Not Your Average Turner’s
Barbara Lightner APRN, CNP-CSC
MetroHealth Systems
Cleveland, Ohio

Objectives:
• Discuss Turner’s syndrome
• Discuss ambiguous genitalia
• Discuss treatment options

Disclosure
• I have nothing to disclose
Patient history

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

History

- Family History
  - Mother 5'6", regular menses, menarche at 11 years
  - Father 5'7"

- Social History
  - Lives with mom. 4th grade. Has IEP for reading, writing and math

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.
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/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.

Laboratories

<table>
<thead>
<tr>
<th>Diagnostic Procedure</th>
<th>Result</th>
</tr>
</thead>
<tbody>
<tr>
<td>Dihydrotestosterone</td>
<td>1835.5</td>
</tr>
</tbody>
</table>

Reference range: 5.0 to 49.9 ng/mL

(NOTE)
Laboratories

<table>
<thead>
<tr>
<th>Test</th>
<th>Flag</th>
<th>Result</th>
<th>Unit</th>
</tr>
</thead>
<tbody>
<tr>
<td>Anti Mullerian Hormone (AMH)</td>
<td></td>
<td>15.0</td>
<td></td>
</tr>
</tbody>
</table>

**Reference Values:**

<table>
<thead>
<tr>
<th>Age</th>
<th>Range</th>
</tr>
</thead>
<tbody>
<tr>
<td>Neonate</td>
<td>10.3-88.7</td>
</tr>
<tr>
<td>Infant</td>
<td>38.1-91.1</td>
</tr>
<tr>
<td>Toddler</td>
<td>60.2-101</td>
</tr>
<tr>
<td>Adult</td>
<td>5.0-5.6</td>
</tr>
<tr>
<td>Female</td>
<td>0.0-1.1</td>
</tr>
<tr>
<td>Adult</td>
<td>0.0-8.9</td>
</tr>
</tbody>
</table>

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

Prader Scale

**The Prader Scale Of Genitalia**

- **Stage 1**: Normal male genitalia.
- **Stage 2**: Abnormally shaped clitoris, slightly reduced vaginal opening.
- **Stage 3**: Phallic body inter dilatation in size, small vaginal opening with separate vaginal opening. Penile shaft is present.
- **Stage 4**: Phallic body inter dilatation in size, with single orificial area and may be slightly broader than the labia.
- **Stage 5**: Looks more male than female, with an open anus, and a normally shaped penis-like phallicus. A multihernial vaginal opening at base of the phallic, phallicusHiypoplasia in male.
- **Stage 6**: Very small, normal penis with the vaginal opening at or near the hip, and the scrotum forming lab shaped.
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
Growth Chart
Bone age

- Chronological age: 9 years 5 months.
- Bone age: 6 years 10 months.
- Standard deviation: 10.7 months

**IMPRESSION**
Delayed bone age, more than 2 standard deviations below chronological age.
- 5/16/18

laboratories

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
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.

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

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?