Congenital Disorders of the Newborn

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Disclosures

• The Presenter has no disclosures to make.
Objectives

1. Assess common congenital disorders of the newborn.
2. Recommend appropriate intervention/plan of care after diagnosis.
3. Summarize updates in newborn screening recommendations.
4. Examine diagnosis of congenital anomalies through the lens of genomics.
5. Integrate new guidelines and advanced assessment skills into future practice changes.
Where do you work?

A. Newborn Nursery, Hospital Setting
B. Pediatric Primary Care, Outpatient Setting
C. Neonatal ICU
D. Other
Genetic Considerations of the Newborn

• Incidence of chromosomal abnormalities: 6 in 1000

• Causative genetic diagnosis in developmental delays: 14%

• Number one cause of death in infants < 1 year of age, accounting for 20% of all infant deaths
Newborn Screenings

• Since 1963 in MA for PKU (HRSA)

• Saves 12,500 babies annually

• Guthrie vs. Point of Care

• Components vary by state

• HHS Advisory Panel (2000)
  • Uniform Screening (31)

https://www.healthy.arkansas.gov/programs-services/topics/newborn-screening
Criteria for Population-Based Screening: Core, Secondary, and N/A

• Substantial Public Health Benefit
• Feasibility of Screening
• Satisfactory Laboratory Methods
• Appropriate Laboratory Facilities and Quality Control
• Appropriate Counseling, Treatment and Follow-Up
• Acceptable Costs
• Effective Education & Evaluation of Program Outcomes
Advances in NBS Technology

• MS/MS vs. DNA extraction
• Digital Microfluidics (“lab-on-a-chip”)
• DNA genome sequencing on-demand
  • Rapid Diagnosis
  • Family Planning
  • Expediting Research
  • Disease Prevention
• Genomic Sequencing and Newborn Screening Disorders Program
  • $5 million grant
  • 4 group comparison
  • Studying effectiveness of genome sequencing for population screening
Ethical Considerations

• Consent
  • Opt-Out policies
  • Privacy and Confidentiality

• Residual Dried Blood Spots
  • 20 states address retention, 13 address information, 17 have no laws
  • NBSTRN developed VRDBS
    • Repository of >2 million bloodspots for researchers
  • Lessons learned from Minnesota and Texas
  • Incidental Findings
Perinatal History

- Maternal age, gravida, para
- Maternal Blood group & Rh factor
- Historic duration of pregnancy (# weeks)
- Prenatal conditions/complications
- Intrapartal Complications
- Medications used during labor
- Family History

- Delivery anesthesia
- Delivery date & time
- Type of delivery & rationale
- Hours of ROM before delivery
- Length of Labor (>24hr & <6 hrs-fetal stress)
- GBS status
- Birth weight
- APGAR score
Genomic Aspects of Perinatal History

- Maternal Health & Habits
- Parental Health & Age at conception
- Ethnicity
- Consanguinity
- Teratogen Exposure
- Amniotic Fluid Volumes
- Fetal Activity

- Pre-conception counseling
- Amniocentesis
- Ultrasound
- Chorionic Villus Sampling
- Abnormal Fetal U/S
Assessment by Observation

- Overall size, shape, & symmetry
- Activity State
  - Quiet sleep
  - Active sleep
  - Drowsy
  - Alert
  - Fussy
  - Crying
- Posture, tone
- Skin color, signs of trauma

- Respiratory effort
  - Grunting
  - Nasal flaring
  - Retractions
  - Respiratory Rate
- Jitteriness
  - No abnormal gaze or eye movements
  - Stimulus sensitive
  - Predominant motion is tremor-not clonic jerking
Dysmorphic Features
Genetic Considerations in Childhood
Considerations

- Number of Systems Involved
- Major or Minor
- Family Characteristics
- RAPID Approach
  - Pedigree
  - Physical Assessment
  - Developmental Assessment
Growth

• Accurate Measurements
• Standardized Growth Charts
• Decreased weight gain— GI or nutritional
• Decreased height velocity— endocrine
• Both— familial vs. multi-system
• When to Worry
Examples of Dysmorphic Features

• Head
  • Dyssmorphic Skull
  • Craniosynostosis
  • Micro/Macrocephaly
  • Frontal Bossing
  • Facial Assymetry
  • High Forehead
  • Low Set posterior hairline
  • Vertical furrowing of central forehead
  • Round/Long/Flat Facies
  • Finely arched/Heavy eyebrows

• Eyes
  • Long eyelashes
  • Ptosis
  • Epicanthal folds of the eyes
  • Upward or downward slanting palpebral fissures
  • Wide-Set Eyes
  • Almond Shaped Eyes
  • Coloboma
  • Iris Abnormality
  • Strabismus
  • Nystagmus
  • Glaucoma
  • Congenital Cataracts
Examples of Dysmorphic Features

• Choanal Atresia
• Oppose thumb to volar forearm
• Skeletal disproportion
• Cubitus Valgus
• Arachnodactyly
• Ectrodactyly
• Pes planus
• Toe walking
• Malar hypoplasia
• Bifid uvula
• Hypodontia
• Conical anterior teeth
• Micrognathia
• Webbed neck
• Redundant Skin
• Café-au-lait spots
• Micropenis
• Pectus excavatum/carinatum
Case Study

Hypotonic Posture
Umbilical Hernia
Coarse Facial Features
Large Anterior Fontanelle

Macroglossia

Photo Credit: https://emedicine.medscape.com/article/919758-overview
What is the most likely diagnosis?

A. Congenital Adrenal Hyperplasia
B. Congenital Hypothyroidism
C. VACTERL Syndrome
D. Cri du Chat
After treatment initiated......

