



A/Prof Paul Hofman

Associate Professor at the Liggins
University of Auckland

The Diagnosis and Management of Short Children - Concurrent Workshop Repeated

Saturday, 17 August 2013

Start 8:30am

Duration: 55mins

Lounge 2

Start 9:35am

Duration: 55mins

Lounge 2



South GP CME 2013

General Practice Conference & Medical Exhibition

15-18 August | Edgar Centre | Dunedin

BLESSED ARE THE CRACKED
FOR THEY ARE THE ONES WHO
LET IN THE LIGHT

A Practical Approach to Short Stature

Paul Hofman
2013

Why bother?

3% of people will always be below the third percentile!
In the vast majority of cases there is no medical problem...

And yet

People complain of being short even when normal e.g. on the 25th percentile

There is great anxiety among many families with short children...especially where the parents are also short.

Is being vertically challenged a handicap?

SHORT STATURE and Society

Tall stature is valued – perhaps even more in shorter societies!

Males are taller than women in general and historically in most societies hold power.

Ruling classes possibly due to better nutrition and genetic selection are taller.

Thus height has become associated with power, intelligence and dominance.

In the USA height has been named as near top of the list in traits people value when choosing a mate

Heightism is as more prevalent than racism!

Short people are often the butt of jokes (look online at some of the websites!)

Many positive comments associated with tall stature
look 'up' to
stand 'tall'
holding in 'high' regard

Whereas there are negative associations with small size
talking 'down'
possessing 'short-comings'
'short man syndrome'

Heightism

There is no denying that we place a high premium on height, be it social, sexual, or economic, and our preference for height pervades almost every aspect of our lives.

Heightism is "one of the most blatant and forgiven prejudices in our society." Economist John Kenneth Galbraith (height 6'8").

1996 essay titled 'My Inner Shrimp' by Gary Trudeau creator of *Doonesbury* – “for the rest of my days I will be a recovering short person”. His final height was over 184cm but was very short as a child.

Pathological Short Stature

Short stature or even more sensitive, poor growth velocity can reflect underlying pathology.

Linear growth is extremely sensitive to any alteration in either the external or internal environment.

Poor growth is therefore a very sensitive, objective parameter indicating an underlying problem but it is not very specific.

SHORT STATURE

Common perceived problem in the community and most common referral to paediatric endocrinologists.

Boys referred >> girls

99% of short stature is NORMAL VARIANT

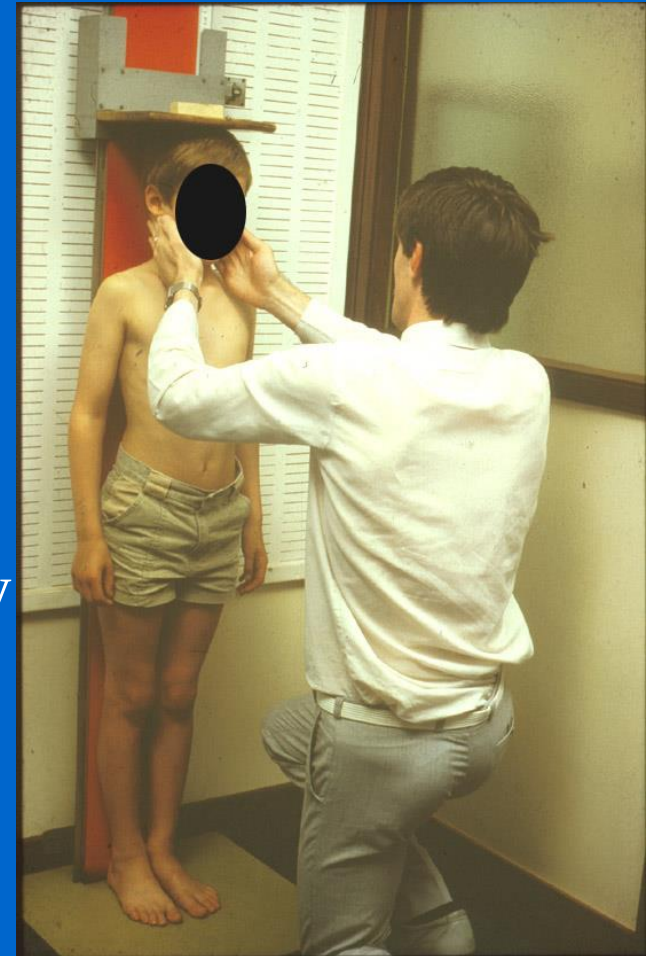
IT IS ESSENTIAL TO UNDERSTAND NORMAL
VARIANT GROWTH AND DISTINGUISH THIS FROM
PATHOLOGICAL CAUSES OF SHORT STATURE.

SHORT STATURE Awareness!

To identify short stature you have to

1) MEASURE the patient

- have an accurate stadiometer
 - firm base
 - rigid measuring bar
 - standard length rod for checking height consistency
- know how to measure reliably and consistently
 - no shoes
 - straight legs
 - no tip toeing/ slouching
 - face in neutral plane with slight traction



SHORT STATURE Awareness!

2) PLOT the data!

No one can accurately assess short stature by eyeballing a child.

Appropriate growth charts are needed. These available free by Pfizer or on the APEG website (Australasian Paediatric Endocrine Group) under clinical resources.

<http://www.apeg.org.au/>

Looking at growth patterns is more useful than one point ie measure and plot all children at least annually if possible.

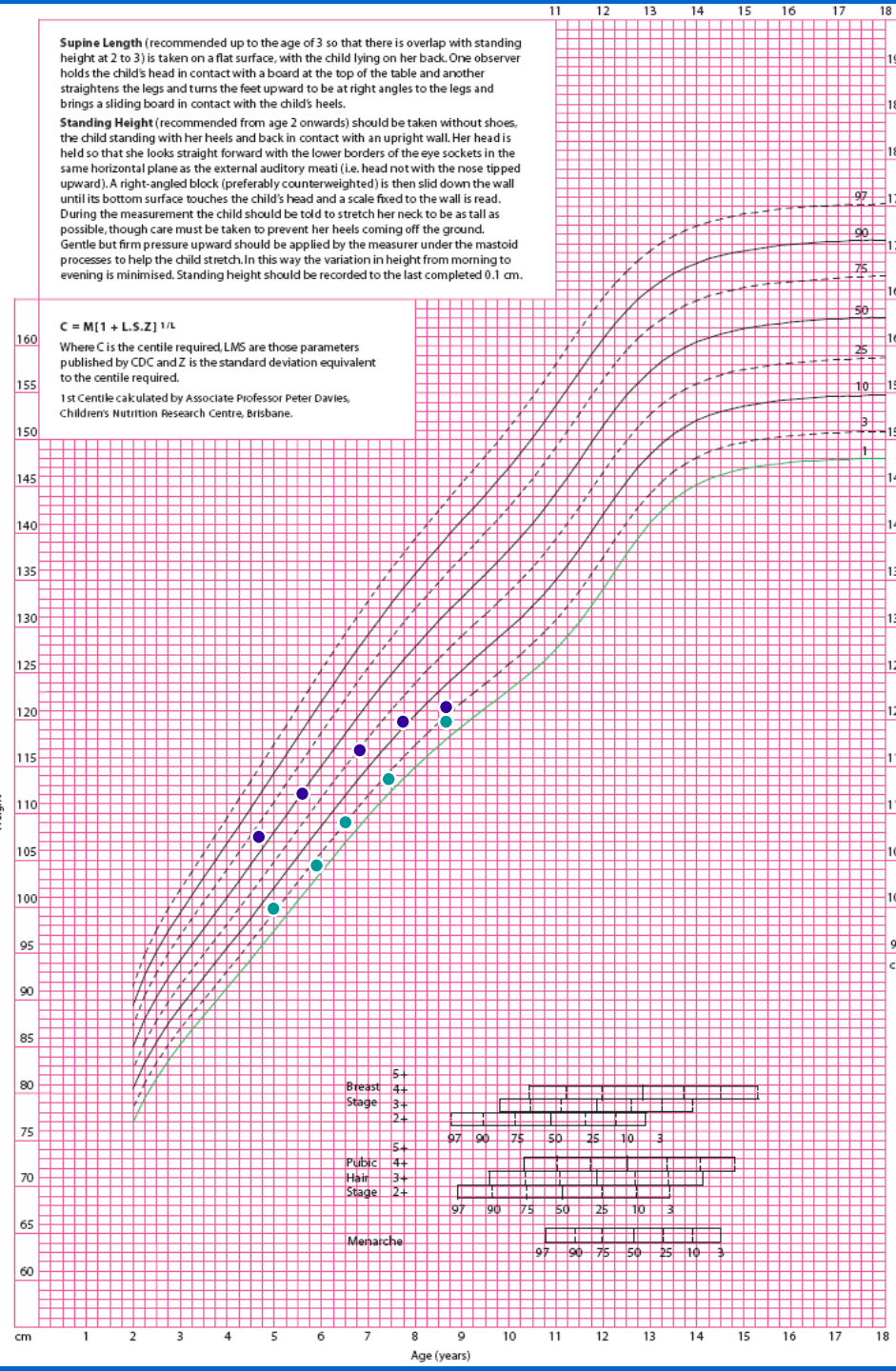
Supine Length (recommended up to the age of 3 so that there is overlap with standing height at 2 to 3) is taken on a flat surface, with the child lying on her back. One observer holds the child's head in contact with a board at the top of the table and another straightens the legs and turns the feet upward to be at right angles to the legs and brings a sliding board in contact with the child's heels.

Standing Height (recommended from age 2 onwards) should be taken without shoes, the child standing with her heels and back in contact with an upright wall. Her head is held so that she looks straight forward with the lower borders of the eye sockets in the same horizontal plane as the external auditory meati (i.e. head not with the nose tipped upward). A right-angled block (preferably counterweighted) is then slid down the wall until its bottom surface touches the child's head and a scale fixed to the wall is read. During the measurement the child should be told to stretch her neck to be as tall as possible, though care must be taken to prevent her heels coming off the ground. Gentle but firm pressure upward should be applied by the measurer under the mastoid processes to help the child stretch. In this way the variation in height from morning to evening is minimised. Standing height should be recorded to the last completed 0.1 cm.

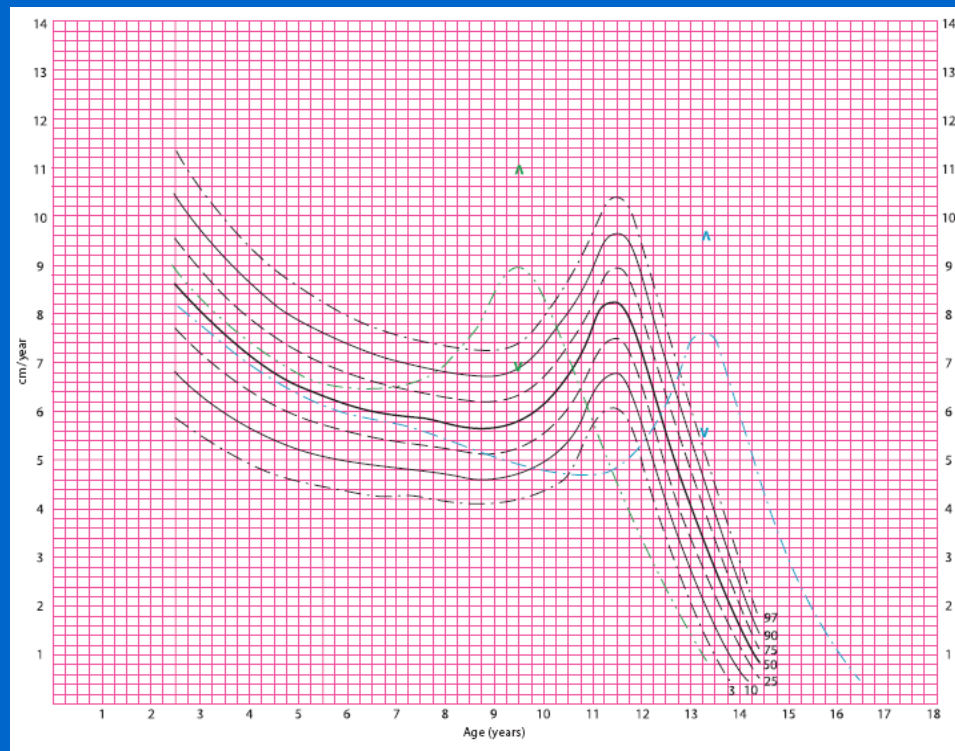
$$C = M[1 + L.S.Z]^{1/4}$$

Where C is the centile required, LMS are those parameters published by CDC and Z is the standard deviation equivalent to the centile required.

