

# Not Your Average Turner's

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## Objectives:

- Discuss Turner's syndrome
- Discuss ambiguous genitalia
- Discuss treatment options

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## Disclosure

- I have nothing to disclose

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## Patient history

- 10 4/12 yo female
- Term infant
- BW 3470
- Born with ambiguous genitalia

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## History

- Family History
- Mother 5'6", regular menses, menarche at 11 years
- Father 5'7"
  
- Social History
- Lives with mom. 4<sup>th</sup> grade. Has IEP for reading, writing and math

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## Laboratories

- 2/12/08 FISH analysis - ISCN NOMENCLATURE: nuc ish  
Xcen(DXZ1x1),Ycen(DYZ3x1)[[33]/
- Xcen(DXZ1x1),Ycen(DYZ3x1)[83]
  
- ISCN KARYOTYPE DESIGNATION: 45,X[38]/46,XY[12] (karyotype is that of an infant with mosaicism as described)

COMMENTS: In a total of 50 cells examined in detail, a bimodal number of 45 (38 cells) and 46 (12 cells) chromosomes was observed to be present. In each of the cells observed to contain 45 chromosomes, there is a single morphologically normal X chromosome present. There is no evidence of the presence of a second sex chromosome in any of these cells. In each of the cells containing 46 chromosomes, there is present a single X chromosome, and a small "G group" sized (marker) chromosome.

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## Pelvic ultrasound

- **ULTRASOUND FINDINGS:** Posterior to the bladder is an elongated structure which has central fluid echogenicity. The structure is anterior to a gas filled rectum. This has the sonographic appearance of a uterus with fluid in the canal.

### IMPRESSION:

Fluid-filled uterus within the pelvis. Two structures are identified within the inguinal canals which have the sonographic appearance of testicles. Within these probable testicles, no ovarian follicles are identified. Superior to the scrotum/fused labia, there appears to be the base of a penis. Given the presence of a uterus, evidence for testicles, and ambiguous genitalia, this may represent a case of true hermaphroditism. In true hermaphrodites, the gonads may show ovarian and testicular tissue, however on this exam only testicular appearing tissue is seen.

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## Laboratories

**INHIBIN B**  
Status: Final result Visible to patient: No (Not Released) Or: Ambiguous genitalia  
Component: Inhibin B  
Inhibin B: 47  
Comment: (NOTE)  
UNIT OF MEASURE: pg/mL

REFERENCE RANGE FOR Inhibin B:  
Male: < 400 pg/mL  
Female: < 15 pg/mL  
Normal Menstrual cycle: 15 - 200 pg/mL

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## Laboratories

Diagnostic Procedure	Result
Dihydrotestosterone	* 1635.5
Reference range: 0.0 to 49.9	
Unit: pg/mL	
(NOTE)	

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## Laboratories

Test	Flag Results	Unit	Reference Value
Anti Mullerian Hormone (AMH)	55.0	ng/mL	REPORTED 01/21/2009 13:22
Reference Range:			
Age Range			
Males			
0-13d	15.5-48.7		
14d-11m	39.1-93.1		
12m-6y	48.0-83.2		
7-8y	33.8-60.2		
Adult	3.0-5.4		
Females			
0-6y	0.0-7.1		
Adult	0.0-6.9		

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## Specialty Evaluations

- Genetics
- Endocrine-brief treatment with growth hormone in 2012
- Cardiology-normal evaluation, no follow up
- Urology-was told to come back for surgical intervention as teen to function as male or female

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## Prader Scale

### The Prader Scale Of Genitalia

- Stage 0** Normal female genitalia.
- Stage 1** Mildly enlarged clitoris, slightly reduced vaginal opening.
- Stage 2** phallus being intermediate in size, small vaginal opening with separate urethral opening. Posterior labial fusion present.
- Stage 3** Further enlarged phallus than Stage 2, with single urogenital sinus and nearly complete fusion of the labia.
- Stage 4** Looks more male than female, with an empty scrotum and a normal-sized penis-like phallus. A small urethral/vaginal opening at the base of the shaft/phallus (hypospadias in a male).
- Stage 5** This is complete male virilisation - a normally-formed penis is present, with the urethral opening at or near the tip, and the scrotum formed, but empty.
- Stage 6** Normal male presentation of the penis with normal testes.