Tone improved
Umbilical hernia reducing in size

More alert
Macroglossia resolved

Photo Credit:
Case Study

What is the most appropriate question to ask in this mother’s prenatal history?

A. Did you travel during your pregnancy?
B. Do you have a history of any sexually transmitted infections?
C. Do you have a family history of microcephaly?
D. Did you experience a traumatic injury during pregnancy?
Case Study

Finding One

Finding Two

Finding Three

Photo Credit: https://www.cdc.gov/ncbddd/birthdefects/downsyndrome.html
What is the most likely diagnosis?

A. Turner’s Syndrome
B. Marfan’s Syndrome
C. Down’s Syndrome
D. Fetal Alcohol Syndrome
Case Study

• Preauricular Skin Tag
  • Otherwise normal physical exam
  • Unremarkable prenatal history

https://neoreviews.aappublications.org/content/4/4/e99.figures-only
What is the most appropriate test to order at this time?

A. Urine and Meconium Drug Screen
B. Hearing Screen
C. Renal Ultrasound
D. Xray: Plain Skull Films
Case Study

- Male infant at 18 hours of life
- Initial exam at 4 hours of life unremarkable
- Weak cry
- O2 sat 83% RA, 84% on blow-by
- Glucose 16
- Tachypnea, RR=108, Grunting, Intercostal Retractions
What is the most likely diagnosis for this infant?

A. Transient Tachypnea of the Newborn
B. Coarctation of the Aorta
C. Spontaneous Pneumothorax
D. Total Anomalous Pulmonary Venous Return
Snowman Sign

# Central Cyanosis

<table>
<thead>
<tr>
<th>Cardiac</th>
<th>Pulmonary</th>
</tr>
</thead>
<tbody>
<tr>
<td>Worsens with crying</td>
<td>Improves with crying</td>
</tr>
<tr>
<td>Comfortable at rest</td>
<td>Respiratory distress</td>
</tr>
<tr>
<td>No response to $O_2$</td>
<td>Responsive to $O_2$</td>
</tr>
<tr>
<td>May have abnormal EKG</td>
<td>Normal EKG</td>
</tr>
<tr>
<td>Abnormal CXR</td>
<td>Normal cardiac silhouette</td>
</tr>
</tbody>
</table>
Pulse Oximetry

- AAP/AHA consensus statement
- Figure:
  - A: Cyanotic CHD
  - B: Differential Cyanosis: CoA
  - C: Reverse Differential Cyanosis: TGA

Case Study
Noted on physical exam

What is the most likely diagnosis?

A. VACTERL Syndrome
B. TORCH Syndrome
C. Holt-Oram Syndrome
D. Spina Bifida/Myelomeningocele
Case Study

Reports increased spit-up

Poor Feeder

Na 121, Potassium 6.5, Glucose 25

What is important to rule out?

A. Congenital Hypothyroidism
B. Angelman Syndrome
C. Bladder Exstrophy
D. Congenital Adrenal Hyperplasia
Case Study

Otherwise normal exam

What is the most appropriate provider order to enter?

A. Implement seizure precautions
B. Order an echocardiogram
C. Obtain a renal ultrasound
D. Draw labs: CBC, CMP, LFT
Case Study

Healthy Male Newborn

No other physical exam anomalies

https://www.mayoclinic.org/birthmarks/sls-20076683?s=2
What is the most likely diagnosis?

A. Mongolian Spots
B. Neurofibromatosis Type I
C. Tuberous Sclerosis
D. Congenital Nevi
Case Study

Systolic murmur noted on exam.

http://www.craniofacial.org/en/content/velocardiofacial-syndrome-vcfs
What is the most likely diagnosis?

A. Trisomy 13
B. Velocardiofacial Syndrome
C. Cri du chat Syndrome
D. Rhett Syndrome
Craniostenosis/Craniosynostostenosis

- Premature fusion of skull sutures
- Look for absent fontanels
- Normal brain growth inhibited
- Usually develops in utero
- 20% associated with genetic syndrome
- Surgery usually performed at 4–7 months

https://www.cdc.gov/ncbddd/birthdefects/craniosynostosis.html
Nose

• Observe for symmetry
• Ensure nare patency (infants are obligate nasal breathers)

• Positional deformities may result from birth process
  • If able to manipulate, will probably correct itself
  • If non-pliable, may be associated with congenital deformity

• Choanal Atresia
Case Study

- Webbed Neck
- Edema of the dorsum (hands and feet)
- Short fingers

What is the most likely diagnosis?

A. Acyanotic Heart Defect
B. Cyanotic Heart Defect
C. Congenital Adrenal Hyperplasia
D. Turner Syndrome
What is the most likely diagnosis?

A. Treacher-Collins Syndrome
B. Potter’s Syndrome
C. Cri du Chat Syndrome
D. Prader Willi Syndrome
Marfan Syndrome

- Autosomal-dominant
- Connective tissue disorder
- Cystic medial necrosis of aorta
- Joint laxity
- Subluxation of ocular lens
- Arm span exceeds height

https://www.marfan.org/marfan-related-conditions/neonatalmarfan
Myelomeningocele

- Neural tube defect
- 75% cases occur in lumbosacral regions
- 1:1,000 live births
- Flaccid paralysis of lower extremities
- Absence of DTR
- Lack of response to touch & pain
- Constant urinary dribbling
- Mortality 10-15%
- 70% normal intelligence

https://pedclerk.bsd.uchicago.edu/page/myelomeningocele
Summary

• Examine patients through a genetic lens
• Genetic Counseling
• GINA