1st Centile calculated by Associate Professor Peter Davies, Children's Nutrition Research Centre, Brisbane.



Girls 2-18 year Growth Charts



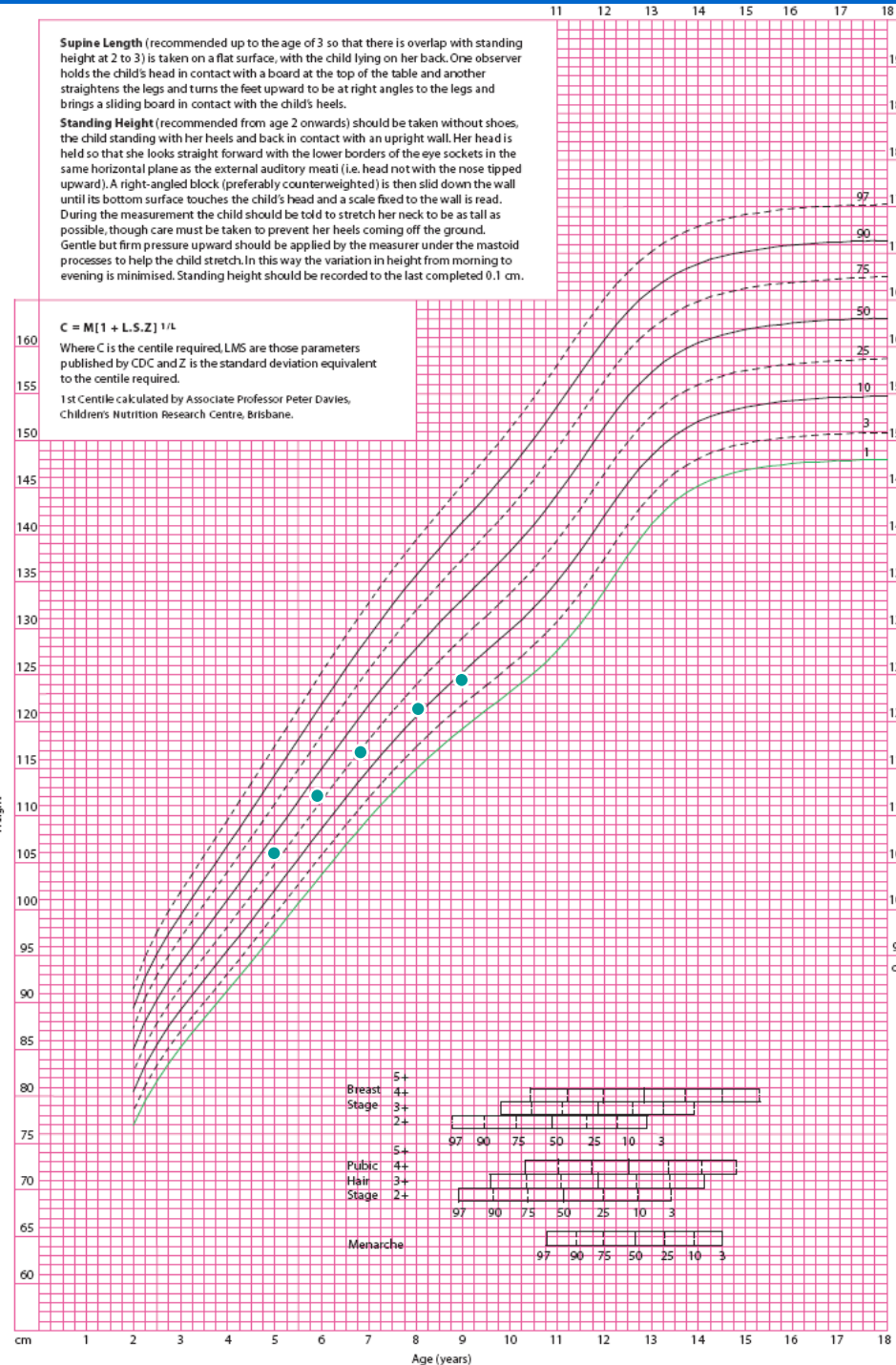
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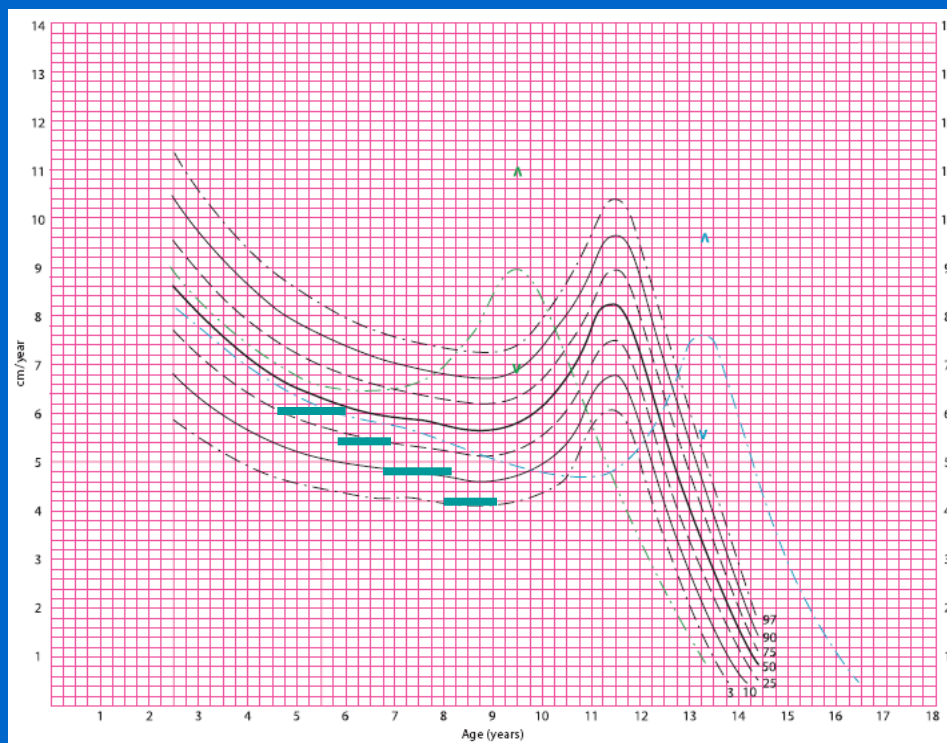
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Girls 2-18 year Growth Charts



Normal Growth

REQUIREMENTS for growth/ anabolism

- Nurturing, caring environment
- Adequate nutritional supply and the ability to digest and absorb the food (ie the child should be well nourished)
- Appropriate hormonal milieu
- Appropriate extracellular and intracellular environment.

Normal Variant Growth – Constitutional Delay of Growth and Development

- Child's height is well below that expected from the mid parental height.
- **Growth velocity is normal** (ie >25th %ile)
- Normal birth weight
- Family hx of pubertal delay and late growth common.
- **Bone age delayed** (although usually not more than 2 years)
- Final adult height normal (usually close to mid parental height)

Normal Variant Growth- Constitutional Delay of Growth and Development

Characteristic history is normal growth for the first 6-9 months after which they cross percentiles downwards for 6-12 months. They then grow parallel to this percentile until late childhood.

Puberty is entered late with a corresponding delay in epiphyseal fusion and consequently extra growth.

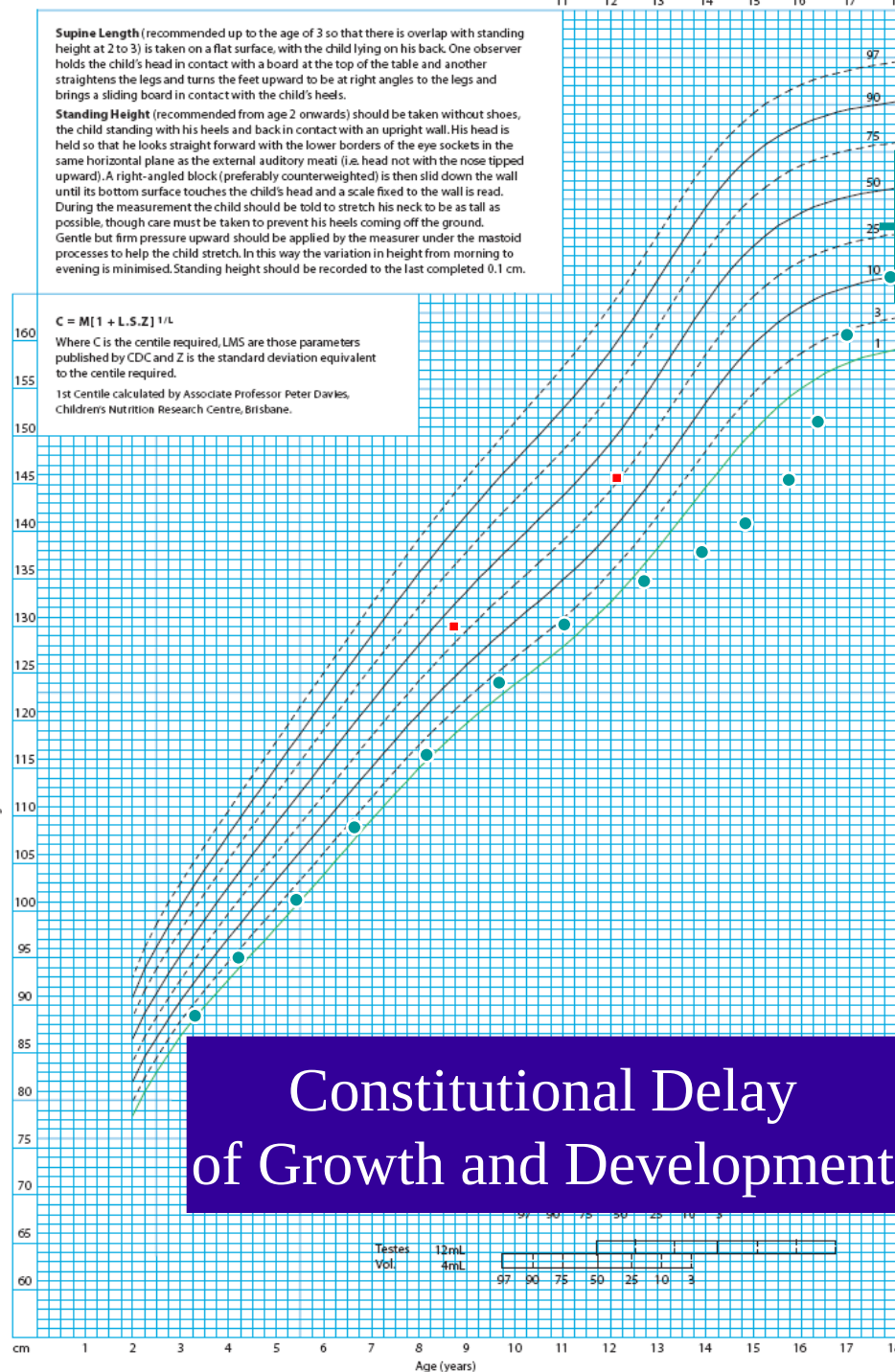
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MPH

Constitutional Delay
of Growth and Development

Normal Variant Growth- Familial Short Stature

- Short parents (beware one short parent - can represent a dominant genetic problem).
- Normal growth velocity (ie >25th PC)
- Normal birth weight
- Bone age appropriate for chronological age.
- Final height consistent with mid parental height.
- Typically these children grow down to their height percentile over the first 6 months after birth and then grow along and parallel to this percentile.

