

Short Stature

PPA clinical meeting 3.12.2008

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Case History

- **Personal Profile**
- **Name** **XX**
- **Age** **13 yrs**
- **Resident of Islamabad**

- **She has been coming to us since 5 yrs**

Case History

○ Presenting Complaints

- **Not gaining height** **5 yrs**
- **Not gaining weight** **5 yrs**

Case History

○ History of Presenting Complaints

- Parents brought their daughter to OPD with C/O

not gaining Height & Weight as compared to rest of their children.

- There was H/O

Anorexia, nausea, constipation and easy fatigue ability.

Case History

- There was no concurrent H/O
Loose motions, persistent cough,
palpitations etc.

Also no H/O

joint pain, vertigo , dizziness

or

Trauma, surgery , cranial irradiation

Case History

- **Birth History:**

 - SVD in CDA Hospital.

- No prenatal exposure and illness or evidence of IUGR,

- At birth average Height and weight

- No perinatal problems i.e.

 - Prolonged jaundice,

 - Hypoglycemia,

 - Puffy extremities.

Case History

o Nutritional History:

Breast fed for 6 months.

Family food is not taken satisfactorily.

o Developmental History:

Sitting at 6 months, Walking at 1 yr

No developmental delay is recorded.

o Immunization History :

EPI schedule

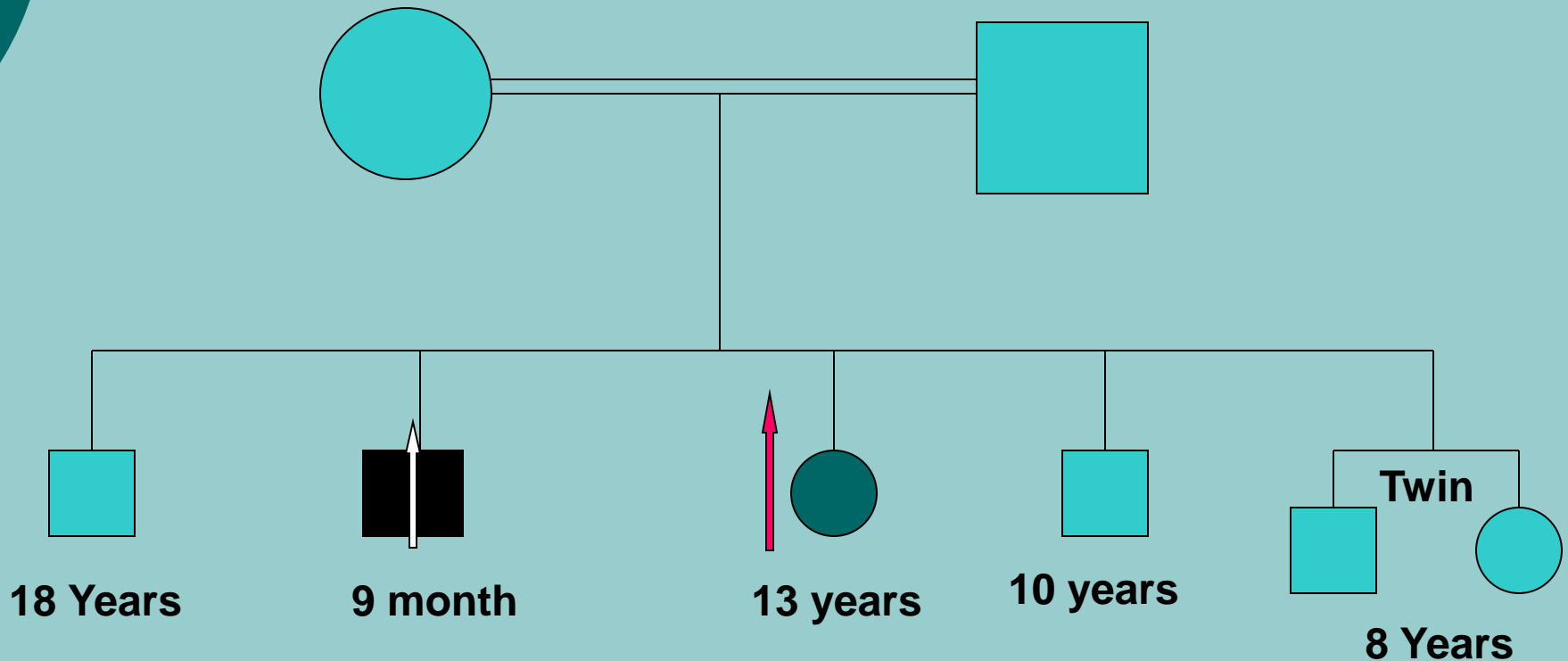
BCG scar present

Family History:

Parents are first cousins with average height & built.

