Endocrinology Emergencies in the Newborn
Cindy Young, BSN, RN, CPN
Ambulatory Endocrine Care Manager

Objectives

1. List the pathophysiology of an inheritance pattern of inborn errors of metabolism
2. Describe clinical presentation and symptoms of inborn errors of metabolism
3. Explain studies used to diagnose inborn errors of metabolism and the role of the nurse
4. Identify neonatal emergencies of the thyroid gland disorder, hypothyroidism, Grave’s disease and understand the diagnostic tests and role of the nurse
5. Discuss the neonatal adrenal disorders and treatment
6. Discuss panhypopituitarism and hypoglycaemia in the newborn

Conflict of Interest Disclosure

No conflict of interest related to the content of the presentation
In 1811, care of the newborn was managed by nurses and midwives in the home.

Nurse assessment skills remain critical. The nurse assessment and “concern” about a patient often influence the timeliness of diagnosis and interventions.
Uncommon and a challenge with diagnosis

Significant overlap with other neonatal emergencies

Challenge to implement screening programs & improve current tests

Neonatal Metabolic and Endocrine Emergencies

Metabolic
Inborn Errors of Metabolism

Endocrine
Neonatal Thyroid Disorders
Adrenal Disorder
Hypopituitarism

Neonatal Metabolic Emergencies
Inborn Errors of Metabolism (IEM)

Pathophysiology
Gene mutation, deficiencies in enzymes and cofactors lead to a blocked metabolic pathway
# Neonatal Metabolic Emergencies

## Inborn Errors of Metabolism (IEM)

<table>
<thead>
<tr>
<th>Pathophysiology</th>
</tr>
</thead>
<tbody>
<tr>
<td>Gene mutation, deficiencies in enzymes and cofactors lead to a blocked metabolic pathway</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Fetal effect</th>
</tr>
</thead>
<tbody>
<tr>
<td>Usually absent at birth due to placental protection</td>
</tr>
</tbody>
</table>

## Inheritance Pattern

- Generally autosomal recessive
- Some are X linked

---

## Autosomal Recessive

### X-Linked

![Autosomal Recessive Inheritance Diagram](image-url)
Clinical Presentation

- Usually appear normal at birth
- Within hours, days, sometimes weeks or months develop non-specific symptoms

Clinical Presentation of IEM

Signs and symptoms may mimic sepsis

- Neuro-lethargy, irritability, seizures, coma
- Respiratory-increased or decreased effort
- Ketonuria
- Jaundice
- Cardiomyopathy, arrhythmias
- Unusual color or odors

Ketonuria with IEM

- Ketonuria is uncommon in newborn infants.
- Anyone caring for newborn with severe illness and ketonuria must consider inborn error of metabolism
May Point to Possible IEM

- Acute onset and rapid progression of symptoms after an interval of normal health
- Unusual severity of symptoms
- Symptoms may correspond with feeding
- In general, the accumulation of toxic intermediates takes place between day 2 and 5 of life
- History of unexplained neonatal or infant death in family

Diagnosis

- Prenatal
- Newborn Screening
- Neonatal Diagnosis
- Post Mortem

General Management of IEM

- Supportive care: respiratory support, IV fluids, antibiotics
- Nutrition: dietary restrictions
- Removal of toxic substances: dialysis
- Administration of cofactors (vitamin)
- Liver transplant
Role of the Nurse

- Acute assessment and comprehensive history
- Metabolic work-up: ABG, CBC, lytes, glucose, urinalysis, ammonia*
- Baby’s advocate
- Parent / caregiver advocate
- Consult and coordinate care

Role of the Nurse

- Assisting the family with a crisis situation
- Assess the coping resources of parents and family
- Understand that grieving can occur as response to a hoped for “perfect” infant
- Anticipatory guidance

Nursing Care-Ambulatory

*Keep a close eye on labs and follow-up care after discharge!
Specific Disorders of IEM

1. Amino Acid Disorders
2. Organic Acid Metabolism / Methylmalonic Acidemia
3. CHO Metabolism Disorder: Galactosemia
4. Urea Cycle / Hyperammonemia Disorder
5. Fatty Acid Oxidation Disorder: MCADD

Amino Acid Disorder
Phenylketonuria (PKU)

<table>
<thead>
<tr>
<th>Pathophysiology</th>
<th>Presentation</th>
<th>Inheritance Pattern</th>
</tr>
</thead>
<tbody>
<tr>
<td>• Deficiency of enzyme, phenylalanine hydroxylase, needed for conversion of phenylalanine to tyrosine</td>
<td>• Abnormal newborn screen</td>
<td>• Inherited autosomal recessive trait</td>
</tr>
<tr>
<td>• Phenylalanine is part of all complete proteins</td>
<td>• Vomiting</td>
<td>• Incidence 1:12,000 newborns</td>
</tr>
<tr>
<td></td>
<td>• Difficulty feeding</td>
<td></td>
</tr>
<tr>
<td></td>
<td>• Infantile spasms</td>
<td></td>
</tr>
<tr>
<td></td>
<td>• Mousy smell</td>
<td></td>
</tr>
<tr>
<td></td>
<td>• Hypopigmentation</td>
<td></td>
</tr>
<tr>
<td></td>
<td>• Eczema</td>
<td></td>
</tr>
</tbody>
</table>