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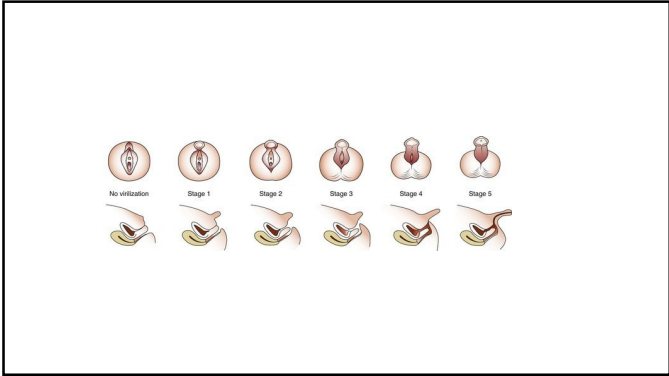
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Human Chorionic gonadotropin (hcg) stimulation test

- For patients who have testicular tissue
- Given daily or every other day-various protocols
- Lab measurement at baseline and within 1-3 days after last dose

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Hcg stimulin test

Component	12/11/2008	12/12/2008	1/9/2009	5/16/2018
Testosterone, Total	139 (H)	129 (H)	821 (H)	<10

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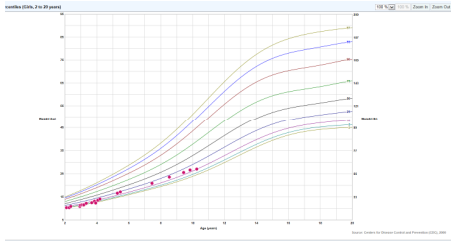
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# Growth Chart



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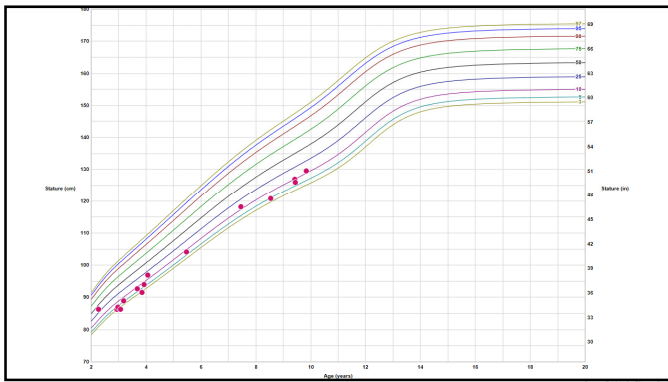
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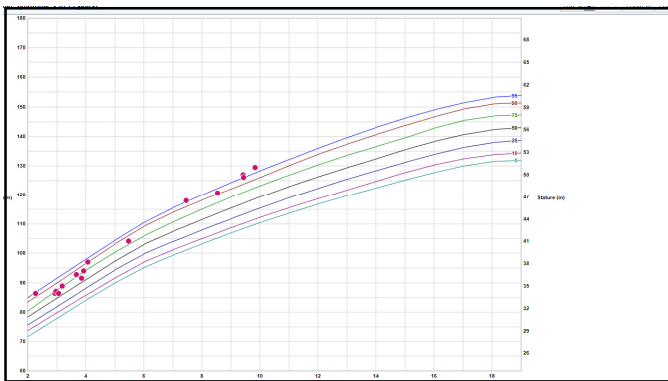
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## Bone age

- Chronological age: 9 years 5 months.

Bone age: 6 years 10 months.

Standard deviation: 10.7 months months

### IMPRESSION

Delayed bone age, more than 2 standard deviations below chronological age.

- 5/16/18

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## laboratories

Laboratory Results	
Component	5/16/2018 UM
Glucose	117
Sodium	131 (L)
Potassium	3.3
Carbon Dioxide	22
Chloride	105
BUN	9
Creatinine	0.45
Calcium	9.5
Anion Gap	14 (H)
Estimated GFR	10
WBC	6.3
HbC	2.0
Hemoglobin	12.0
Hematocrit	37.0
MCV	31
MCH	28.7
MCHC	14.8
Platelet	310
RBC-CL%	3.8
RDW	14.4
Erythrocyte Total	450.0
Lutealizing Hormone	40.26 (U)
FSH	7.4 (U)
Born-C Eia	150.0
TSH (high sens.)	0.005
T4 (Thyroxine) Free	1.01

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## Physical exam-5/2018

- General: Well appearing child, wearing T-shirt (Dream Big), pants, hair in two ponytails. Well mannered, inquisitive.
- HEENT: Coarse facial features with thick eyebrows. PERRL. EOMI. MMM, no pharyngeal erythema. 6 teeth/quadrant
- Neck: thyroid not palpable, no cervical adenopathy
- Lungs: CTAB
- Breasts: Tanner I
- CV: RRR, no murmur
- Abdomen: Non-tender, no palpable masses
- GU: Tanner II base of phallus, phallus measures 4x1.5 cm. No palpable gonads urethral opening on perineum, tiny vaginal orifice visualized
- Ext: pulses +2, no CCE
- Skin: No acne. No body odor, whorled skin pattern, increased body hair on extremities

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## Psych Evaluation

- 9 yo with disorder of sexual development, female assigned at birth who is unsure about gender identity at this time. Is not ready to provide decision on gender assignment.

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## Plan

- Restart growth hormone 0.8 mg sq daily x 1 week then increase to 1 mg x 1 week, then 1.2 mg sq daily
- Growth hormone dosing: 0.5mg/kg/day
- Continue psych counseling

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## Discussion

- Should we offer growth hormone?
- Should we offer pubertal suppression until gender identity is decided?
- Lupron/Supprelin/Triptodur
- What is the risk for cancer with surgical delay?

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