BI 358 Lecture 12

I. **Announcements** Presentations Group I next Tuesday! Sign-in, attendance, guest feedback, .pptx vs Prezi… submit < 4 pm M. Q? Today’s lecture prep for Dr. Godfrey’s & Dr. Bradshaw’s guest lectures, Feb 25th + Mar 3rd.

II. **Congenital Hand Differences** Prep for Dr. Godfrey

III. **Neonatal & Pediatric Physiology** Prep for Dr. Bradshaw

A. What’s a *neonate*? Age range for pediatric patients?
B. Some differences?
   1. Markers to predict problems *(NB: rare ~95%x OK!)*
   2. *cf:* Neonate vs. adult human values (selected)
   4. Heart differences?
   5. More frequent, yet still uncommon problems: congenital genetic defects, Tetralogy of Fallot, Down syndrome, Edward's syndrome, Cystic fibrosis

C. Development & Pediatrics tour, Tanner scale. Ref: Moore, Persaud, Shiota (MPS); Johnson (RVJ) +...
Jenna Godfrey, MD, MSPH
Pediatric Upper Extremity
Adult Hand & Wrist

20 in 10,000 or 0.002
More common in ♂ than ♀

Congenital Hand
Differences in
the Media!

https://orthoinfo.aaos.org/en/diseases--conditions/congenital-hand-differences
Upper Extremity Bones

- Clavicle
- Scapula
- Humerus
- Ulna
- Radius
- Carpals
- Metacarpals
- Phalanges

Image Source: [http://spmsubjects.blogspot.com](http://spmsubjects.blogspot.com)
Hand

Image Source: http://en.wikipedia.org/wiki/Hand
Neonate
Gr. neos/new
L. -nat/born

infant
1st 4wk > birth

Charles A. Hoffmeister, MD
Neonatal/Perinatal Specialist
Pediatrics = Gr. παιδιά/paidiá/children
Γιατρός/Giatrós/physician

medical branch that treats child: development, care, disease treatment
In the USA:
13.4 million readers
57,695 pediatricians
2,100 nominated
7 chosen
1 is our Dr. B

You’re one of our favorite pediatricians and
*Parents* magazine’s, too!

Pilar Bradshaw, M.D., F.A.A.P.
PROVIDING INTERVENTION AND ADVOCACY FOR CHILDREN WHO ARE VICTIMS OF OR WITNESSES TO CRIME

https://kidsfirstcenter.net/
<table>
<thead>
<tr>
<th>VARIABLE</th>
<th>NEONATE</th>
<th>ADULT</th>
</tr>
</thead>
<tbody>
<tr>
<td>WT (lb)</td>
<td>7</td>
<td>♂ 110  ♂ 150</td>
</tr>
<tr>
<td>WT (lb, range)</td>
<td>4.5 – 11</td>
<td>wide variation</td>
</tr>
<tr>
<td>HR (b/min)</td>
<td>130</td>
<td>~2 × 70</td>
</tr>
<tr>
<td>RR (breaths/min)</td>
<td>40</td>
<td>3 × 12-15</td>
</tr>
<tr>
<td>BV (mL)</td>
<td>! 16.7 × 300</td>
<td>( \frac{1}{17} ) × 5000</td>
</tr>
<tr>
<td>CO/(\dot{Q}) (mL/min)</td>
<td>100 × 50</td>
<td>( \frac{1}{100} ) × 5000</td>
</tr>
<tr>
<td>BP (mm Hg)</td>
<td>70/50</td>
<td>?? × 120/80</td>
</tr>
<tr>
<td>BMR (relative)</td>
<td>2x Adult</td>
<td>1</td>
</tr>
<tr>
<td>FLUID Δ(relative)</td>
<td>7x Adult</td>
<td>1</td>
</tr>
</tbody>
</table>

< 6.0 premature

H₂O Homeostasis!
1. Ductus Arteriosus
2. Foramen Ovale
3. Ductus Venosus

G&H 2016 fig 84-4
G&H 2011 fig 83-4
How so fast? Cell divisions in as little as 4 hr!

Cells: 1, 2, 4, 8, 16, 32, 64, ... 100 trillion!

24 hr/d

$0, 4, 8, 12, 16, 20, 24$

Baby: ~38-40 wk

Distinct human appearance: 28 wk later

Embryoblast

Embryo

Organogenesis

$t = 0$

3 wk

8 wk ≥ 9th wk

Fetus =
Embryo?
Fetus?
Baby?
Fetus @ 28 wk or 7 mo
1100 g (1.1 kg) ≈ 2.5 lb

J Langman 1981 Medical Embryology p 80
### As a Pregnant Female –

**Where you’ll gain the weight**

<table>
<thead>
<tr>
<th>Component</th>
<th>Range</th>
</tr>
</thead>
<tbody>
<tr>
<td>Your baby</td>
<td>6½ to 9 pounds</td>
</tr>
<tr>
<td>Placenta</td>
<td>1½ pounds</td>
</tr>
<tr>
<td>Amniotic fluid</td>
<td>2 pounds</td>
</tr>
<tr>
<td>Breast enlargement</td>
<td>1 to 3 pounds</td>
</tr>
<tr>
<td>Uterus enlargement</td>
<td>2 pounds</td>
</tr>
<tr>
<td>Fat stores and muscle development</td>
<td>4 to 8 pounds</td>
</tr>
<tr>
<td>Increased blood volume</td>
<td>3 to 4 pounds</td>
</tr>
<tr>
<td>Increased fluid volume</td>
<td>2 to 3 pounds</td>
</tr>
</tbody>
</table>

**Total**

22 to 32½ pounds

Surprise!

