

# A 19-Year Old with Congenital Adrenal Hyperplasia (CAH) and Decreased Bone Mineral Density

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# History of the Present Illness

- 19-year old Black male with classic, salt-wasting congenital adrenal hyperplasia, complicated by prior problems with medication adherence, presents to Endocrinology clinic in follow-up
- Last seen in the endocrinology clinic ~1 year prior



# Relevant History

## Past Medical History

- o CAH: diagnosed on day #4 of life, born at 34 weeks
  - o Prednisone 2.5 mg bid
  - o Fludrocortisone 0.1 mg bid
- o Central precocious puberty with short stature
- o Depression, ADD
- o Eczema
  - o Hydroxyzine
  - o Ketoconazole
  - o Triamcinolone

## Past Surgical History

- o Appendectomy
- o Tonsillectomy

## Social History

- o Graduating high school this spring
- o Plans to train to become a police officer
- o Father in military; frequently deployed
- o No tobacco, etoh, or illicit drug use
- o Sexually active – previously reports gay sexual orientation

# Review of Systems

**Constitutional:** Denies fever, chills, activity change, appetite change, and weight loss. Denies increased thirst. **+fatigue.**

**HEENT:** Denies dysphagia, hoarseness, rhinorrhea. Denies vision changes. **+tinnitus is present.**

**Resp:** Denies shortness of breath, cough, wheezing.

**CV:** Denies chest pain, lower extremity edema, palpitations.

**GI:** Denies nausea, vomiting, diarrhea, abdominal pain, constipation.

**GU:** Denies polyuria. Denies erectile dysfunction or diminished libido.

**MSK:** Denies myalgias, joint swelling, arthralgias.

**Neuro:** Denies dizziness, weakness, light-headedness, and numbness.

**Psych:** Denies behavioral problems, anxiety, or dysphoric mood.



# Physical Examination

**Constitutional:** Alert, oriented x 3. Well-developed, well-nourished.

**HEENT:** Normocephalic, atraumatic. PERR. Mucous membranes moist.

**Neck:** Supple. No thyromegaly. There are no thyroid nodules palpated.

**CV:** Regular rate and rhythm. No murmurs, rubs appreciated.

**Pulm/Chest:** Clear to auscultation bilaterally. Without wheezes or rales.

**GI:** Soft, non-tender, non-distended. Bowel sounds are present. No HSM.

**GU:** Penis normal. Normal testicular volume and size. There are no abnormal testicular masses palpated. Tanner Stage V.

**MSK:** Normal range of motion. No edema or tenderness.

**Neuro:** No focal deficits. 2+ biceps reflexes. No tremors present.

**Skin:** Warm, dry, **diffuse eczematous scaly lesions on arms, legs, and neck.**

**Psychiatric:** Normal mood and affect. Normal behavior, judgement, and thought content.

# Diagnostic Evaluation

Glucose	75
Sodium	140
Potassium	3.3
Chloride	103
CO2	28
Anion Gap	9
BUN	12
Creatinine	0.8
GFR	120
Calcium	9.2

Albumin	3.9
Total Protein	7.0
T bili	0.2
Alk Phos	43
AST	25
ALT	19

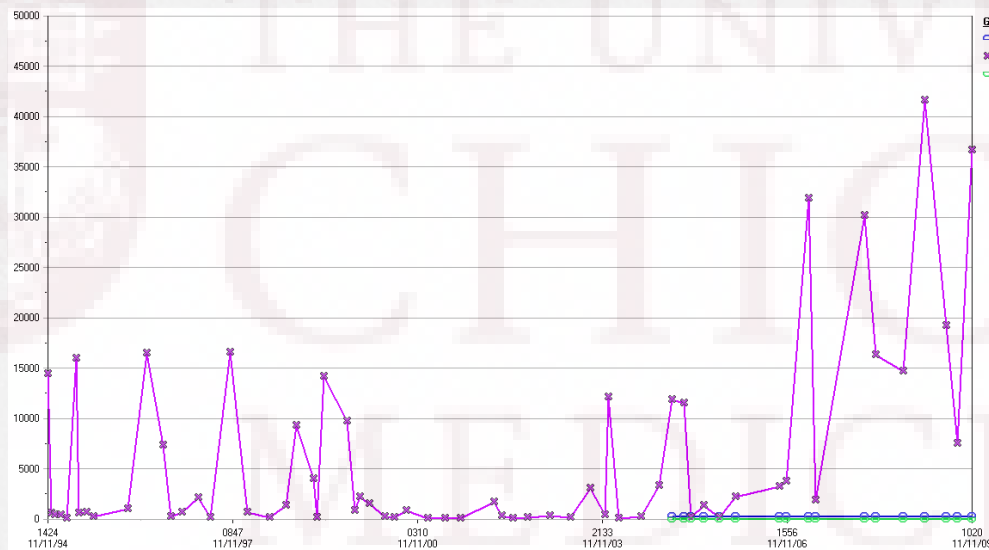
WBC	7.8
HGB	13.5
HCT	41.8
PLT	266



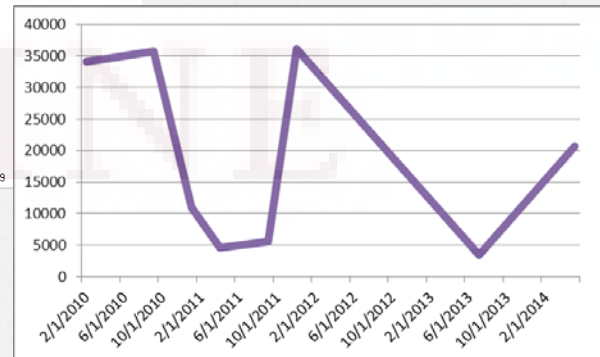
# Additional Evaluation

	12/11	4/12	8/12	7/13	5/14
17-hydroxyprogesterone < 200 ng/dL	36,100			3,440	20,700
ACTH < 52 pg/mL	686	1,230	146	97.6	370
Renin 1.2-2.4 ng/mL/h (12-17 yrs)	13.0	< 0.6	1.6	3.1	4.5
Testosterone (total)					
Androstenedione Tanner Stage V 65-210 ng/dL				65	
FSH 1.2-8.0 mIU/mL Adult male, 18-35				4.3	
LH 2.0-6.8 mIU/mL Adult male, 18-35				2.7	

