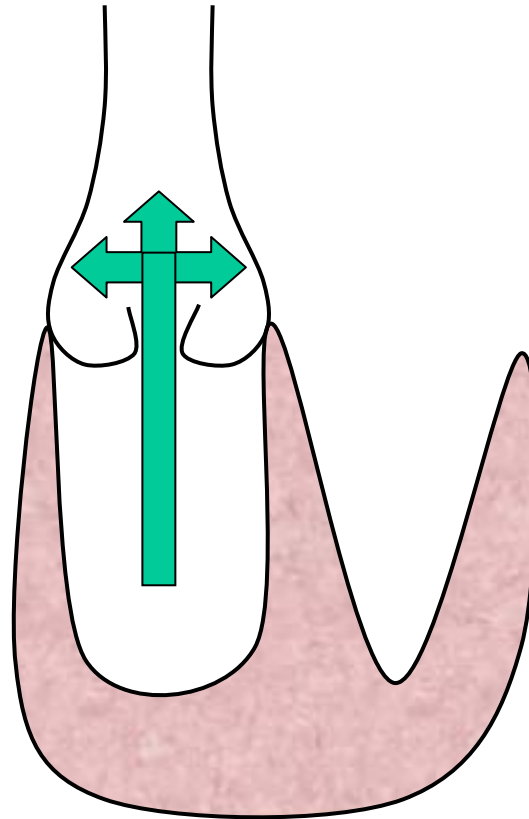


Beta-Blockers and Marfan Syndrome

Decrease Heart Rate

Decrease the force
with which blood is
ejected into the aorta

Slow the rate of
aortic enlargement



Who?
When?
What?
How Much?
Concerns?

Beta-Blockers and Marfan Syndrome

Who? When?

At the time of diagnosis with MFS
(? Wait until aortic enlargement)
Aortic enlargement but not MFS

What?

Infants:	Propranolol
Older children:	Atenolol (Tenormin)

How much? ENOUGH...

Measure heart rate after exercise:

Young children: heart rate < 110

Older children: heart rate < 100

Beta-Blockers and Marfan Syndrome

Concerns?

Generally well tolerated:

Lethargy, stomach upset often mild and transient
Most children report no problems with school

Should not be used with severe asthma, reactive airway disease

Occasionally, persistent lethargy, impaired learning prohibits use. Protecting the aorta in the context of impaired quality of life is not acceptable.

Alternative: Calcium Channel Blockers

Antibiotics to Prevent Cardiovascular Infection “SBE Prophylaxis”

People at risk:

MVP with regurgitation OR thickened leaflets	low
Artificial heart valve	high
Aortic repair with artificial or natural material	

Dental Work, Other Procedures Expected to Contaminate the Blood With Bacteria

		Children	Adults
Low Risk	Amoxicillin (oral, 1 hour b.)	50 mg/kg	2.0 g
High Risk (?)	Ampicillin (IM / IV, 30 min b.)	50 mg/kg (2.0)	2.0 g
	Gentamycin (IM / IV, 30 min b.)	1.5 mg/kg	1.5 mg/kg (120)

Exercise Restrictions and Marfan Syndrome

Concerns:

A sustained elevation of heart rate and blood pressure can promote aortic enlargement

A sharp blow to the head or chest can aggravate eye or vascular disease

Certain activities can contribute to joint discomfort and accelerate arthritic changes

Don't:

Participate in competitive, contact, or isometric activities such as weight lifting

Maintain activities that cause joint discomfort

Do:

Remain active with low intensity aerobic exercise

(**Roller Coasters:** depends on the intensity of the experience and the response of the child)

What's wrong with competitive soccer for a 5 year old?

- In most instances, there is no immediate danger.
- BUT, exercise restriction is an investment in the future. The goal is to promote a lifestyle that will maximize the situation 20 years later.
- After a few years, many children define themselves by the sport that they play. All their friends are competitive soccer players. All their support systems and social infrastructure resolves around soccer. At this point, restricting participation is devastating.
- Direct children toward sustainable activities at a young age.

Recognize that many kids need to try to win. To be the best. To be part of a group committed to a common goal.