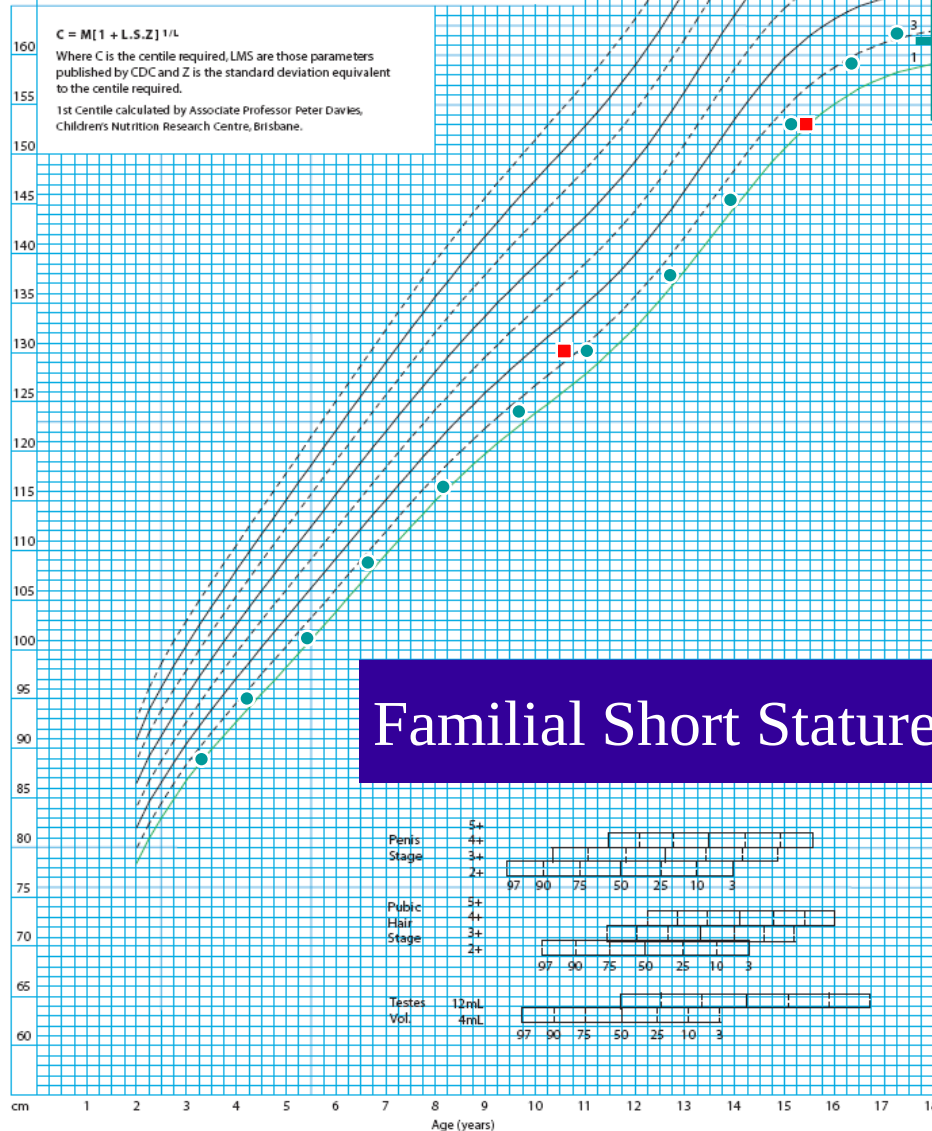
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Familial Short Stature

MPH

Short Stature - Classification

Large number of causes

All pathological causes of short stature will cause a poor growth velocity (<25th PC) and a crossing of percentiles downwards on a growth chart.

Short Stature - Classification

Proportionate (ie leg length to body length and arm span are normal)

Disproportionate (shortening of either limbs or trunk)
Almost always due to a skeletal dysplasia (eg hypo and achondroplasia)

A large no. of conditions, which combined are still uncommon.

Define by measuring armspan (= height $\pm$ 5cm) or sitting height (easier to measure and charts readily available)

Short Stature - Proportionate

Post Natal

Normal Variant

Pathological

- Nutritional (starvation commonest cause of SS worldwide)
- Chronic Illness ie renal, cardiac, GI, respiratory, neurological.
- Severe neglect can cause psychosocial dwarfism
- Chronic Drug Intake mainly glucocorticoid tx.

Short Stature - Proportionate

Prenatal

Intrauterine Growth Retardation

uteroplacental insufficiency

congenital infection

syndromal (Downs, Noonans etc)

Chromosomal (Turner syndrome (XO) commonest cause of pathological SS in girls (1:2000 female births - always consider in girls!)

Antenatal Drug abuse (esp. alcohol)

Short Stature - History

Growth pattern

Antenatal history and **birth weight**

Hx of any chronic illness or medication

Hx of malnutrition/ neglect

Family Hx

Mid-parental height $\frac{(\text{Mother's ht} + \text{Father's ht} \pm 13\text{cm})}{2}$

95% confidence intervals are $\pm 8\text{cm}$

Hx of delayed puberty

Short Stature - Examination

Measure and plot height (plus other previous measurements if available)

Limb asymmetry/disproportion

Pubertal Status

Dysmorphism

Evidence of organ system disease

Short Stature - Investigation

Standard Investigation

BONE AGE (skeletal survey occ. required if a skeletal dysplasia is suspected)

If there is an abnormal growth pattern (measurements at least 4 months apart (preferably 6-12 months apart).

Thyroid function tests

FBC + ESR

U+Es, Creatinine, (LFTs)

Urinalysis/ pH

Karyotype (girls)

Coeliac screen

(faecal spec/ LFTs/ capillary blood gas)

Short Stature - Investigation

If growth failure persists

Refer to paediatric endocrine services

Short Stature - Diagnosis

A pathological growth pattern can be established from growth data, if necessary taken over a 4-12 month period.

Plotting previous growth data will often help establish the length of time the problem has been present.

A normal growth velocity **over a 12 month period** virtually excludes a pathological cause for short stature.

Summary

- 1) Measure and plot children at least annually
- 2) A normal growth velocity essentially excludes significant pathology.
- 3) Always calculate the midparental height and ask about a history of pubertal timing.
- 4) A bone age is the only 'must do' investigation. Waiting until you can calculate a growth velocity before doing other tests is otherwise appropriate.
- 5) In short girls growing poorly ALWAYS do a karyotype.

Case 1

A concerned mother presents her 5 year old son with short stature

Really only noticed he is short now

What are the common ages of presentation with short stature?

Case 1

What are the common ages of presentation with short stature?

Starting school

Starting high school

Case 1

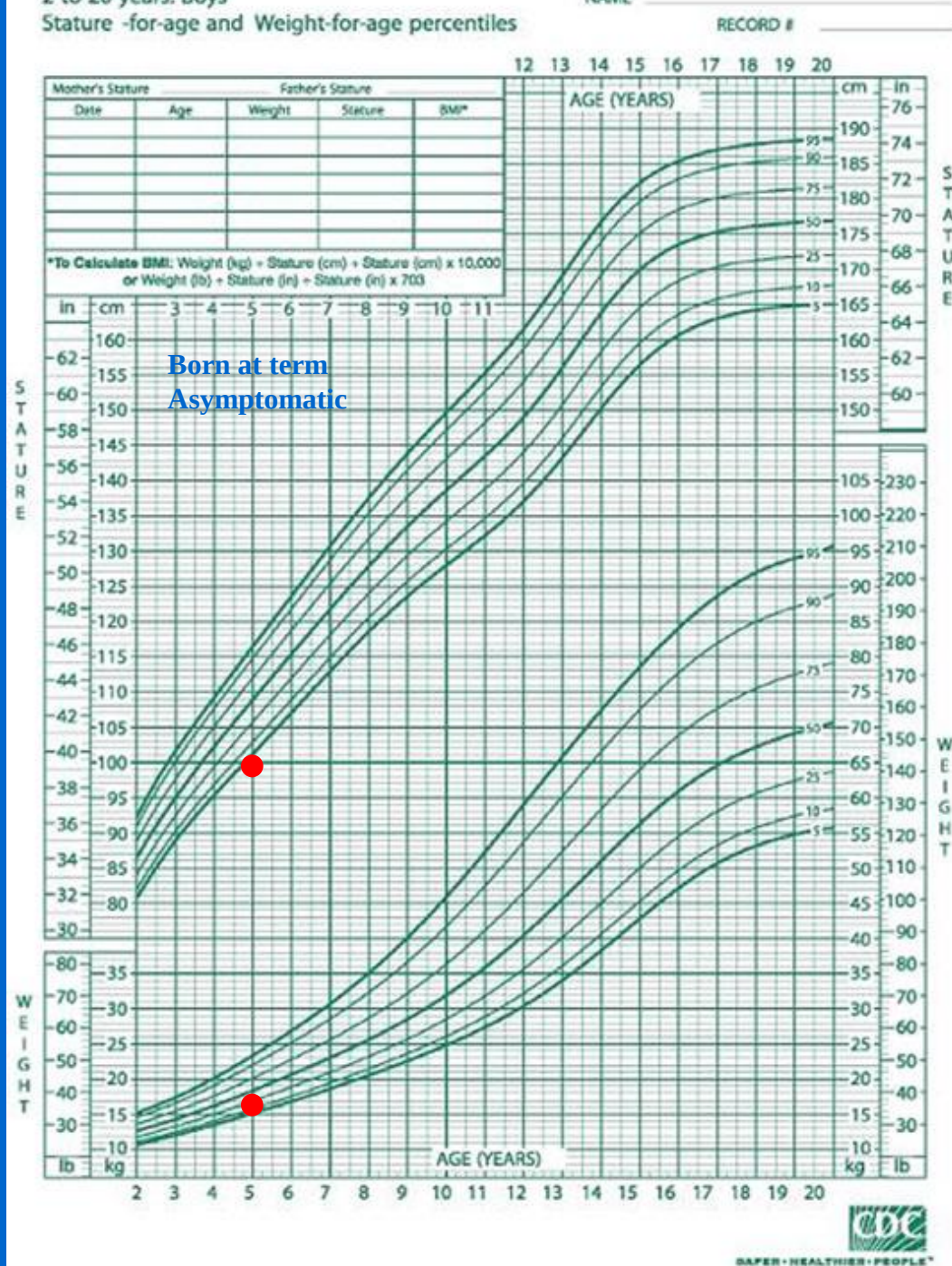
Born at term, normal delivery,

Normal birth weight

Completely healthy

.

Case 1



Case 1

What more do you want to know and why?

-

Case 1

What do you want to know and why?

No interval history of note

No medications

No pregnancy illnesses/drugs

Birth weight 3,600 gm

Father 166 cm

Mother 151 cm

Parents school teachers

Mothers menarche 12 years, Father has no idea

Mid-parental height

Mother and fathers heights

$$\text{MPH} = \frac{M(\text{cm}) + F(\text{cm})}{2} \pm 13 \text{ cm}$$

MPH range $\pm 8 \text{ cm}$

$$(151 + 13 + 166) / 2 = 165 \text{ (157-173) cm}$$

3rd percentile

Plot on growth chart

Case 1

What diagnostic investigations should
you perform now?

Case 1

What diagnostic investigations should
you perform now?

Bone age only

5 years (=Chronological Age)

Short stature screening

1375 referrals to CHMC Cincinnati

325 had Ht <3rd PC, normal history/exam

Normal variant short stature 99%

Sisley J Pediatr 2013; May 21 [Epub]

Short stature screening

New pathology 1.3% (2 coeliac disease)

Screening cost to identify new pathology case:
\$US105,107.00

“Healthy short children do not warrant
non-directed, comprehensive screening”

Sisley J Pediatr 2013; May 21 [Epub]

Case 1

What is the single most important piece of clinical information you should try and obtain?

