Name that Growth Issue: A Critical Approach to Diagnosing that “Common” Growth Problem

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Disclosure of Conflict of Interest

I have no actual or potential conflict of interest in relation to this program/presentation.

Objectives

- By the end of this session, participants should be able to discuss the varying approaches and differential diagnoses in a short stature evaluation.
- By the end of the session, participants should be able to apply knowledge to future growth evaluations at their home institution that will lead to improved diagnoses for that “routine” growth evaluation.
The Usual Short Stature Evaluation

- Review of Systems
- Physical Exam
- Laboratory studies
- Imaging

History and Physical Examination

- Birth History - Small for Gestational Age, Intrauterine Growth Retardation
- General History - Chronic Illness
- Family History - Genetic, Psychosocial
- Physical Examination - Proportions, Abnormalities
- Growth Chart - Growth Velocity, Change in Growth Pattern

Normal growth rates during childhood

- Girls’ peak growth rate: 11.5 yrs
- Boys’ peak growth rate: 13.5 yrs

National Center for Health Statistics
Growth evaluation

- Laboratory studies
  - Endocrine: IGFBP-3, IGF-1, TSH, Cortisol?; GH stimulation testing?
  - Non endocrine: CBC, CMP, Inflammatory markers (ESR or CRP), Celiac panel
- Radiology studies
  - Bone age
  - MRI of pituitary gland?

Differential Diagnosis

- Normal variants
  - CDGP
  - FSH
- Pathologic Conditions
  - Non-Endocrine
    - Chromosomal Abnormalities
    - Congenital anomalies
    - Disorders of bone formation
    - Chronic systemic disease (CKD, Celiac disease)
    - Nutritional Disorders
    - Infection
    - Inflammatory or Connective tissue disease
    - NCGA
- Endocrine
  - Hypothyroidism
  - GHD
  - Hx of CPP
  - Cushing disease

Case #1

- Initially presented at age 10 years 8 months with concerns of growth deceleration
- ROS negative
- Family Hx
  - MPH 5’7”
  - No endocrine issues
  - No known autoimmune issues
Case #1 - Growth Chart

Growth deceleration began at age 5 years from 75th%ile. By age 9, 3rd%ile.

Case #1 - Physical Exam

- Presenting photo
- Hypertrichosis on back
- Wide neck
- Prepubertal

 Evaluation at initial presentation

- Bone age delayed by 3 years
  - At 10 years 7 months, bone age just 7 years 10 months
What’s Next?
- Let’s discuss!
- Differential Diagnosis
- Evaluation

What we did?
- Evaluation
  - Laboratory studies
- Radiology
  - MRI of pituitary gland

Case #1 Outcome and Treatment
- Diagnosis- Hashimoto thyroiditis
- Treatment- Levothyroxine
  - Dose escalation
  - Flip back to growth chart
  - ADD growth chart with adult height
Case #1 - Growth Chart

Growth deceleration began at age 5 years from 75th%ile. By age 9, 3rd%ile.

Case #1 – Growth Chart following treatment

Case #2
- 15 year 4 month old female presented for a growth evaluation
- Concern about height (4'6.5") and lack of growth for past 2 years
- Normal weight gain
- Menarche age 12 years old – cycles regular
Case #2

- **ROS**
  - Recent diagnosis of ADHD and anxiety within the past year
  - Treated with Vyvanse and Prozac
- **Foot pain** – EMG normal

- **Family Hx**
  - MPH: 5'3.5"
  - Paternal uncle with Type 1 DM, no other endocrine disease, MGM 5'

- **Physical Exam**
  - Tanner 5
  - High arched palate
  - Normal thyroid exam

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**Case #2**

- **Evaluation**
  - Bone age – 17 years of age

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**Case #2: What is Next?**

- **Evaluation**
  - Laboratory studies ?
- **Differential Diagnosis**
Case #2 – Outcome and Treatment

- Evaluation
- Diagnosis
- Hypothyroidism: Levothyroxine – 50 mcg daily
- OTHER??

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Case #3

12 years 2 month male originally presented at age 10 years 4 months for concerns of short stature (height 11th percentile)

PMHx:
- Birth weight: 5lb 10oz
- Gestational age: 37 weeks gestation
- No of fetal intracranial hemorrhage and venous anomaly – monitored only

ROS:
- Negative for headaches
- Mild intermittent asthma
- Developmentally normal
- Family Hx
  - Adjusted MPH: 69th
  - Thyroid disease – MGM

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Case #3

- Physical Exam
  - Prepubertal

- Evaluation
  - Bone age read as 6 years at a chronologic age of 10 years
  - AHP normal for family's genetics
  - Laboratory studies unremarkable
    - CBC, CMP, IGF-1, IGFBP-3, TSH, fT4, Celiac panel, ESR
What’s Next?

- Let’s discuss!
- Differential Diagnosis
- Further Evaluation

What we did

- Further Evaluation
- Laboratory studies
  - Provocative growth hormone stimulation test
    - GH peaked at 5.93 and 6.9
    - Repeat testing of pituitary hormones, including cortisol – ALL normal
  - Radiology
    - MRI of pituitary gland
    - Pituitary hemorrhage
- Treatment
  - Growth hormone with monitoring of MRI

Patient 4 months after starting GH
Patient 4 months after starting GH

Case #4

- 16 year 2 month old male presenting with growth deceleration and weight gain despite pubertal development

Case #4: Growth Chart
Case #4

R/O:
- Straight gain despite activity with denied excessive intake
- Vasovagal syncope
- Puberty: Tanner 4 PH; Tanner 5 Genitals (testes 15 cc)
- Family History: MPH 66.5"

PMHx: History of sixth nerve palsy repair at age 11 years

Physical Exam:
- Round facies
- Puberty: Tanner 4 PH; Tanner 5 Genitals (testes 15 cc)

Bone age at CA of 16 years 1 month, read as 14 years 6 months

Case #4: Evaluation

Case #4: What is Next?

Further Evaluation

Differential Diagnosis
Case #4: What we did

- Laboratory evaluation
  - IGF-1, IGFBP-3, TSH, fT4, CBC, CMP, Celiac panel, ESR
  - Additional tests:
    - 24 hour urine cortisol
    - Dexamethasone suppression test

- Imaging
  - MRI of pituitary gland

Case #4: Outcome and Treatment

- Outcome
  - Cushing disease

- Treatment
  - Transphenoidal surgery
  - Adrenal insufficiency
  - Steroid wean
  - Aromatase inhibitor

Questions?