3 brothers & 1 sister are of normal height & built

1 Brother died at the age of 9 months due to gastroenteritis.



Case History

- **Family History:**

 - mother is carrier of thalasseamia trait

 - No family H/O

 - Tuberculosis, Fits, Diabetes mellitus,
Mental retardation, Short stature.

- **Socio economic History:**

 - Lower middle class.

 - Healthy environment including adequate sleep,
exercise, psychosocial factors are also favorable for
growth & development.

General Physical Examination

- A young thin built girl with pallor,
no dymorphism or disproportions
looking short for her age
- Anthropometric measurements
< 3rd centile
- Vital signs with in normal range.
- No sign of malnutrition or other
remarkable physical findings.

Systemic Examination

- GIT:

Abdomen soft, non tender,
no visceromegaly /ascites

- CVS :

Apex beat 4th ICS medial to MCL
S1+S2+0

- Resp S :

No significant finding

Systemic Examination

- CNS :

 - Intact higher mental functions & speech

 - Motor & Sensory systems &

 - Cranial nerves intact

 - Cerebellar signs absent.

- SMR :

 - No sign of puberty

Stature measurements 8 yrs

wt : 14 kg
Ht : 102 cm } < 3rd centile
Weight age : 3 yrs
Height age : 4 yrs

Ratio : U/ L 52/50 cm = 1.04 : 1

Arm span : 104 cm

Target Height: 150.5cm

Mid parental Height : 153 cm

Management Plan

Initially for 6 months we managed her with Dietary advice and multivitamins & Iron but no significant improvement in height & weight.

- Detailed evaluation was done by doing certain tests.
- **Differentials for short stature were**
 - *Constitutional delay,
 - *Familial short stature &
 - *Idiopathic short stature
 - *Endocrinal causes
- Growth measurements were plotted.

Investigations

- **Blood CP:**

WBC	$9.8 \times 10^3 / \mu\text{l}$
Hb	9.2 G /dl
Platelets	$358 \times 10^3 / \mu\text{l}$

- **Hb electrophoresis :**
Beta thalassemia trait

- **RBS :** **98 mg %**

Radiological Aid

- Bone Age at 9 yrs
more than 7 & less than 9 yrs
- Chest X ray : Clear lung fields ,
normal cardiac shadow
- Abdominal USS : Normal profile

Chest X Ray



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Hina
26.09.07.
30
2/11/0



Investigations

- Stool RE
- Urine RE
- RFT
- LFT
- TFT

WNL

- Chromosomal Karyotyping:
Report awaited
- Cealiac profile was negative.

Growth Hormone levels

2005

Basal level : 1.23 m IU /l (< 14 n IU /l)

GH after L dopa: 4.76 n IU/ l (NV: 0)

2007

Basal level : 0.26 m IU /l (< 14 n IU /l)

GH after L dopa: 9.7 n IU/ l (NV: 0)

Suboptimal response to GH stimulation test.
Insulin Tolerance test was advised.