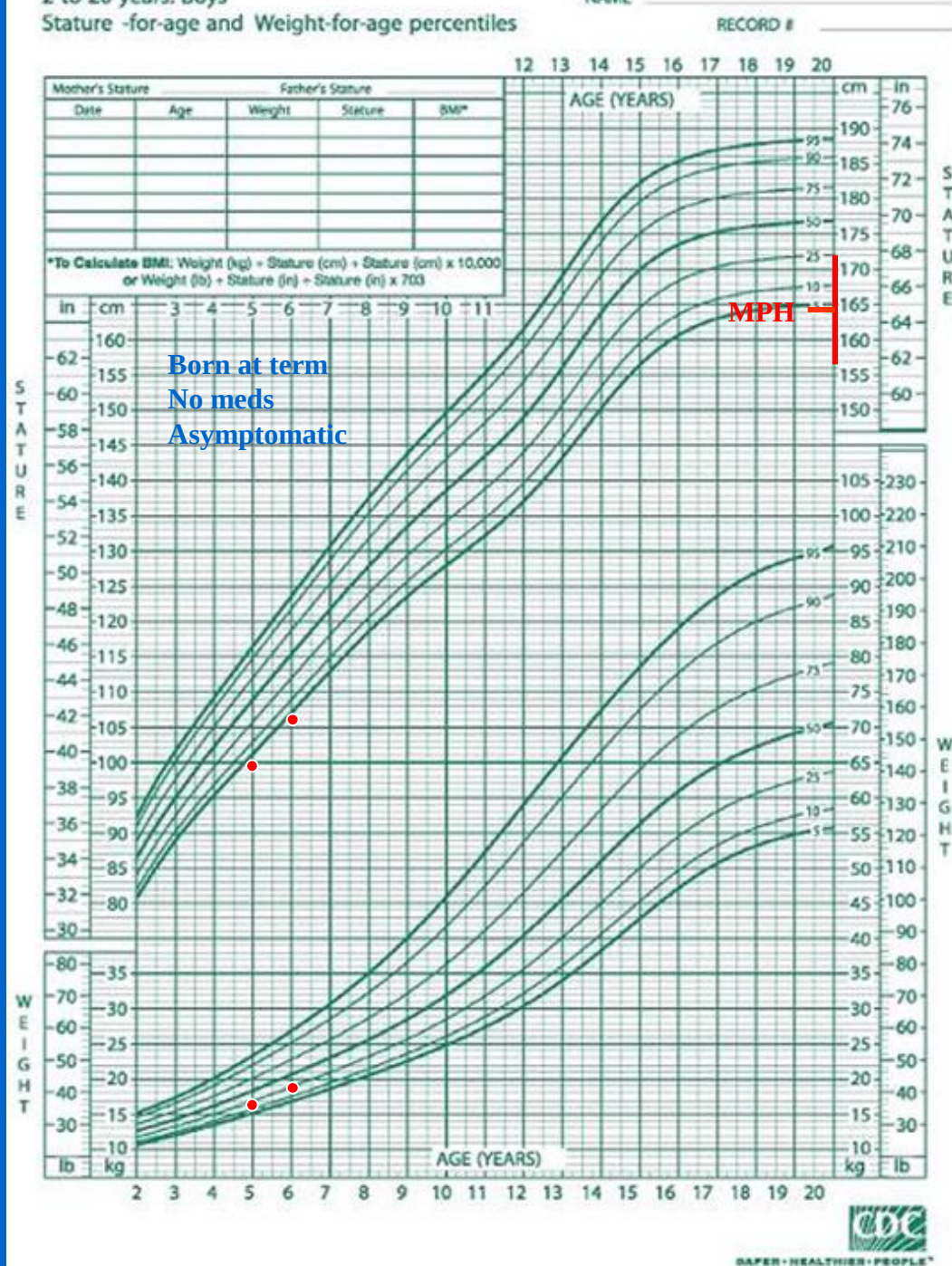
Height velocity (HV)

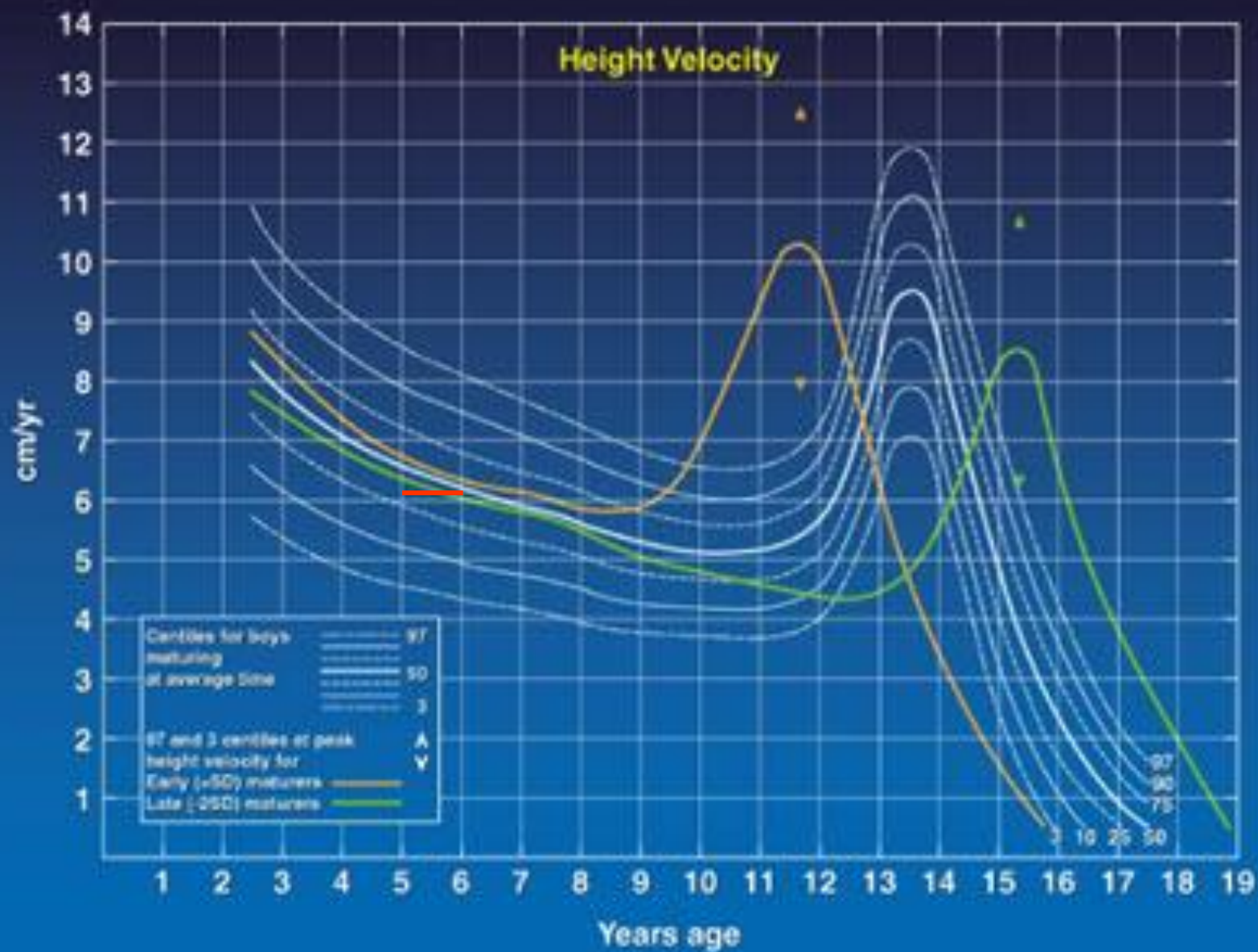
Measured over a minimum of 6 months

HV differentiates normal variant short stature from pathological short stature

Normal height velocity 25-75 PC

Case 1





Case 1

What is the most likely diagnosis?

Case 1

What is the most likely diagnosis?

Familial short stature

Case 2

A 12 year old girl presents with short stature.
She is a keen netballer and is finding it difficult to compete.

Recurrent otitis media all through early childhood (grommets)

No maternal pregnancy illnesses/drugs

Birth weight 3,200 gm

Father 181 cm, early developer

Mother 162 cm, menarche 12 yrs

MPH 165 cm (50th percentile)

Father bus driver, mother house wife

Oldest of two children (brother 6 yrs old, tall)

NAME _____

RECORD # _____



MPH

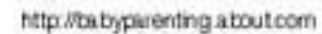
Case 3
Aged 7 yrs

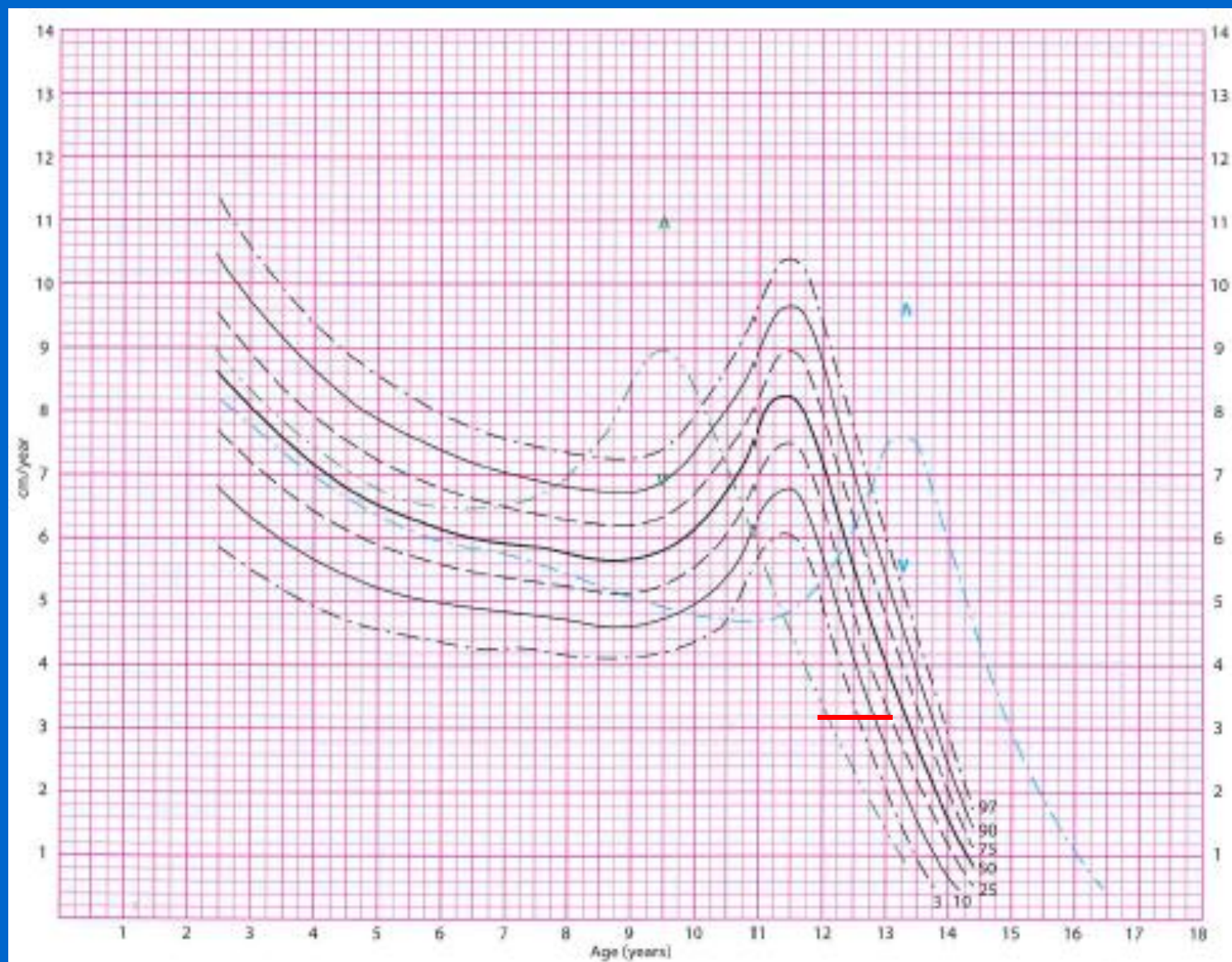




Stature-for-age and Weight-for-age percentiles

RECORD ●





Case 2

Are further investigations required?