# 17-Hydroxy- Progesterone Levels



Graph Legend  
17-OH Progesterone (Peds E...  
17-OH Progesterone (Peds E...  
17-OH Progesterone (Peds E...





# Diagnostic Evaluation

25-OH Vitamin D	9

TSH	1.05

## BMD Evaluation

L1-L4 spinal BMD: 0.848 g/cm<sup>2</sup>;  
Z-score of -3.2.

Left femoral neck BMD is 0.953  
g/cm<sup>2</sup>; T score of -1.7.

## CT Abdomen and Pelvis

Appendix not visualized. Inflammatory changes in distal ileum and cecum may be due to inflammatory bowel disease or other etiology. Adrenal hyperplasia. Prominent groin nodes.

# Growth

## Over-treatment

- ❖ Growth Suppression
- ❖ Hypertension
- ❖ Iatrogenic Cushing's



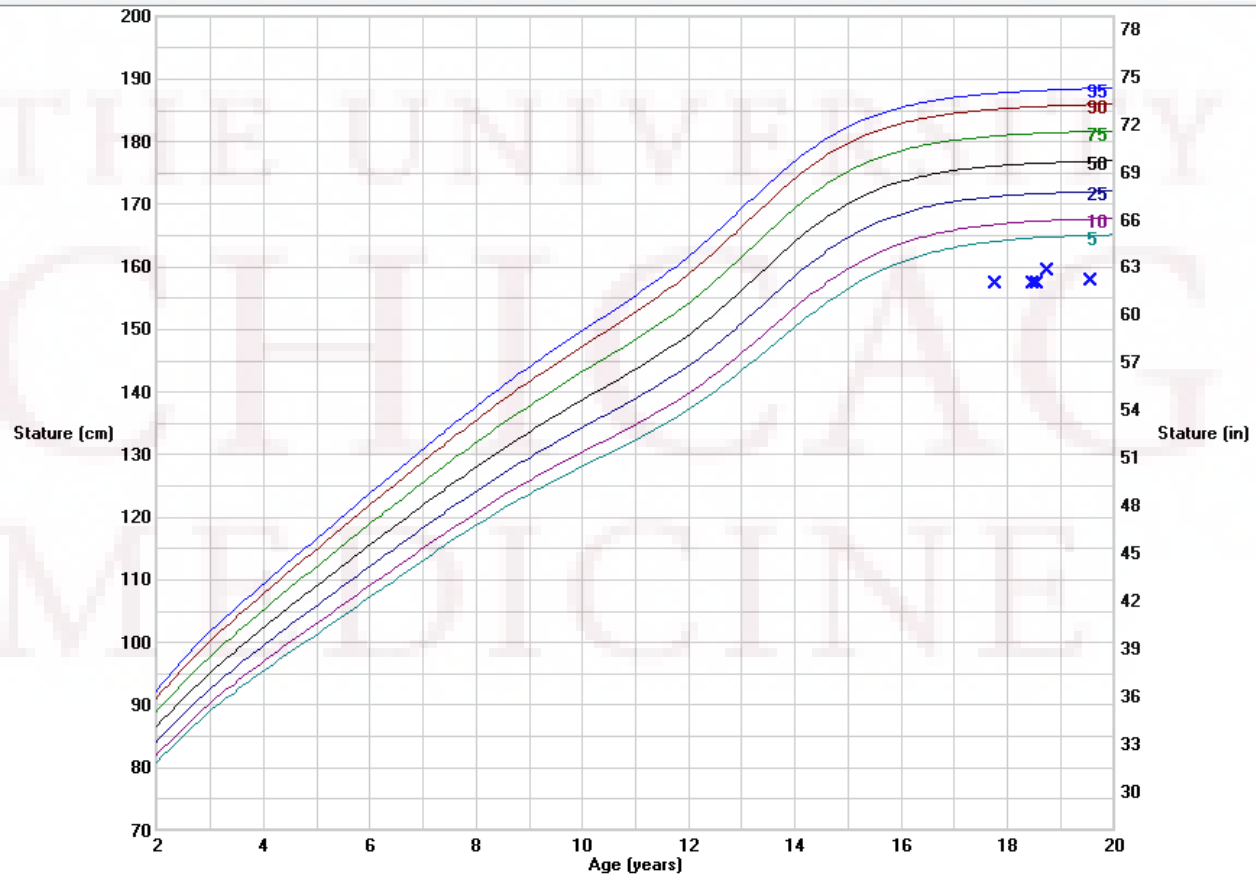
## Under-treatment

- ❖ Risk of adrenal crisis
- ❖ Increased adrenal androgen production
- ❖ Accelerated bone age and loss of growth potential



Stature-for-age Percentiles (Boys, 2 to 20 years)

Zoom: 100%



Source: Centers for Disease Control and Prevention

# Clinical Questions



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