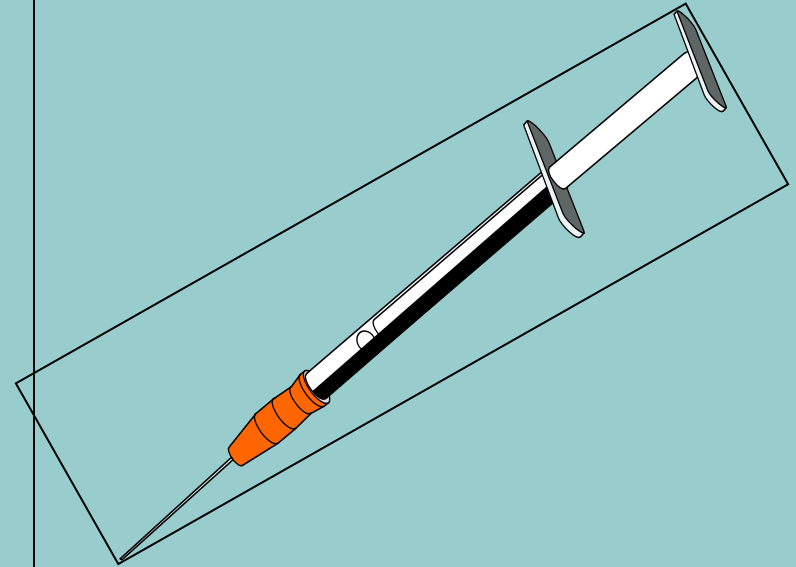
Growth Hormone levels & Insulin Tolerance Test

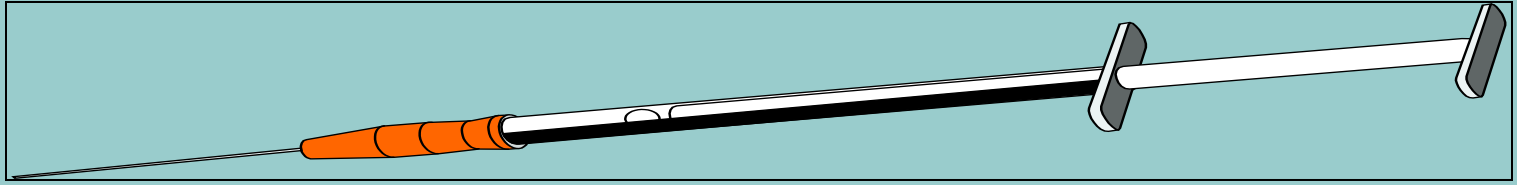
Plasma glucose basal	4.4	(3.3—6.1 m mol/L)
Plasma glucose (at induction)	1.7	
Glucose 30 min	4.9	
60 min	5.0	
GH basal level	0.19	(<14m IU/L)
GH (at induction)	3.8	
GH 30 min	3.0	
GH 60 min	0.52	

Inadequate response to insulin tolerance test

Final Diagnosis

- Short stature due to isolated Growth Hormone Deficiency.





- Replacement Therapy was started in April 2008 after consultation & evaluation by Dr Gulbeen Shahid (endocrinologist PIMS)
- Eutropin 4iu SC injections
3 times /wk at night.



○ Age yrs	Height	Weight
○ 8	102 cm	14 kg
○ 8.5	103 cm	15 kg
○ 9	106 cm	15 kg
○ 9.5	107 cm	15 kg
○ 10	111 cm	16 kg
○ 10.5	115 cm	16 kg
○ 11	120 cm	17 kg
○ 11.5	120 cm	18 kg
○ 12	121 cm	20 kg
○ 12.5	122 cm	21 kg
○ 13+	130 cm	23 kg