Case 2

Are further investigations required?

Karyotype

TFTs

FBC+ESR

Electrolytes and renal function

Case 3

Are further investigations required?

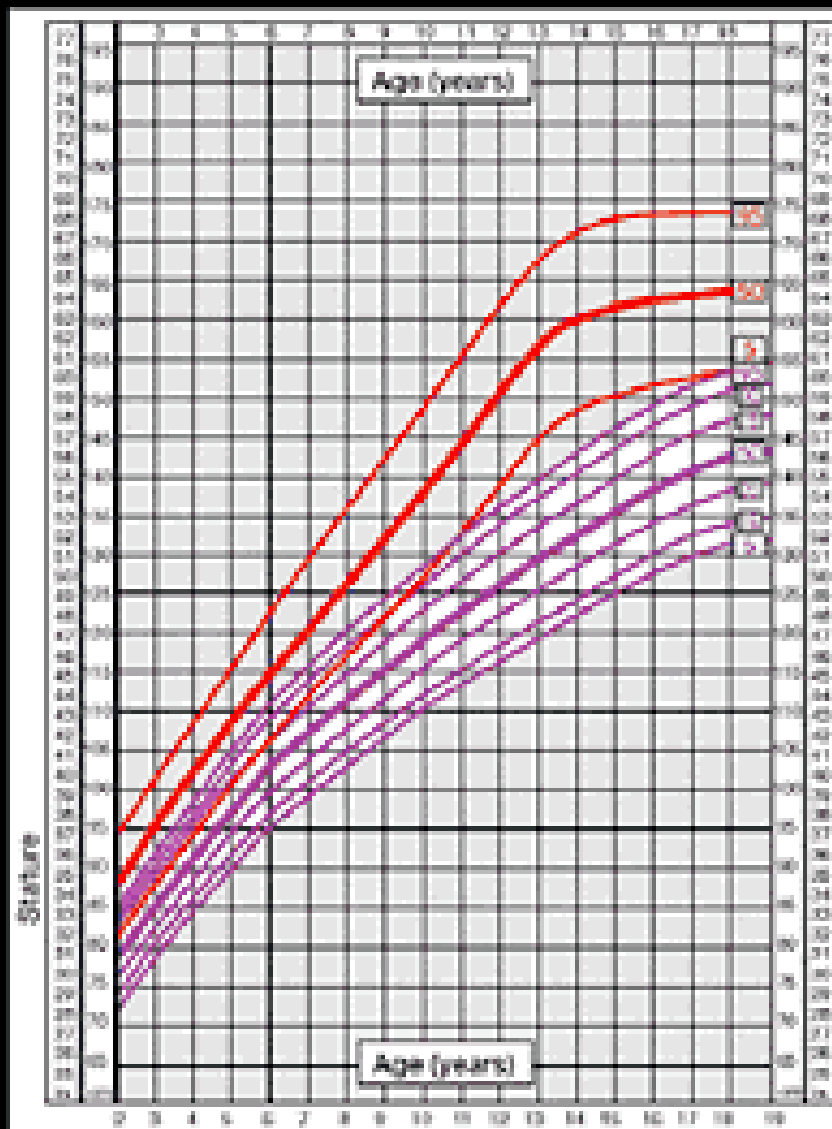
Karyotype: XO

TFTs

FBC+ESR

Electrolytes and renal function

Growth Chart: Girls With Turner Syndrome



Turner Syndrome

Consider in all girls with unexplained short stature or Ht below MPH range.

Commonest feature is short for MPH (100%).

50% will only have short stature as clinical feature.

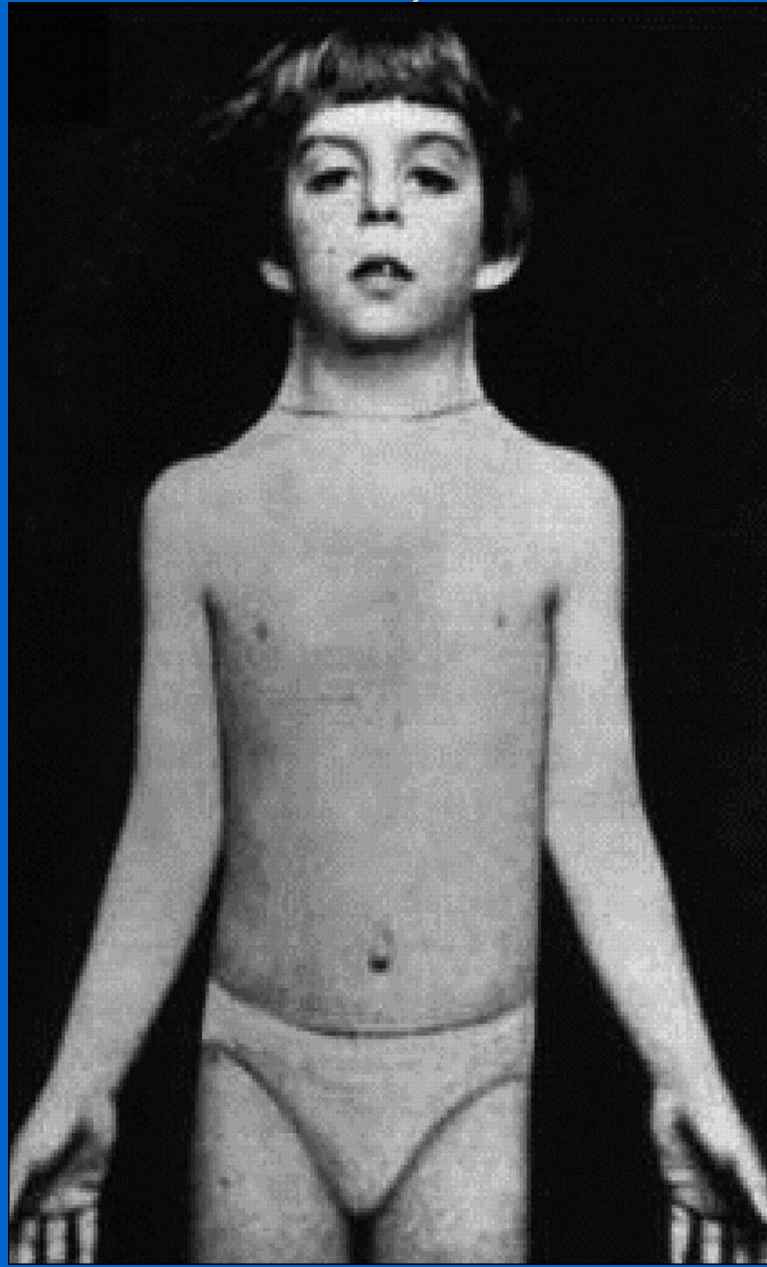
Present with short stature, poor HV or delayed puberty.

Management:

Growth hormone therapy for short stature

E₂ for puberty and beyond

Turner Syndrome



Case 3

A concerned mother presents her 13 year old son with short stature

Noticed at starting secondary school how short he was compared to peers – “they all look like men!”

Mild asthma treated with flixotide 200 mcg bid

No pregnancy illnesses/drugs

Birth weight 3,250 gm at term

Father 175 cm

Mother 165 cm

MPH 176.5 cm (50th percentile)

Father accountant, mother lawyer

Youngest of three children

What other aspects of history do you want to know?