Solutions:

- academic pursuits
- artistic pursuits
- selected athletic pursuits (sports that stress skill rather than endurance)

Golf

Bowling

Archery

Sports strategy (coaching)

Cardiac Surgery and the Pediatric Patient

Aorta:

Options:

Homograft:	no anticoagulants	reoperation
Composite Graft	anticoagulants	stable
Valve Sparing	no anticoagulants	? Stable

Mitral Valve

Most common indication for heart surgery in pediatric

Marfan syndrome

Generally considered only after failure of medical therapy

Repair rather than replacement possible in most cases

Can be performed simultaneously with aortic surgery

Cardiac Surgery and the Pediatric Patient

Dealing with Chest Wall Deformity:

In selected circumstances, severe chest wall indentation (pectus excavatum) can complicate cardiovascular surgery. It is best to discuss this possibility with the cardiovascular surgeon long before surgery is planned. If necessary, the chest repair should be performed and healed before proceeding with the heart operation. Simultaneous chest repair and heart surgery is not recommended.

More about pectus repair...

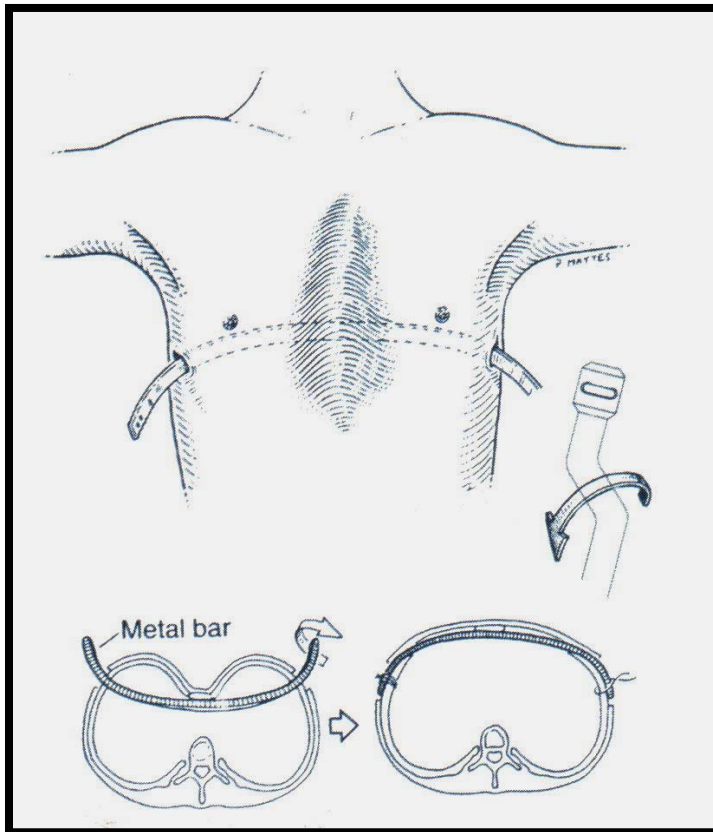
Many patients have restrictive lung profiles on pulmonary function tests, BUT this does not change after pectus repair performed after early childhood.

If repair is performed too early (pre-adolescence), pectus will likely recur.

Pectus repair is largely a cosmetic issue. This does not minimize importance!

Nuss Procedure

Who?
When?