Head

Head
What are my chances of having a child with a birth defect?  \( \leq 5\% \)

Of every 100 babies born in the United States, 95 to 97 are born healthy (no major medical or surgical intervention is necessary). According to the March of Dimes Birth Defects Foundation:

- One of every 175 is born with a congenital heart defect.
- One of every 400 is born with clubfoot.
- One of every 700 is born with cleft lip and palate.
- One of every 800 is born with Down syndrome.
- One of every 2,000 is born with spina bifida.

To put this list into perspective, consider the following:

- The odds of having twins are about one in 100.
- The odds of having triplets are about one in 8,000.
Tetralogy of Fallot

1. Aorta Displacement
2. Pulmonary Stenosis
3. Ventricular Septal Defect
4. R Ventricular Hypertrophy

f = 3.3 per 10,000 live births
15% TOF 22q11 deletion
7% TOF trisomy 21
≥ 4% TOF NKX2.5 mutation

G&H 2016 & 2011 fig 23-5
## Chromosome abnormalities: What are your risks?

<table>
<thead>
<tr>
<th>Age</th>
<th>Risk for Down syndrome</th>
<th>Total risk for clinically significant chromosome abnormalities</th>
</tr>
</thead>
<tbody>
<tr>
<td>20</td>
<td>1/1,667</td>
<td>1/526</td>
</tr>
<tr>
<td>21</td>
<td>1/1,667</td>
<td>1/526</td>
</tr>
<tr>
<td>22</td>
<td>1/1,429</td>
<td>1/500</td>
</tr>
<tr>
<td>23</td>
<td>1/1,429</td>
<td>1/500</td>
</tr>
<tr>
<td>24</td>
<td>1/1,250</td>
<td>1/476</td>
</tr>
<tr>
<td>25</td>
<td>1/1,250</td>
<td>1/476</td>
</tr>
<tr>
<td>26</td>
<td>1/1,176</td>
<td>1/476</td>
</tr>
<tr>
<td>27</td>
<td>1/1,111</td>
<td>1/455</td>
</tr>
<tr>
<td>28</td>
<td>1/1,053</td>
<td>1/435</td>
</tr>
<tr>
<td>29</td>
<td>1/1,000</td>
<td>1/417</td>
</tr>
<tr>
<td>30</td>
<td>1/952</td>
<td>1/385</td>
</tr>
<tr>
<td>31</td>
<td>1/909</td>
<td>1/385</td>
</tr>
<tr>
<td>32</td>
<td>1/769</td>
<td>1/322</td>
</tr>
<tr>
<td>33</td>
<td>1/602</td>
<td>1/286</td>
</tr>
<tr>
<td>34</td>
<td>1/485</td>
<td>1/238</td>
</tr>
<tr>
<td>35</td>
<td>1/378</td>
<td>1/192</td>
</tr>
<tr>
<td>36</td>
<td>1/289</td>
<td>1/156</td>
</tr>
<tr>
<td>37</td>
<td>1/224</td>
<td>1/127</td>
</tr>
<tr>
<td>38</td>
<td>1/173</td>
<td>1/102</td>
</tr>
<tr>
<td>39</td>
<td>1/136</td>
<td>1/83</td>
</tr>
<tr>
<td>40</td>
<td>1/106</td>
<td>1/66</td>
</tr>
<tr>
<td>41</td>
<td>1/82</td>
<td>1/53</td>
</tr>
<tr>
<td>42</td>
<td>1/63</td>
<td>1/42</td>
</tr>
<tr>
<td>43</td>
<td>1/49</td>
<td>1/33</td>
</tr>
<tr>
<td>44</td>
<td>1/38</td>
<td>1/26</td>
</tr>
<tr>
<td>45</td>
<td>1/30</td>
<td>1/21</td>
</tr>
</tbody>
</table>

**Implications relative to Dr. Kaplan’s lecture & delaying pregnancy!**

RVJ, Mayo Clinic p 59.
95% of Down Syndrome Trisomy 21
90% of Cases → Eggs Are Abnormal

Normal ♀

Down Syndrome ♂

Quad Screen? 4 Blood Chemistry Tests

2\textsuperscript{nd} trimester, neural tube defects & chromosomal abnormalities, 81% sensitivity, 5% false +

- **AFP:** \textit{alpha-fetoprotein}, fetal liver
  - High, neural tube defects (spina bifida)

- **hCG:** \textit{human chorionic gonadotropin}, placenta
  - High, Down syndrome (Trisomy 21)
  - Low, Edward's syndrome (Trisomy 18)

- **Estriol:** placenta + fetal liver

- **Inhibin-A:** placenta + ovaries

[http://www.americanpregnancy.org/prenataltesting/quadscreen.html](http://www.americanpregnancy.org/prenataltesting/quadscreen.html)
Amniocentesis or Chorionic Villus Sampling?
**Down Syndrome Fetus**

*NB*: 1:1400 incidence for maternal age 20-24; 75% spontaneously aborted. Flat frontal facies, anomalous auricles, simian crease, clinodactyly.