Case 3

Mother menarche 14 yrs

Father still growing when started university

18 yr old brother stopped growing at secondary school

16 year old sister menarche at 14 yrs

Case 2

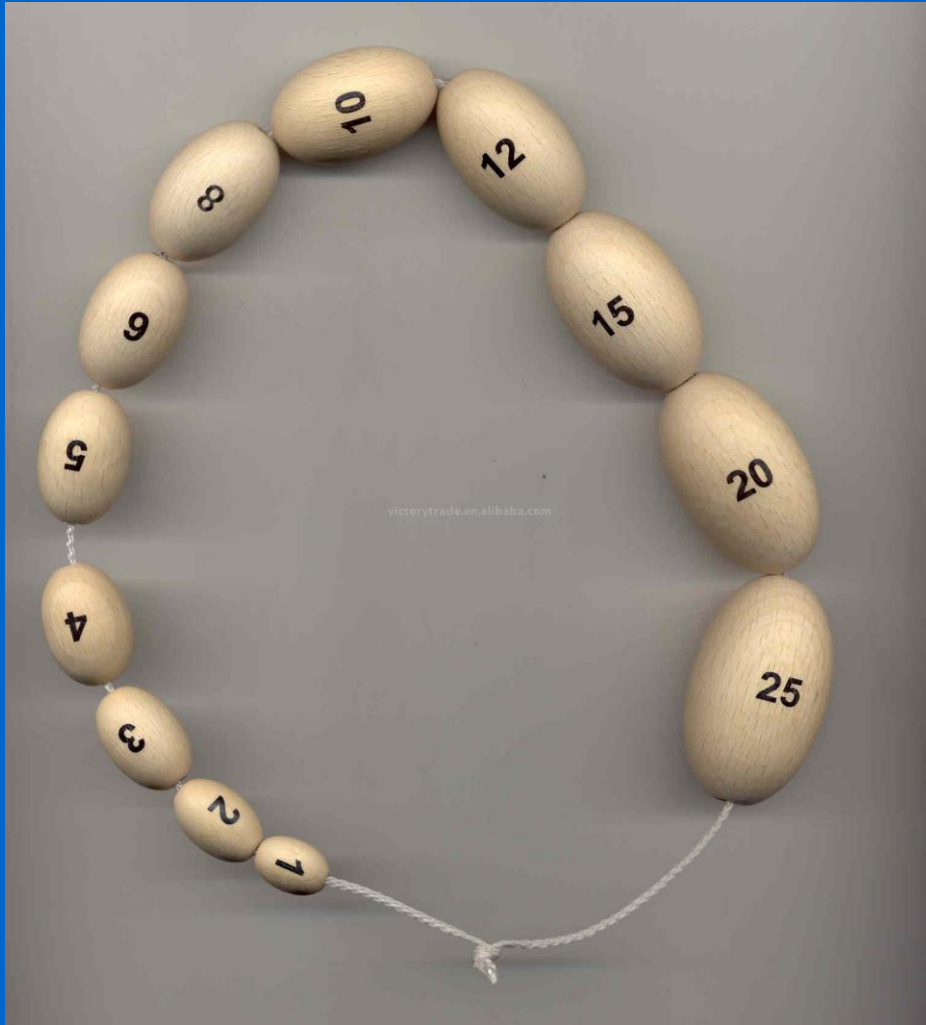
Examination normal

Testes 3 ml

Tanner stage 1 pubic hair

Orchidometer

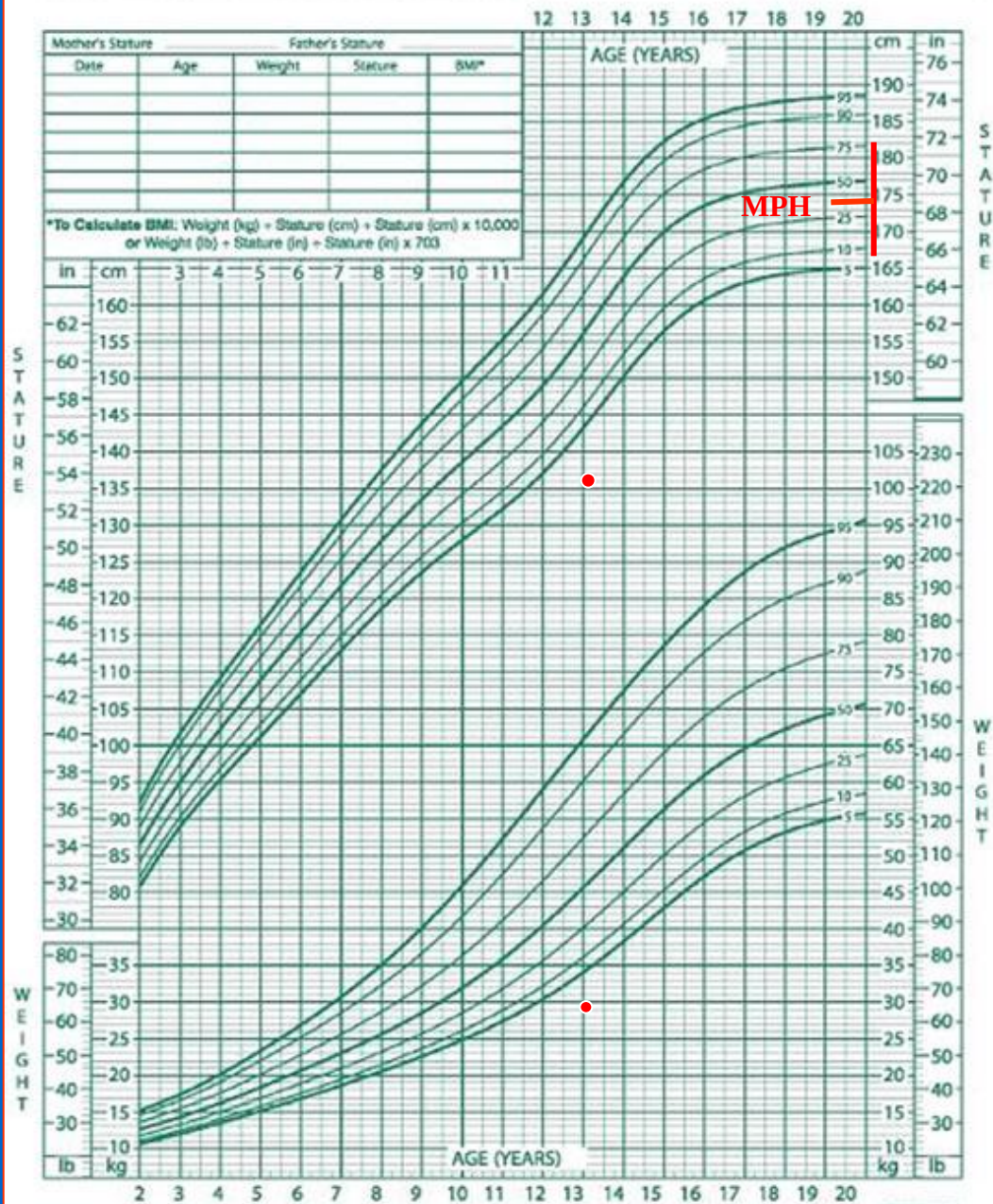
The paediatric endocrinologist's “rosary beads”



Case 3

Stature -for-age and Weight-for-age percentiles

RECORD #

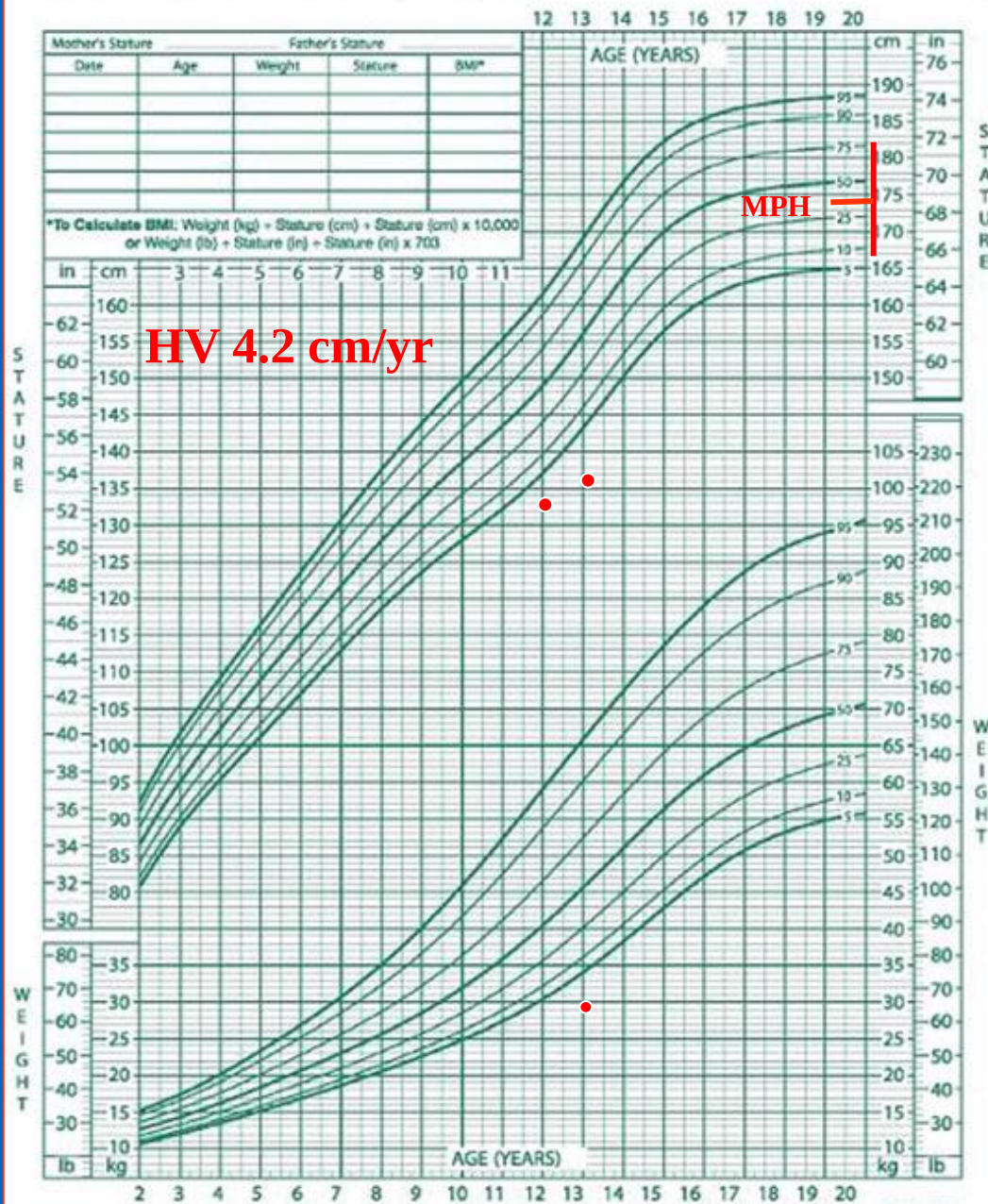


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Case 3

Stature -for-age and Weight-for-age percentiles

RECORD #



Case 3

Mild asthma treated with flixotide 200 mcg bid

No pregnancy illnesses/drugs

Birth weight 3,250 gm

Father 175 cm

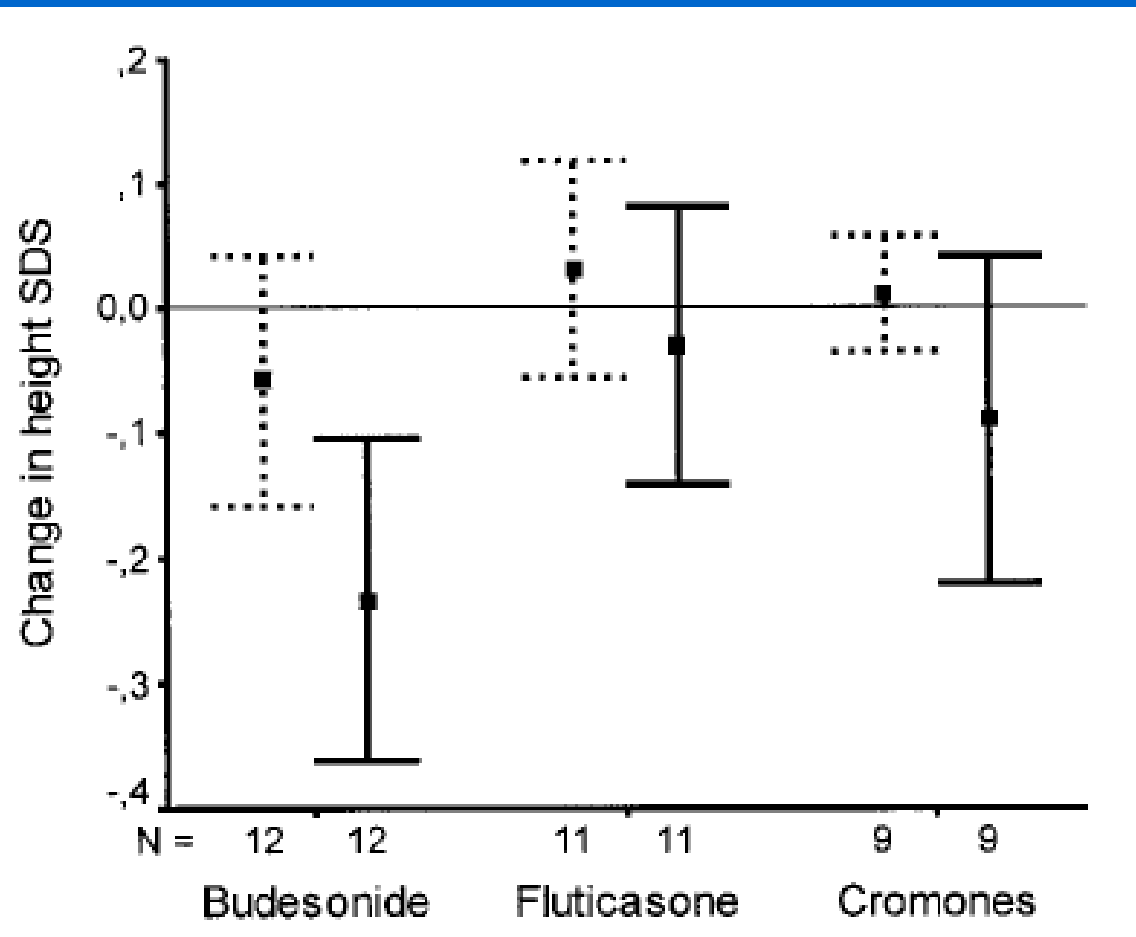
Mother 165 cm

Father accountant, mother lawyer

Youngest of three children

Is it likely that flixotide has affected his growth?