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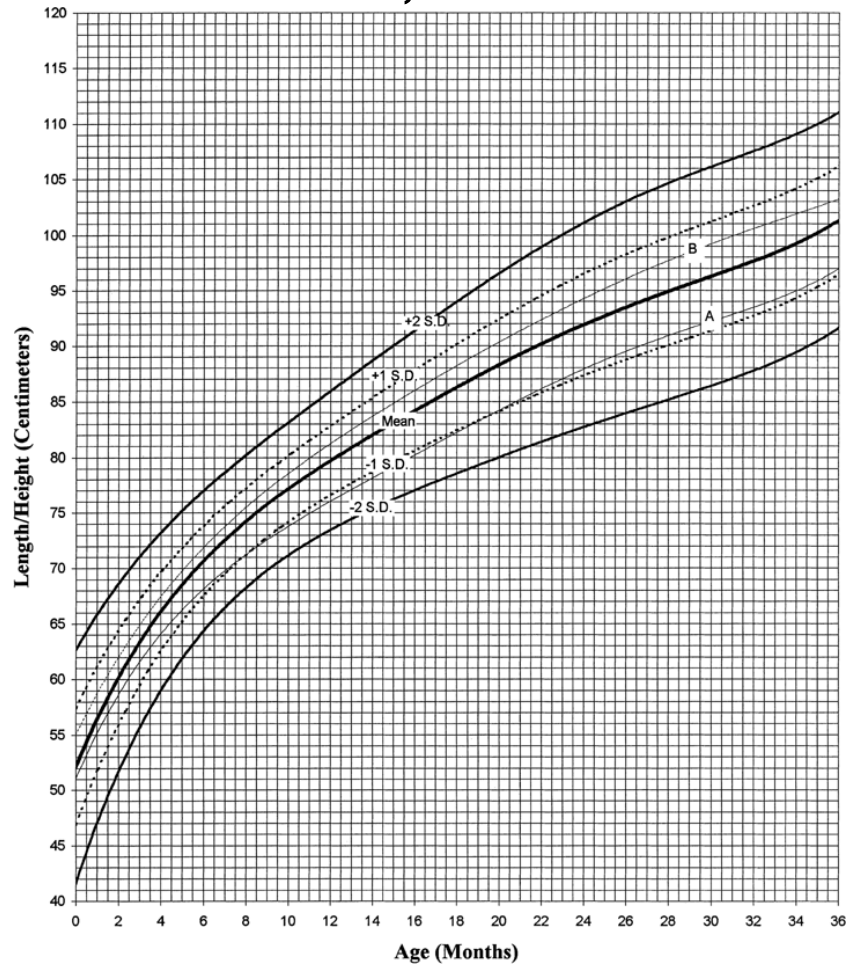
Growth

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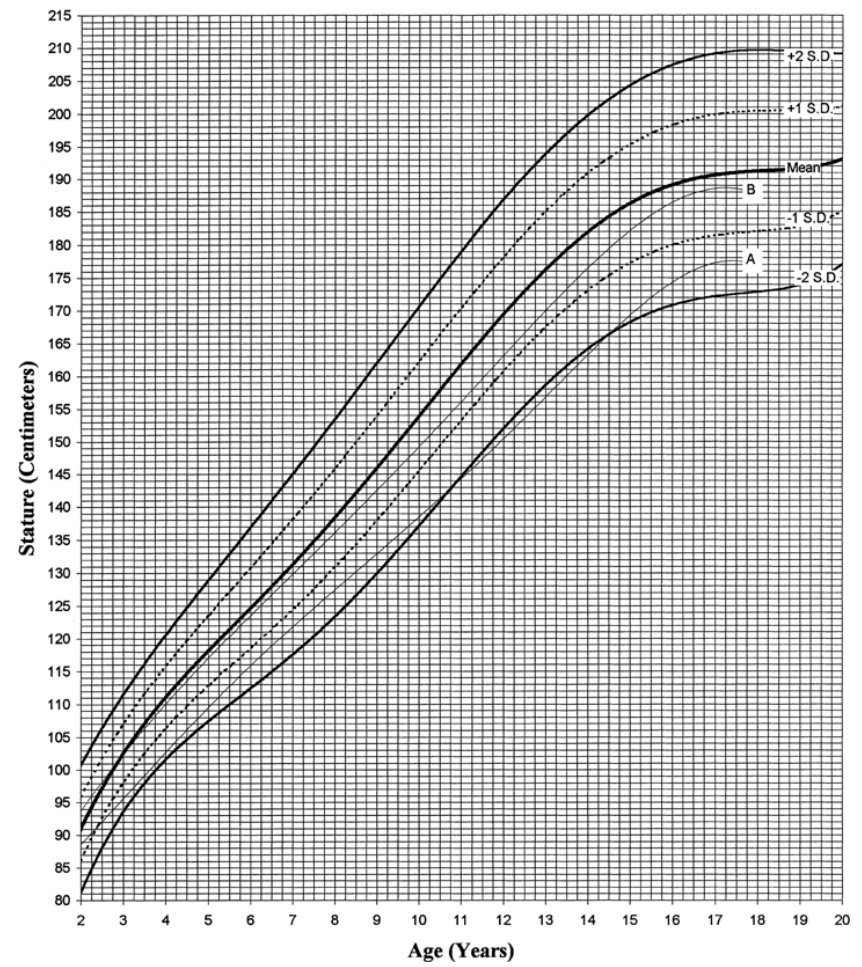
Communication