*SOURCE*: KL Moore, TVN Persaud & K Shiota (MPS)1994
# DOWN SYNDROME NEONATE

## 10 KEY FEATURES (Hall)

<table>
<thead>
<tr>
<th>Feature</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. Facial profile flat</td>
<td>90%</td>
</tr>
<tr>
<td>2. Hypotonia</td>
<td>80%</td>
</tr>
<tr>
<td>3. Poor Moro reflex</td>
<td>85%</td>
</tr>
<tr>
<td>4. Joint hyperflexibility</td>
<td>80%</td>
</tr>
<tr>
<td>5. Skin excess nape of neck</td>
<td>80%</td>
</tr>
<tr>
<td>6. Palpebral fissures slanted</td>
<td>80%</td>
</tr>
<tr>
<td>7. Pelvic dysplasia</td>
<td>70%</td>
</tr>
<tr>
<td>8. 5\textsuperscript{th} finger mid-phalynx dysplasia</td>
<td>60%</td>
</tr>
<tr>
<td>9. Auricles anomalous</td>
<td>60%</td>
</tr>
<tr>
<td>10. Simian crease</td>
<td>45%</td>
</tr>
</tbody>
</table>
Dizygotic Twins Discordant for Down Syndrome
FIGURE 2. Down syndrome. A, Young infant. Flat facies, straight hair; protrusion of tongue; single crease on inturned fifth finger.
Recessive Disorders
eg, Cystic Fibrosis

RVJ, Mayo Clinic p 61.

f = 4 in 10,000 live births
CFTR gene, 7q31.2
long arm chromosome 7

Most Common Position. Ideal!!

RVJ, Mayo Clinic p 317.

cf: G&H 2016 fig 83-9
G&H 2011 fig 82-9
Occiput Posterior/Sunnyside up! Oh No! …Yikes!

Largest presenting diameter!

RVJ, Mayo Clinic p 318.
Breech! eg, Frank

RVJ, Mayo Clinic p 319.
A baby who is positioned horizontally across the uterus, rather than vertically, is in a transverse lie position. Most babies in this position have a cesarean birth.
Baby @ birth
38 wk or 266 d
> conception!
3200 g (3.2 kg)
≈ 7 lb
35 cm
## Apgar Scores: How Healthy Is Your Newborn?

<table>
<thead>
<tr>
<th>Sign</th>
<th>Points</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>0</td>
</tr>
<tr>
<td>Appearance (color)</td>
<td>Pale or blue</td>
</tr>
<tr>
<td></td>
<td></td>
</tr>
<tr>
<td>Pulse (heartbeat)</td>
<td>Not detectable</td>
</tr>
<tr>
<td>Grimace (reflex irritability)</td>
<td>No response to</td>
</tr>
<tr>
<td></td>
<td>stimulation</td>
</tr>
<tr>
<td>Activity (muscle tone)</td>
<td>Flaccid (no or</td>
</tr>
<tr>
<td></td>
<td>weak activity)</td>
</tr>
<tr>
<td>Respiration</td>
<td>None</td>
</tr>
</tbody>
</table>

Scores determined for each sign are totaled. The highest possible score is 10. By 5 minutes of age, most healthy babies have scores of at least 7. A score less than that indicates that the baby warrants careful watching.

Virginia Apgar, MD, Anesthesiologist, 1953
Figure 83-7

Fall in body temperature of the neonate immediately after birth, and instability of body temperature during the first few days of life.
Behavioral development of the infant during the first year of life.
Cephalic to Caudal Development

2 mo. (fetal) 5 mo. Newborn 2 yr. 6 yr. 12 yr. 25 yr.

Courtesy Dr. Allen Harlor, PeaceHealth Medical
Cholesterol

Brain Weight

DNA

Courtesy Dr. Allen Harlor PeaceHealth Medical
Infant Eustachian tube smaller + more horizontal!

An infant’s ear is different from an adult’s ear because the eustachian tube is more horizontally positioned. Because of this, drainage from the middle ear occurs less easily, and your baby is at greater risk for an ear infection (otitis media). This condition occurs when the eustachian tube becomes blocked and fluid is trapped. It is marked by swelling and discoloration of the eardrum.

- Fluid-filled middle ear
- Bulging eardrum
- Swelling and inflammation
Tanner Stages of Development
Tanner Stages for Breast Development
Tanner Stages? What are the Ages?
Tanner Stages? What are the Ages?

All 14 $\frac{3}{4}$ yr!!

All 12 $\frac{3}{4}$ yr!!