Fluticasone and growth suppression



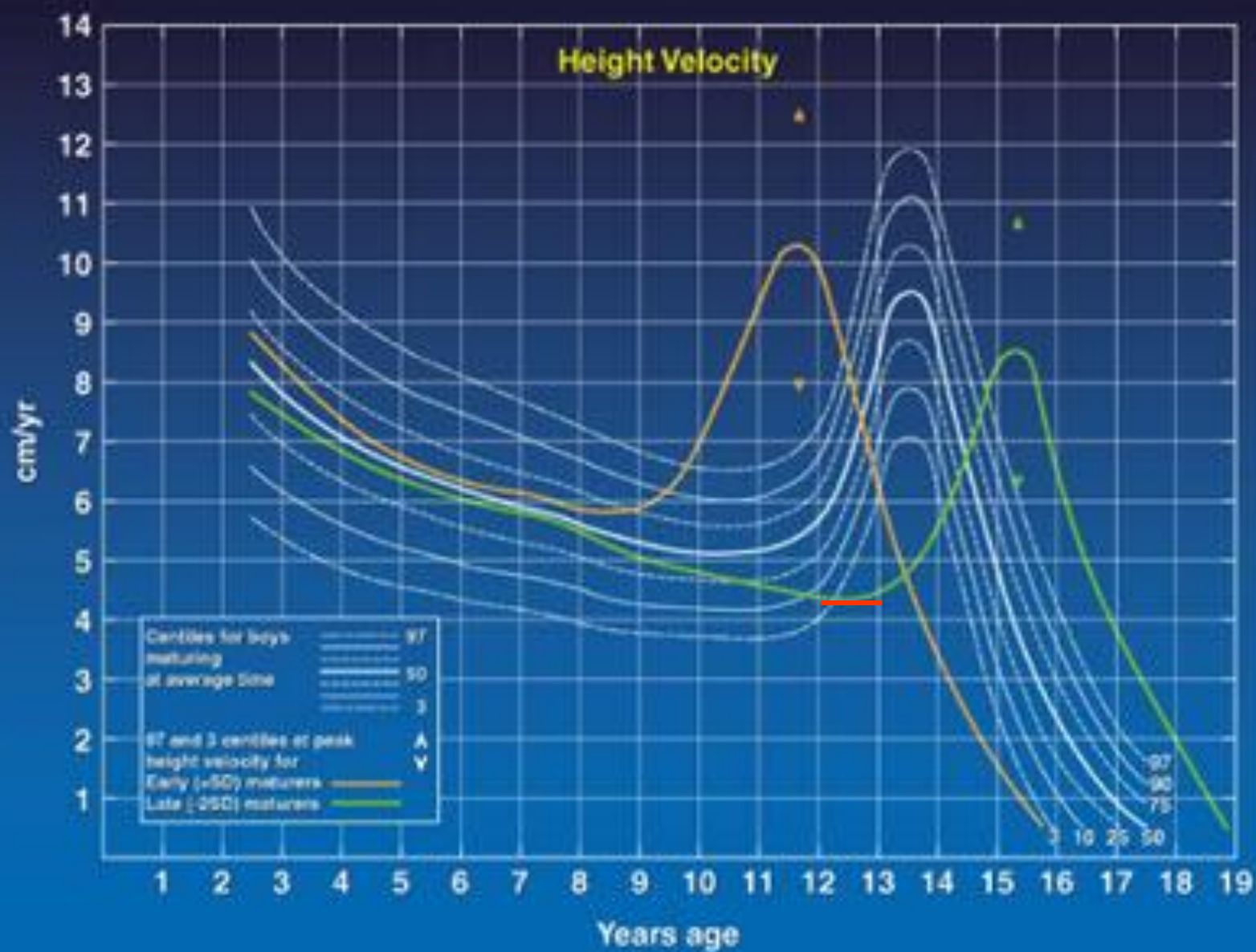
75 asthmatic children
9.5 (5.5-14.7) yrs
Randomised to
Budesonide 200→400 $\mu\text{g}/\text{d}$
Fluticasone 50→200 $\mu\text{g}/\text{d}$
Na cromoglycate

Fluticasone and growth and HPA axis suppression

Only seen with high doses of fluticasone:

Approx >600 $\mu\text{gm/day}$ younger children

Approx $>1,000$ $\mu\text{gm/day}$ older children



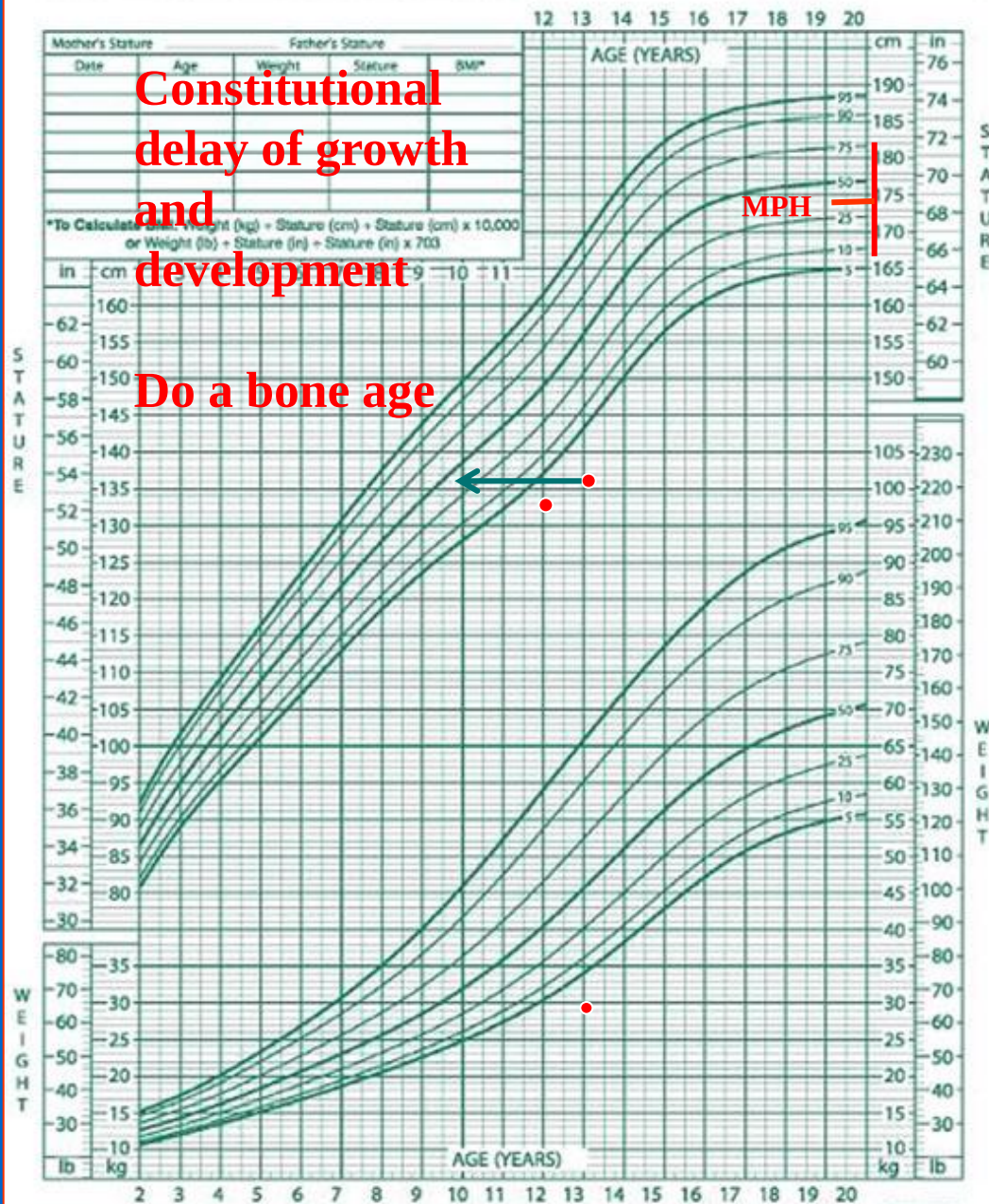
Case 3

What is the most likely diagnosis and how will you manage him now?

Case 2

Stature -for-age and Weight-for-age percentiles

RECORD #



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Case 3

How will you manage him now?

Is he distressed by his size and lack of pubertal development?

May benefit from low dose testosterone

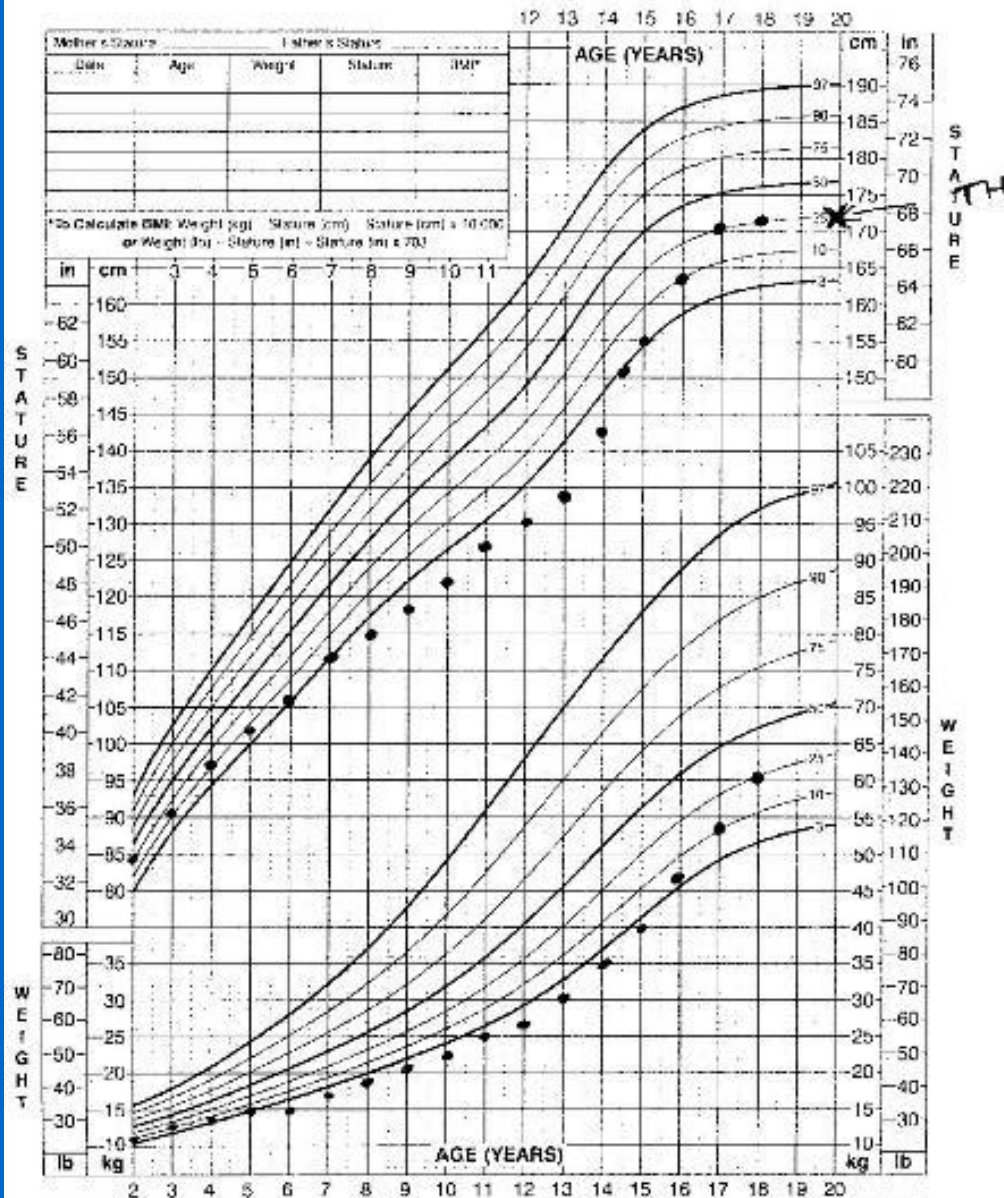
Testosterone undecionate (Panteston) 40 mg/day
x 6 mos or 50- 100 mg Testosterone esters
(Sustanon) IM monthly x 3-6 months

Will not adversely affect final height and will
give some pubertal development and height
acceleration

Case 2

2 to 20 years: Boys Stature-for-age and Weight-for-age percentiles

NAME _____
RECORD # _____



Published May 20, 2000 (modified 11/01/00)
©2000 CDC. Developed by the National Center for Health Statistics in collaboration with
the National Center for Chronic Disease Prevention and Health Promotion (2000)
<http://www.cdc.gov/growthcharts>

CDC
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Case 4

An 8 yr old boy presents with short stature.