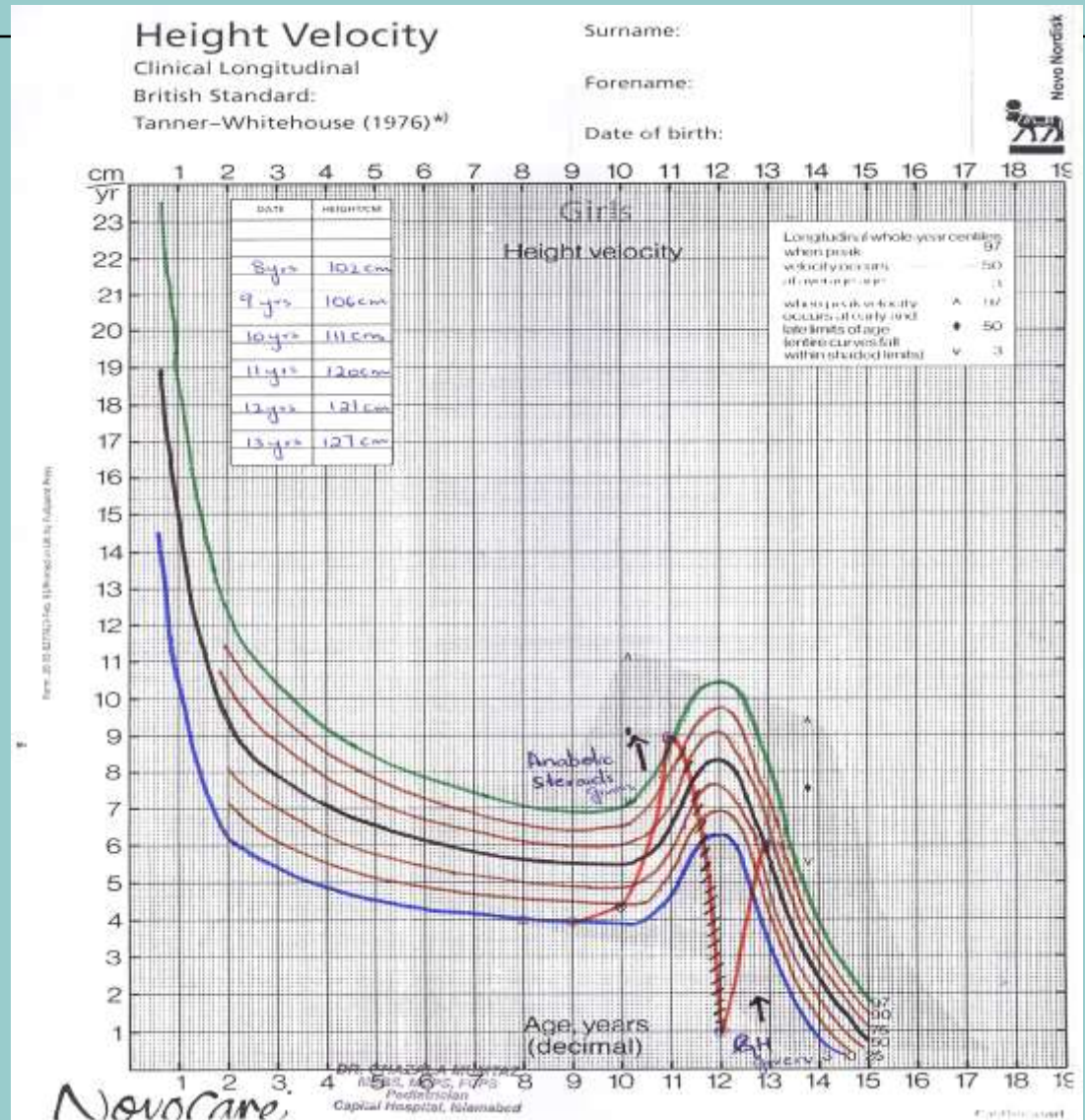
Effects of Therapy

- Gain in Height 8 cm
- Gain in Weight 2 Kg

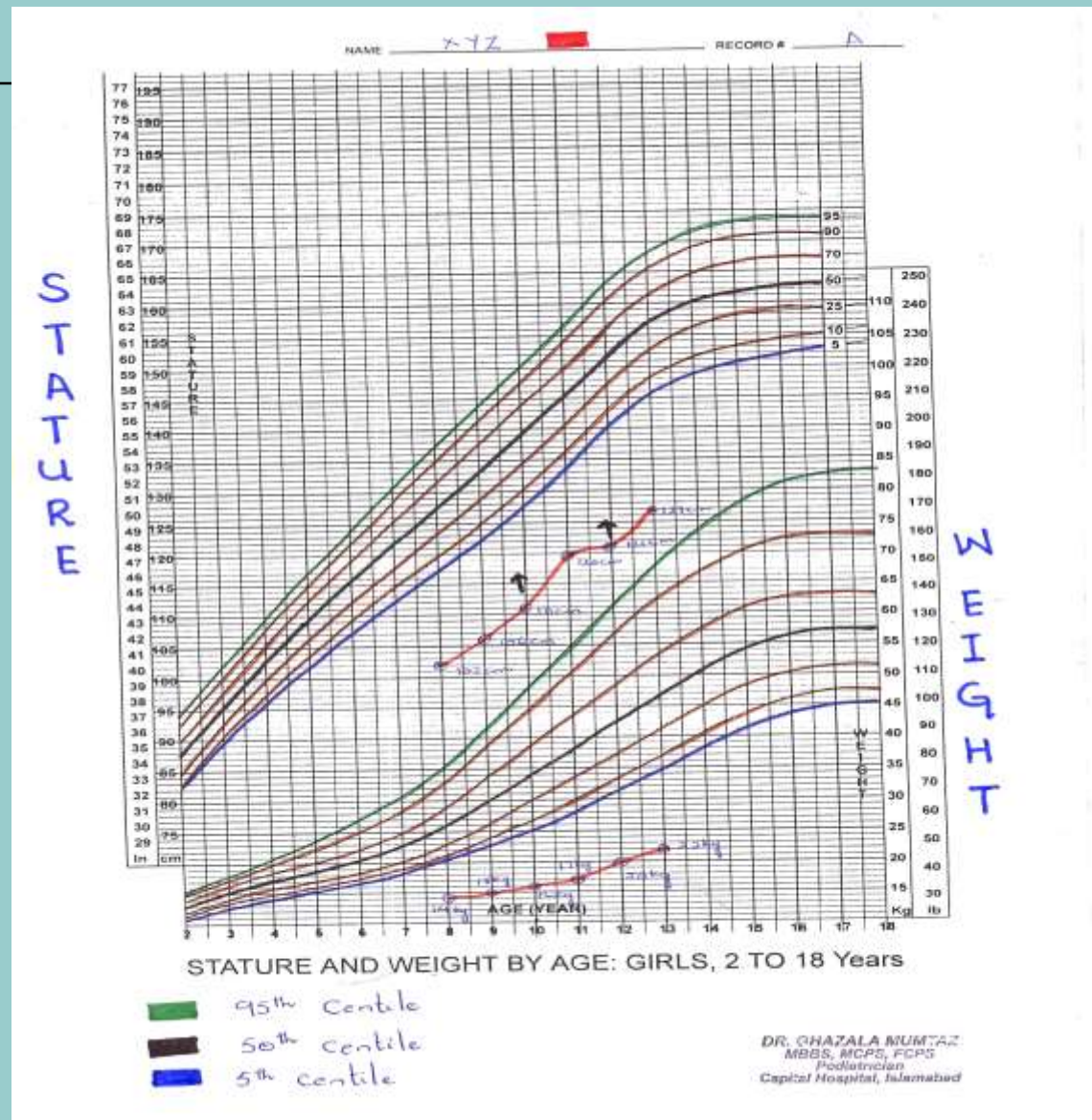
Ratio U/ L :

$$62.5/64.5 \text{ cm} = 0.96 : 1$$

.Height Velocity



Height & weight.



Current Age 13 yrs

- Weight 23 kg = weight age 8 yrs
- Height 130 cm = Height age 9 yrs

- Expected weight = 46 kg
- Expected Height = 157 cm

- Target Height = 150.5cm

Case Summary

- The parents of a healthy 8 yr old girl are concerned that she is the shortest among her siblings & class mates.
- Her height & growth curves as shown already.
- A thorough history reveals that she was a full term infant, has had no significant medical problems, she is developmentally appropriate.
- Other than being small for her age, no abnormalities are noted on physical examination.
- Careful measurements of her upper & lower body segments are normal for age.
- Her parents of average height had normal pubertal ages. Father : 14yrs & Mother : 16yrs

How do you define short stature in a child?

- **DEFINITION**

- Short stature is defined as height

 - < 2 to 2.5 SD

 - (less than 2 to 2.5 standard deviations)

 - below the mean for age and gender

Calculate mid-parent height:

○ **Boys**

Mid parental height = mother's height + father's height
/2 + 7cm

Target centile range = mid parental height +/- 10cm

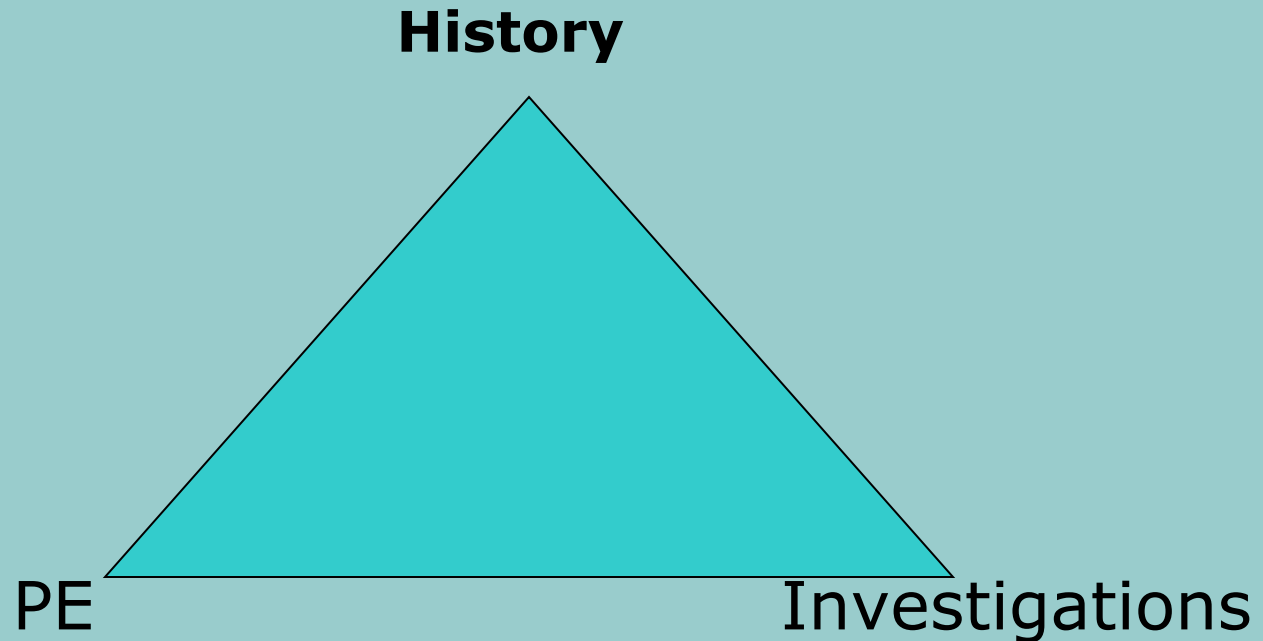
○ **Girls**

Mid parental height = mother's height + father's height
/2 - 7cm

Target centile range = mid parental height +/- 8.5cm

Evaluation of Short Stature

- **Triad of**



*Evaluation of Short Stature

*EXAMINATION

Respiratory System

Chest deformities

Signs of chronic lung disease e.g.
cystic fibrosis, asthma

Cardiovascular System

Signs of congenital heart disease

Hypertension

Signs of cardiac failure

Evaluation of Short Stature

*EXAMINATION

Abdomen

Hepatomegaly

Splenomegaly

Masses

Ascites

Renal

Urine output

Evaluation of Short Stature

*EXAMINATION

CNS

Visual acuity and visual fields

Nystagmus

Signs of hydrocephalus

Focal signs

*INVESTIGATIONS

- ❑ FBC
 - Aneamia
 - Leucocytosis
 - Leucopaenia
 - Thrombocytopaenia
- ❑ ESR, CRP
- ❑ Electrolytes and liver enzymes
- ❑ Carotene, folate, prothrombin time
- ❑ Celiac panel

