MAGIC BOX ON DWARF’S HEAD

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CASE

- 4 Yr 5 month
- Male child
- DOB-11.06.2008
- First born to NCM
- Developmentally normal
- Orissa
- Reliable history
Presenting complaints

- Not gaining adequate height compared to his peer groups
Since when and how?

- Noticed by around **2 years** of age.
- In family functions and when playing with other children.
- Also in group photos of preKG, he is found to be shorter than others.

Consulted nearby hospital, reassured and advised to observe.
Associated problems?

- No h/o any **chronic illnesses**—cardiac, pulmonary or renal, git
- No h/o **limb deformities**, abnormal gait, fractures, trauma
- No h/s/o **hypothyroidism**—constipation, lethargy, feeding difficulty, MR, cold intolerance
- No h/o **malabsorption**—large, bulky, frothy stools
- Any h/s/o **cushings**—
Contd....

- No h/o drug intake - nicotine, hydantoin, warfarin, steroids
- No h/o raised ICT
- No h/o visual or hearing disturbances
**Birth History**

- **Antenatal h/o** - uneventful (Any h/o maternal infections antenatally—TORCH)(drug intake)

- **Natal** - LSCS at 38 weeks.(non progressive labour).
  
  B WT—2.75Kg.length-50 cm(records)

- **Post natal** — hyperbilirubinemia+ PT given for 2 days.
FAMILY HISTORY

- No family h/o short stature or constitutional delay
- Father—165cm
- Mother—159cm
- MPH—166cm (>50th centile)

- 5yrs
- 5months

abortion
Well immunised till date. Optimal vaccines, typhoid only was given (at 4 months length-**62cm**)

- Developmentally normal child
## DIET HISTORY

<table>
<thead>
<tr>
<th></th>
<th>OBSERVED</th>
<th>EXPECTED</th>
<th>DEFICIT</th>
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<tbody>
<tr>
<td>CALORIES</td>
<td>1300</td>
<td>1230</td>
<td>70</td>
</tr>
<tr>
<td>PROTEIN</td>
<td>25</td>
<td>26</td>
<td>nil</td>
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</tbody>
</table>
- Father is railway employee
- Mother is house wife
- Kuppusamy classification grade 3 (lower middle class)
PHYSICAL EXAMINATION
Concious, oriented, afebrile, playful
No pallor, icterus/cyanosis/clubbing.
No lymphadenopathy/edema.

**Vitals:**
Temp - 37.0 deg. celsius.
PR - 96/min.
RR - 28/min.
BP - 110/70 mm Hg
HEAD TO FOOT EXAMINATION

- Shape of head - normal, no dysmorphism
- Face - puffy, **doll like**
- No midline defects
- Hair - normal
- Teeth - 20 in no. No caries
- No signs of vitamin def
- Limbs - normal
- No petechial spots
- Abdomen - normal
- Bilateral testis descended
- Stretched penile length -1.9cm
- Tanner SMR staging -- preadolescent
ANTHROPOMETRY

- Weight—14.4 kg......15\textsuperscript{th} centile
- Height---87cm..........\textless 3\textsuperscript{rd} centile
- HC--49cm..................15\textsuperscript{th} centile
- MAC—14.5cm
- US /LS ratio—1.2:1
- Arm span—86cm
- Weight for height......97\textsuperscript{th} centile
Length/height-for-age BOYS
Birth to 5 years (percentiles)

WHO Child Growth Standards
Systems examination

- CVS: S1 S2 heard, no murmurs.
- RS: NVBS, BAE Equal. WOB - Normal.
- P/A: Soft. No organomegaly/FF.
- CNS:
  - Higher functions/cranial nerves—normal.
  - Spinomotor system: normal
  - Gait: Normal

**Visual assessment:** Acuity, field of vision normal
4 yr 5 month old developmentally normal male child, first born to NCM presented with PROPORTIONATE SHORT STATURE, with MICROPENIS with normal Wt for Ht without any significant family history, and with out any features of nutritional deficiency or limb deformity ......
Provisional diagnosis

- PROPORTIONATE SHORT STATURE with MICROPENIS most probably endocrine etiology
EVALUATION
Investigations

- Baseline investigations - Normal
- Renal function tests - Normal
- Liver function tests - Normal
- Serum Calcium - 9.1 mg%
- Serum Phosphorus - 4.7 mg%
- X ray wrist showed **Bone Age <3 years.**
- **Thyroid Function Test:** Normal
  - T3 - 2 nmol/L
  - Free T4 - 15.3 pmol/L
  - TSH - 6.2 microunits/ml

- **Estimation of Growth Hormone levels (Clonidine Stimulation Test)**
  - Basal - 0.04 ng/ml
  - 30mins - 0.06ng/ml
  - 60 - 0.06ng/ml
  - 90 - 0.058ng/ml
  - 120 - 0.077ng/ml
  - 150 - 0.048ng/ml (GH Deficiency)
- **USG Abd-- normal**
- **IGF- I levels** – <25 (49-289ng/ml)
- **IGF BP3 levels** – 0.5 (0.9-4.3U)
- **Serum Cortisol** – 8 mic gm/dl (4-32)
- **Sr. Prolactin** – 7.3 ngm/ml (3.34 – 26.72)
- **FSH** – 2.9 mIU/L (1.79 -5.12)
- **LH** – 2.4 mIU/L (1.20 – 12.86)
- **Testosterone**---8.9ng/dl (2-20ng/dl)
Prominent subarachnoid space with herniation of CSF into the sella tarsica, compressing pituitary gland  S/O

EMPTY SELLA SYNDROME
- Growth hormone therapy started at 0.3 mg/kg /wk as daily injections s/c.
- Parents are counseled
AFTER 6 months....