Completely asymptomatic

No medications, no hospitalisations

No family history of note

Born at term, 3,400 gm

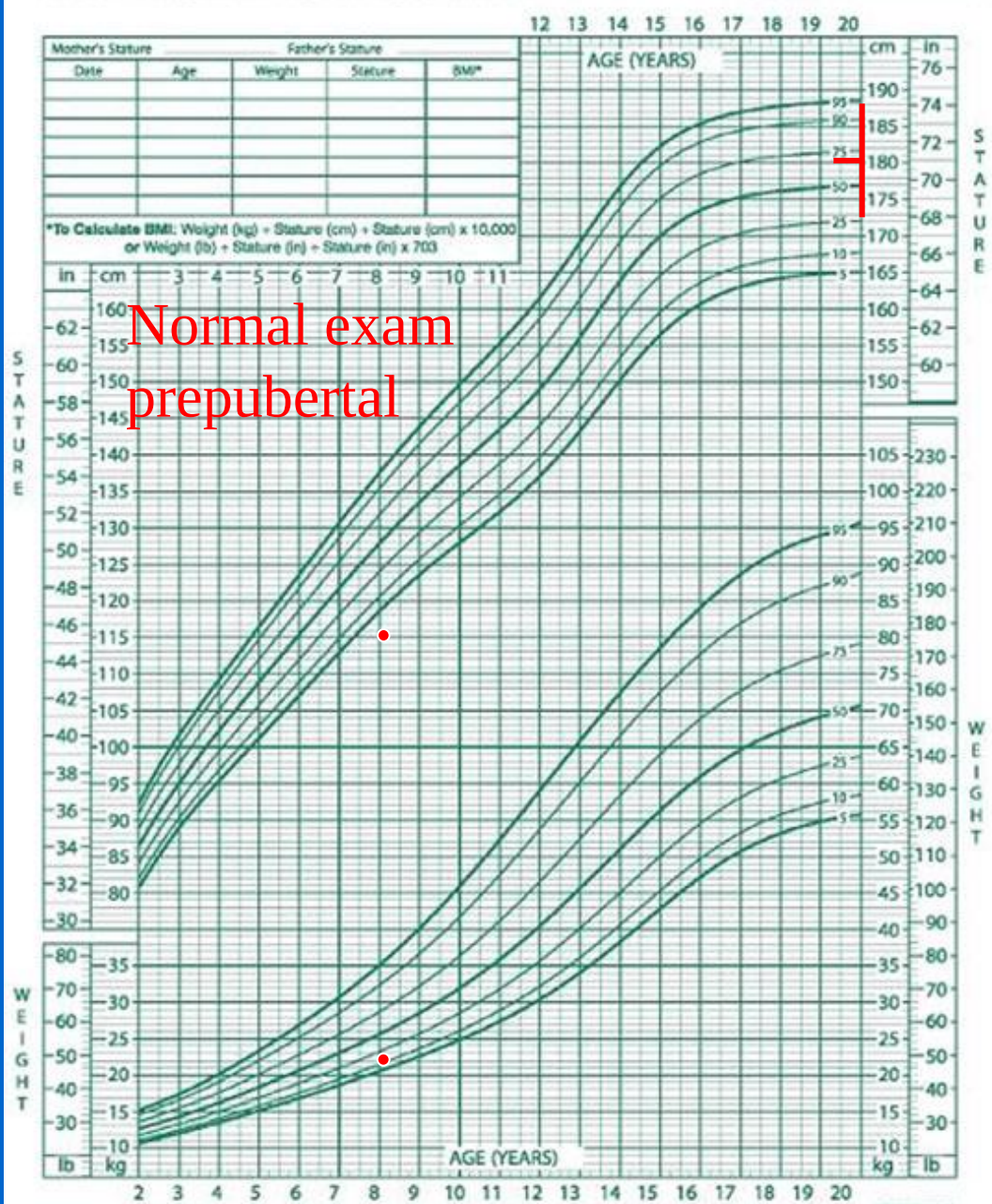
Father 182 cm, puberty history unknown

Mother 163 cm, menarche 12 yrs

Case 4

Stature-for-age and Weight-for-age percentiles

RECORD #

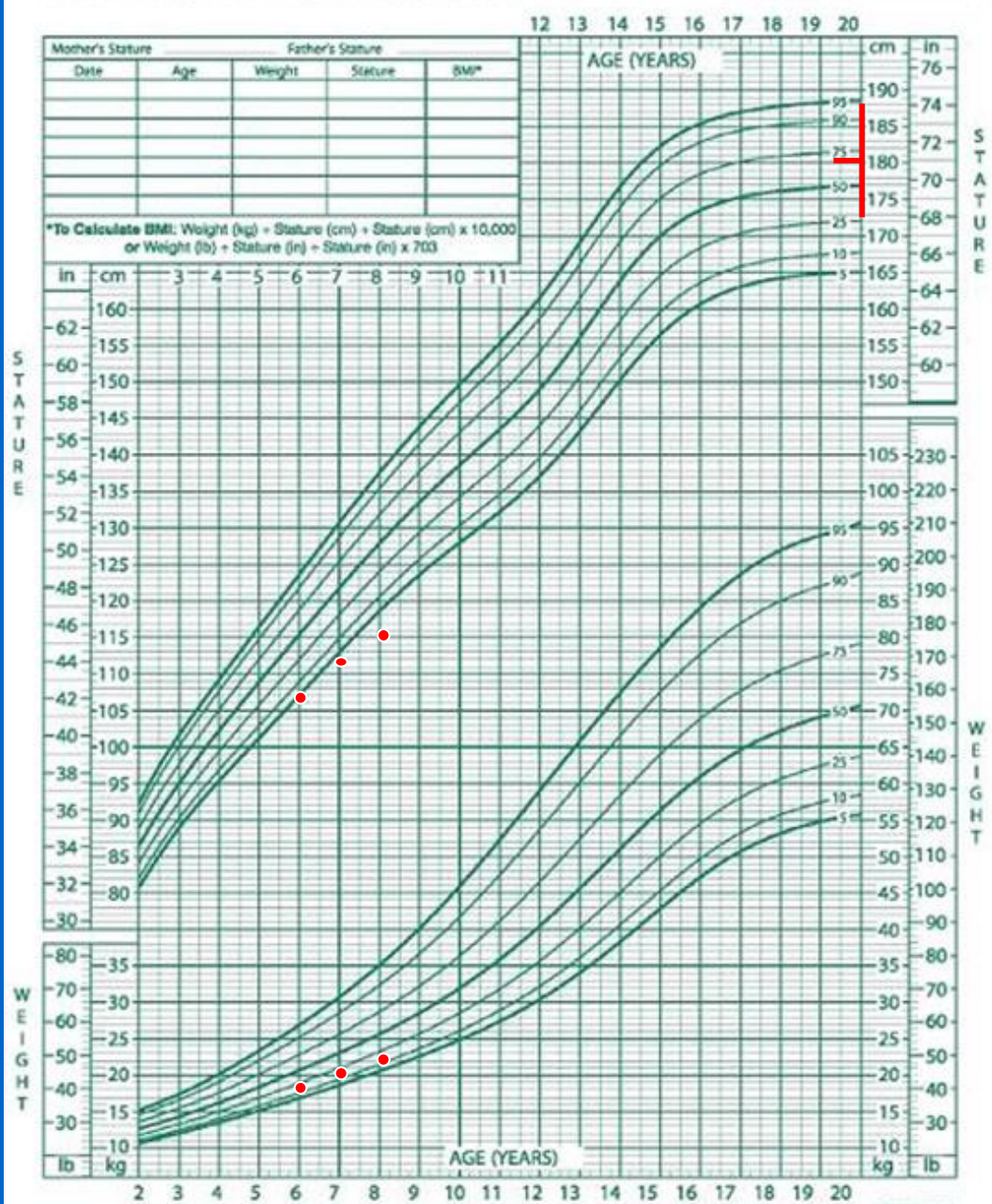


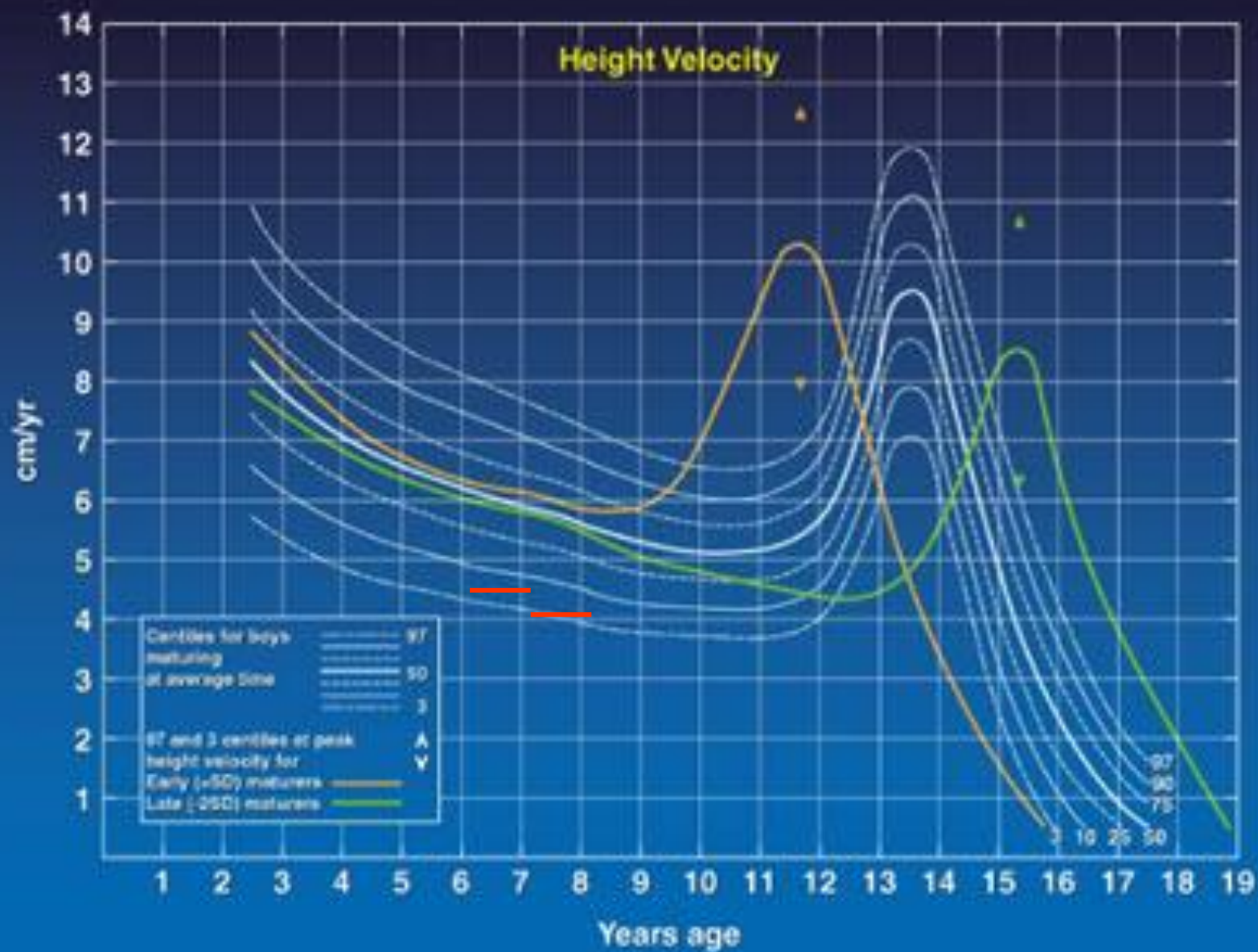
Normal exam
prepubertal

Case 4

Stature -for-age and Weight-for-age percentiles

RECORD #





Initial investigations

Hb 110 g/l MCV 62 WBC 6.4 (N differential)

ESR 4

Urea 2.3 mmol/l Na 141 mmol/l K 4.1 mmol/l

TFTs Normal

LFTs normal

Case 4

What is the most likely diagnosis?

Case 4

What is the most likely diagnosis?

Coeliac disease:

Chronic inflammatory bowel disease (occult)

Coeliac disease (CD)

In young children GI symptoms predominate

Older children often asymptomatic

50% of children with CD asymptomatic

Short stature without GI symptoms 2-8% CD

Csizmadia CGDS et al. Lancet 1999;353:813–14.

Voss LD et al. BMJ 1992;305:1400–2.

Ahmed ML et al. Arch Dis Child 1993;69:361–5.

Coeliac disease (CD)

Microcytosis

Low Haemoglobin

Low serum iron

Delayed bone age