INVESTIGATIONS

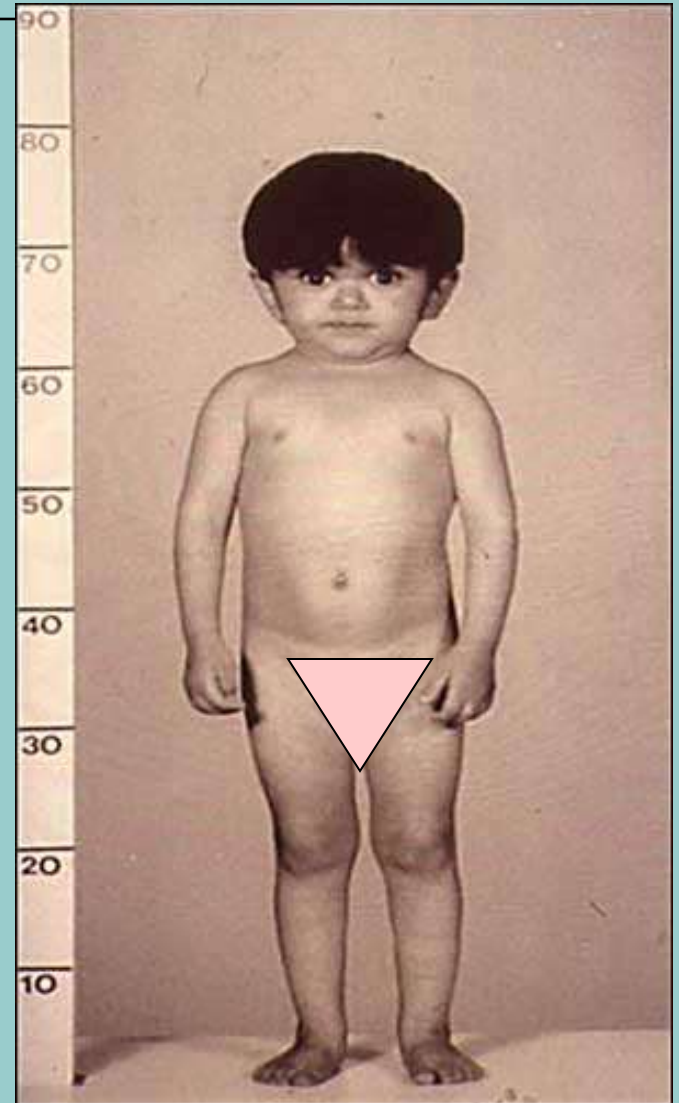
- Urinalysis and pH
- Karyotype
- Cranial imaging- MRI
- Bone age
- TFT
- Prolactin
- IGF 1, IGF BP3
- Growth hormone stimulation tests

MANAGEMENT

- OPTIMISE TREATMENT OF CHRONIC DISEASES
- ENSURE GOOD NUTRITION AND NOT OVERNUTRITION
- APPROPRIATE THERAPY FOR TUMOURS
- REPLACEMENT THERAPY FOR DEFICIENCY SYNDROMES

Features of Congenital GHD

- Normal BW&BH.
- Unexplained hypoglycemia.
- Prolonged hyperbilirubinaemia.
- Clinical appearance.



Features Suggestive of GHD

- **Short stature**
- **Poor growth velocity**
- **Delayed bone maturation**
- **Increased subcutaneous fat**
- **Dentition is delayed**
- **High pitched voice**
- **Age of pubertal onset**

**Diagnosis*

- History.
- Physical exam.
- Auxologic criteria.
 - Height.
 - Mid parental ht.
 - Growth velocity.
 - Bone age.
 - Tanner staging.

**Investigations.*

- **GH profiles.**
 - Spontaneous GH secretion.
 - Urinary GH excretion.
- **Pharmacological tests.**
 - GH provocation test.
- **Serum markers of GH secretion.**
 - IGF-1 and IGFBP-2,3 levels.
- **Radiological investigations.**
- **Other profiles.**

**Treatment*

○ Preparations

Humatrope Eli Lilly 4iu/1.33mg

Eutropin 4 iU LG Life sciences Korea

○ Dose

(3iu=1.0mg)

0.18 – 0.3mg/kg/wk

0.025 – 0.035/kg/day

○ Response depends

Age at the start of treatment

Severity of GHD

Duration of the disease

Genetic potential (parental height)

Dose and frequency of administration of rhGH

**Treatment...*

- **Duration.**
- **When to stop Rx.**
- **Contraindication.**
- **Follow up.**
 - Growth parameters.**
 - Bone age.**
 - Tanner staging.**
 - Side effects of therapy.**

**Side Effects of GH Treatment*

- **Pseudotumor cerebri.**
- **Salt and water retention.**
- **Hypothyroidism.**
- **Slipped capital femoral epiphysis.**
- **Worsening of scoliosis.**
- **Acute pancreatitis.**
- **Possible glucose intolerance and hyperinsulinism.**
- **Leukemia.**

**Other Interventional Therapy*

- Anabolic steroids.
- IGF-1.
- GHRH.
- Delaying puberty.
- Surgery and limb lengthening.

**Other Indication For GH*

- Turner syndrome.
- End stage renal failure.
- Idiopathic short stature.
- IUGR.
- Prader-willi syndrome.

Prognosis

- Prognosis is determined by response to Growth hormone replacement therapy and is generally favorable

Short stature

Thank You

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