- height ..... 93cm (inc by 6 cm)
- weight......16.6cm
- stretched penile length...2.2cm (0.3cm)
DEFINITION

Height Below 3\textsuperscript{rd} centile or more than 2 standard deviations below the median height for that age and sex and according to the population studied
STEPS IN EVALUATION

STEP I:
Is the child really short?

STEP II:
If short, is it proportionate or disproportionate?

STEP III:
SMR

STEP IV:
Determining Aetology

STEP V:
Investigative workup
APPROACH TO SHORT STATURE

Ht less than 3\textsuperscript{rd} centile

NO

Reassurance
Assess Growth velocity

YES

Is the Ht within MPH

ASSESS BONE AGE

NO

Elicit history to r/o
Systemic diseases, Malnutrition, IUGR Dysmorphic/ Chromosomal syndromes, Hormone deficiency

BA=CA>HA

Familial Short Stature

Assess Growth Velocity over 6 months

NO

BA=HA<CA

Constitutional Growth Delay
Arm Span, Height & Upper-Lower Segment Measurements

PROPORTIONATE

GROWTH VELOCITY

NORMAL

- CONSTITUTIONAL DELAY IN GROWTH
- FAMILIAL SHORT STATURE
Chronic Diseases
- Renal
- GIT
- Cardiac
- Pulmonary
- Metabolic
- Ch. Infections
- Immunodeficiency

Endocrinal Causes
- Hypothyroidism
- GH Deficiency
- GH Resistance
- Cortisol Excess
- Cushing’s
- Exogenous Steroids
Arm Span, Height & Upper-Lower Segment Measurements

Disproportionate?

Yes

- Rickets of Various Causes
- Cretinism
- Skeletal Dysplasias
- Multiple Dysostosis

No
At What Age Has the Child Presented With Short Stature?

REMEMBER - **I C P Model** of Growth!
- **Infancy** - Likely to be **Nutritional**
- **Childhood** - **Hypothyroidism** and **Growth Hormone Deficiency**
- **Puberty** - Disorders of **Puberty***(sex steroids)*
Bone age is not delayed & corresponds to the chronological age

Growth Velocity is normal proceeding along the 10th centile

BA = CA > HA
Constitutional delay in growth

\[ BA = HA < CA \]
Bone age is delayed;

Growth Velocity is normal proceeding along the fifth to tenth percentile

This curve shows that a patient "fell off the curve," and had a delayed bone age.

Thyroid replacement "jump starts" growth.
Empty sella refers to the radiological appearance of an enlarged or deformed sella tarcica that is partially or completely filled with cerebrospinal fluid.

Busch used the term “empty sella” to describe this condition.
The term empty sella, is a misnomer as the sella is not completely empty.

Pituitary is always present both anatomically and functionally, though often it is displaced downwards and compressed by CSF pressure, results in abnormal pituitary function.
An **incomplete sellar diaphragm** is an essential pre-requisite for the development of the empty sella.

1. **Congenital deficiency** of the diaphragma sellae
2. **Suprasellar** promoting factors: raised ICT
3. **Pituitary** promoting factors: any **reduction** in the size of pituitary gland

**Physiological involution:**
- pregnancy/menopause
- Replacement of the deficient hormone results in feedback suppression
Pathological involution: (Sheehan’s syndrome) pituitary infarction (vascular d/s, diabetes), head injury, meningitis, cavernous sinus thrombosis

4. Rupture of an intrasellar or parasellar cyst:
Secondary empty sella

Associated with an iatrogenic event such as surgery, radiation, or both

Non-iatrogenic disease such as infarction and infection of the pituitary gland.
Clinical features

- Most persons are asymptomatic and the detection of this abnormality may be incidental.
- Visual abnormalities are infrequent. (more in 2ndry type)
- Children with an empty sella most commonly have GH deficiency, although other pituitary hormone dysfunction may occur.
- Rarely, associated with hormone excess possibly due to coexisting micro-adenoma within the compressed gland.
In children with isolated GH deficiency or multiple pituitary deficiencies, empty sella was observed in 48%.
Diagnosis

- The lateral radiograph of the skull:
  - normal sized sella or enlarged
  - (maintenance of the "closed" configuration)
  - (intrasellar masses or neoplasm "open" configuration)
- Computerised tomography scans
- Magnetic resonance imaging (MRI)
Hormonal profile

(serum thyrotropin and T4, corticotropin and cortisol, LH and testosterone, basal and stimulated growth hormone and prolactin level.)

(mild hyper-prolactinaemia with or without galactorrhoea occurs in approximately 15% of patients)
Complications

- visual field defects
- Cerebrospinal fluid rhinorrhea
- meningitis.
- pituitary dysfunction
Treatment of empty sella

- Asymptomatic and incidentally detected, it requires no specific treatment
- **endocrine dysfunction**—replacement therapy
- **CSF rhinorrhea** may require surgical correction.
- **visual field defects**: needs to be corrected by chiasmopexy
To emphasize on the importance of physical examination esp genetalia in case of short stature to get a clue to diagnosis.

to emphasis the importance of recording anthropometry and plotting on graph during all immunisation visits.
REFERENCE

- Nelson text book of pediatrics