How big will he be?

infants, toddlers

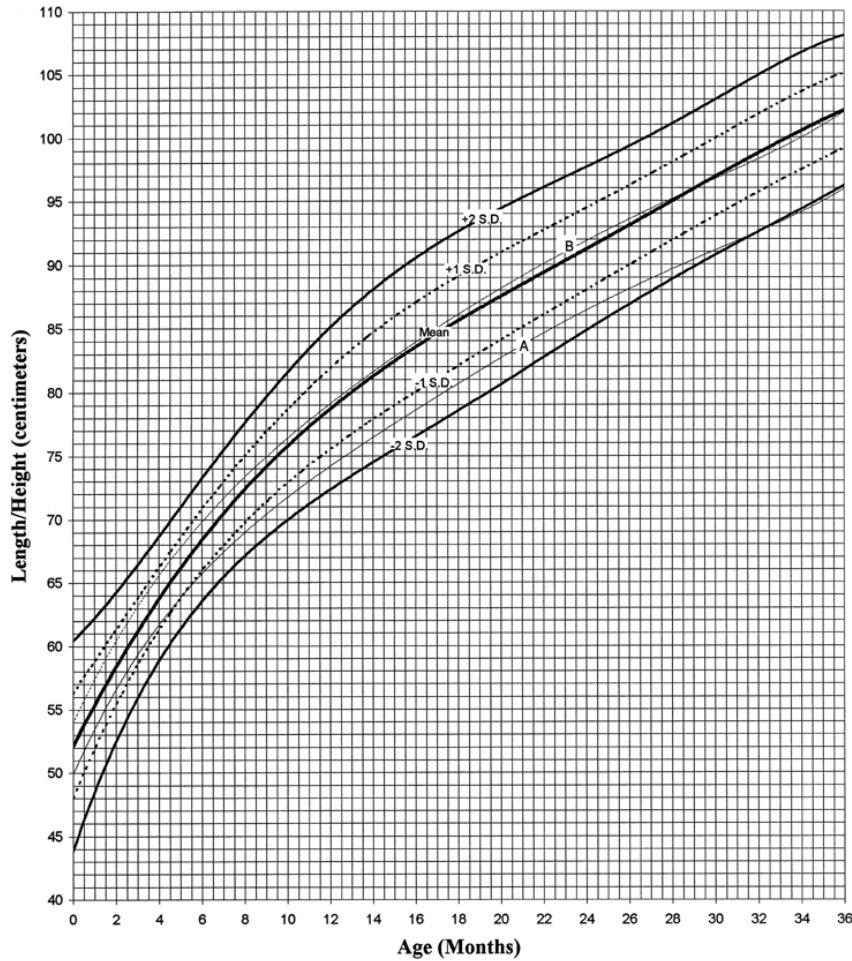


older children

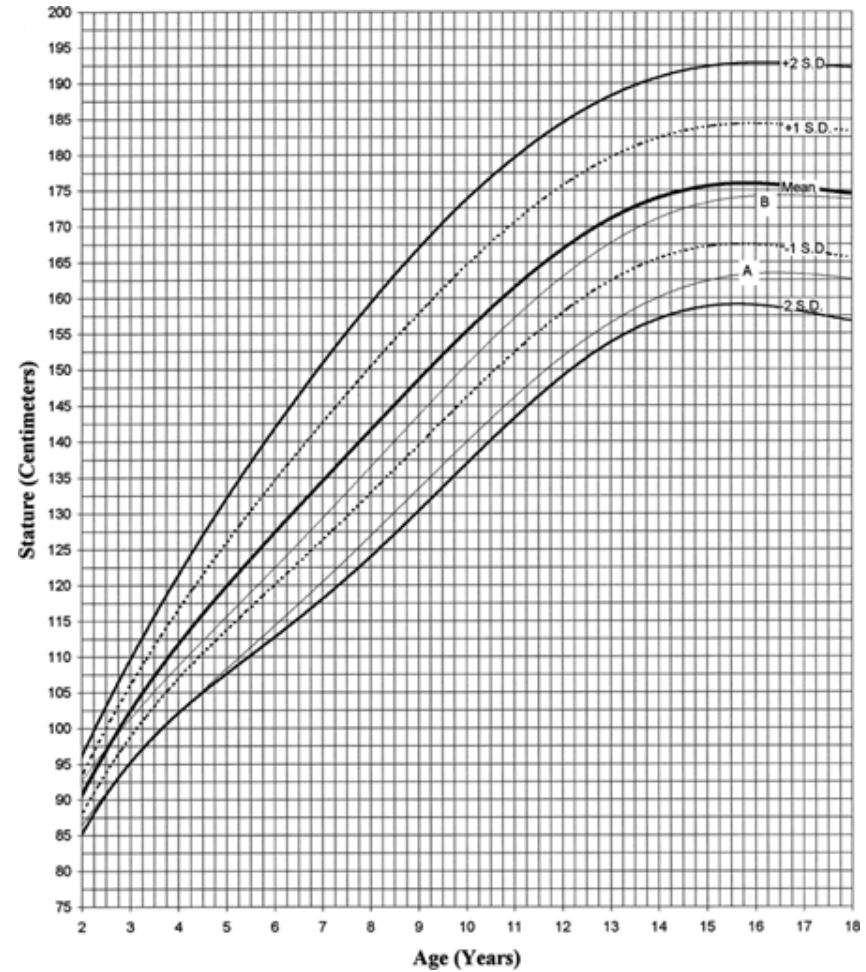


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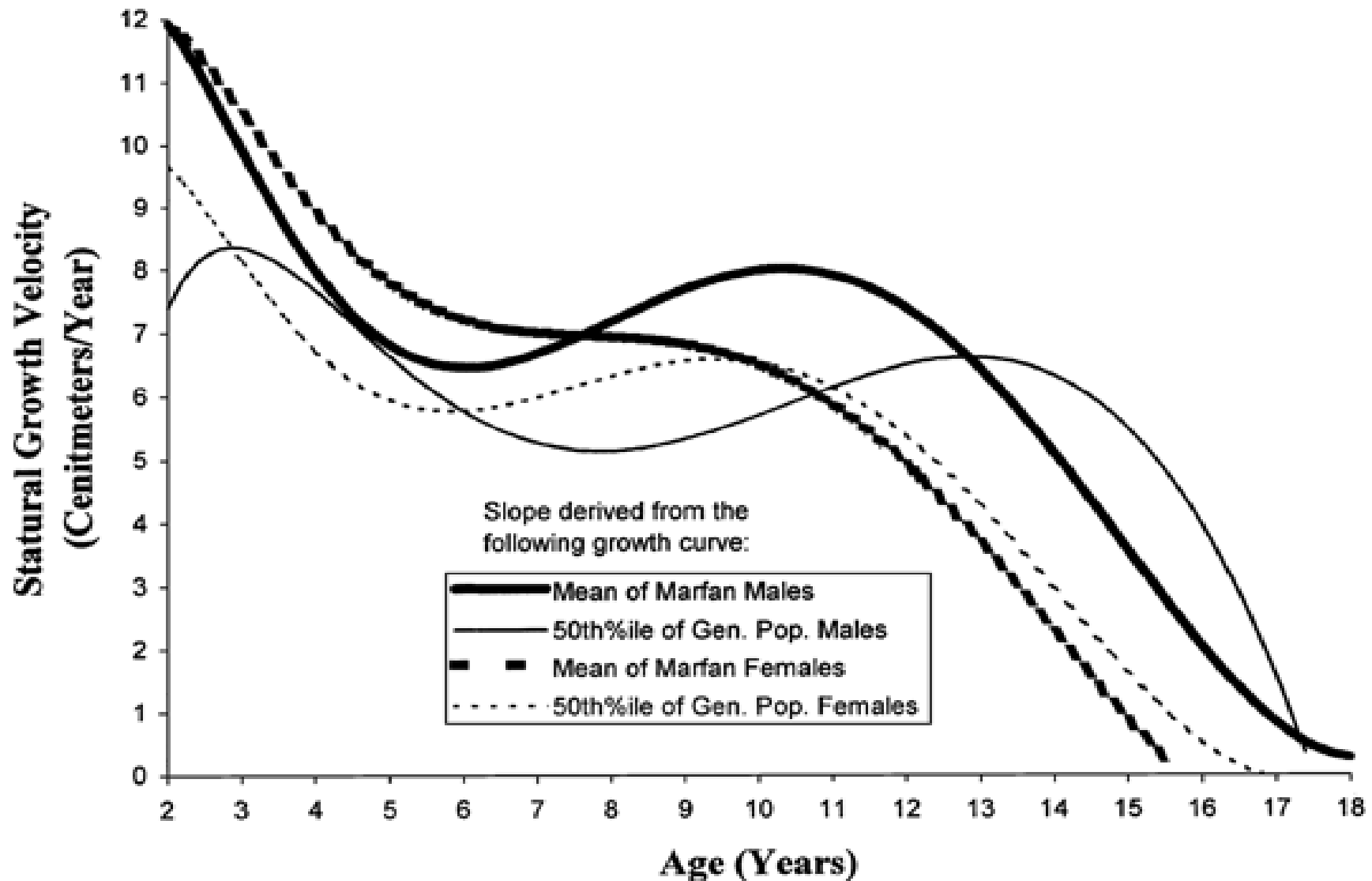
infants, toddlers



older children



Growth Velocity...faster and peaks earlier



Accurate estimate of final height requires determination of bone age. This is best accomplished with the combined expertise of a pediatric radiologist and endocrinologist.

(e.g. if A and B are same age and height, A is predicted to be taller than B once growth is complete.)

A



B

Estrogen therapy for tall girls with MFS...

Concept: Hormone therapy will accelerate maturation of the growth plate, resulting in a decrease in final height.

Success: Decrease of about 2 centimeters compared to predicted height. (Rx at 12yrs)

Modification: Start earlier?

Concerns: Initially accelerates growth.

Social concerns - sexual maturation.

Earlier increase in blood pressure.

Conclusion: Reserve for extreme height.

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Performance Problems and Marfan Syndrome

Learning Disability, Hyperactivity, Attention Deficit



NOT common, perhaps chance association.

Success with treatment with Ritalin if adequately beta-blocked.

Dealing with visual disturbance, muscle weakness, difficulties with coordination

Maximize Potential...

Sit close to board

Extra time for movement between classes

Portable computer

Thick pens / pencils

Participate to the extent of abilities / restrictions

Do **NOT** allow your child to routinely sit idle on the sidelines during gym class.

- 1) Advocate for complete participation in safe and appropriate activities and personalized exercise programs at other times.
- 2) If the school is unwilling or unable to meet these needs, your child is better off doing a life-enriching activity in the computer lab, library, etc.

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Marfan Syndrome

Parents Talking to Children:

Be open, honest, forthcoming.

Children will sense discomfort in silence.

Potential to amplify or misinterpret meaning.

Children will absorb information when they are ready.

Be receptive to questions.

Be sensitive to anxiety, changes in behavior.

Educate, but **BE POSITIVE**.

Provide role models.

Children Talking to Peers:

Friends will appreciate learning about Marfan syndrome.

Stress abilities, but also talk about special needs.

Everyone has limitations and strengths.

Differences are **minor**, and **not “bad”**.

Distributed by:



22 Manhasset Ave
Port Washington, NY 11050
(800) 8-MARFAN
www.marfan.org