PKU Treatment

• Restriction of phenylalanine
• Special low protein infant formula
• Diet is lifelong and awful
• DNA sequencing mutational analysis may be used to determine carriers in families
• Developmental delay if untreated
• Most people with PKU can live as long and healthy as anyone else if the diet is started as infants and continued throughout their life
Recipe for low Phe Pancakes

- 1/3 c wheat starch
- 1/4 c corn starch
- 1/3 c welplan baking mix
- 1/2 t methylcellulose
- 1/2 t cinnamon
- 1 t Ener-G egg replacer
- 1 t metamucil or other fiber product
- 1/2 t baking powder
- 1/2 c mocha mix
- 1/4 c apple sauce
- 1/2 c water

Case study

- Birth History: 8lb, 1 oz; product of full-term, uncomplicated gestation. Apgar scores 8 and 8. No problems during newborn period

- History: Hospitalized with bronchiolitis at 5 months of age. No neurological abnormalities noted on physical exam. Parents note he walked at 1 1/2 years of age.

  At 4 years, was referred for neurological eval because of lack of speech. He was not toilet trained, was unable to feed or dress himself, related poorly to others and occasionally engaged in rocking and head banging. He had eczema on the palms of his hands. An EEG showed seizure activity and he was started on phenytoin. At 6 years, he had progressive encephalopathy, severe mental retardation and seizure disorder.

- Lab: Plasma phenylalanine level was 23 mg/dL (0.4-1.6). Urinary ferric chloride test was positive for phenylketonuric derivatives, despite the lack of musty urine odor. The original newborn filter paper screen for PKU had been omitted.

Amino Acid Disorder
Maple Syrup Disease (MSUD)

Pathophysiology
- Missing enzyme: Branched Chain Ketoacid Dethyldrogenase (BCKAD): needed to change 3 branch amino acid: leucine, isoleucine, and valine. Without enzyme-changed to toxic ketoacids

Presentation
- Abnormal newborn screen
- Vomiting-poor appetite
- Metabolic acidosis; rapid respirations
- Hypertonia
- The urine smells like maple syrup

Inheritance Pattern
- Inherited rare autosomal recessive trait
- Incidence: Less than 1,200,000 births
- Mennonites have a higher incidence: 1:380
**MSUD Treatment**

- Peritoneal dialysis to reduce the amino acid level acutely
- Low protein infant formula
- Avoid cow's milk, regular formula, meat, fish, cheese and eggs. Regular flour, dried beans, nuts and peanut butter may have branch chained amino acids and must be avoided or strictly limited.
- Lifelong treatment with MSUD diet is necessary
- Children are at risk for metabolic crisis when they don’t follow the diet
- Regular lab tests to measure amino acid levels
- Liver transplant
- Genetic testing

**Role of the Nurse MSUD**

- Coordinate care
- Dietary
- Genetics
- Developmental Intervention
- Call the doctor
- Poor appetite
- Behavioral changes
- Vomiting
- Infection or illness
- Educate
- Emotional support
- MSUD family support

http://www.msud-support.org/
Nursing Care MSUD - Ambulatory

Keep a close eye on baby and follow up care after discharge.
Example: Branch chain amino acids (BCAA) due on Friday, June 3.

Organic Acid Disorder
Methylmalonic Acidemia (MMA)

<table>
<thead>
<tr>
<th>Pathophysiology</th>
<th>Presentation</th>
<th>Inheritance Pattern</th>
</tr>
</thead>
<tbody>
<tr>
<td>• Missing enzyme needed to break down certain fats and amino acids</td>
<td>• Newborn screen</td>
<td>• Inherited rare autosomal recessive trait</td>
</tr>
</tbody>
</table>

| | • Ketonuria | • Incidence 1:67,000 |
| | • Vomiting | |
| | • Poor appetite | |
| | • Extreme sleepiness | |
| | • Hypotonia | |
| | • Hypoglycemia | |
| | • Seizure | |

Methylmalonic Acidemia Treatment

• Special formula as infant
• Avoid meat, eggs and dairy products
  - Smaller amounts of the amino acids are found in flour, cereal, some vegetables and fruits
• Urine and blood testing
• Without treatment, brain and nerve damage can occur. Acutely, this can cause coma and death
• Even with treatment, some children continue to have problems with health and development
Nursing Care MMA

<table>
<thead>
<tr>
<th>Coordinate Care</th>
<th>Call the doctor</th>
</tr>
</thead>
<tbody>
<tr>
<td>Dietary</td>
<td>Poor appetite</td>
</tr>
<tr>
<td>Genetics</td>
<td>Behavioral changes</td>
</tr>
<tr>
<td>Developmental intervention</td>
<td>Vomiting</td>
</tr>
<tr>
<td></td>
<td>Infection or illness</td>
</tr>
</tbody>
</table>

Educate

Emotional support

Children Living with Inherited Metabolic Disorders (CLIMB)

http://www.climb.org.uk

Nursing Care MMA-Ambulatory

Triage ill calls follow-up apps

Carbohydrate (CHO) Disorders

<table>
<thead>
<tr>
<th>Galactosemia</th>
</tr>
</thead>
<tbody>
<tr>
<td>Pathophysiology</td>
</tr>
<tr>
<td>----------------</td>
</tr>
<tr>
<td>• Missing enzymes that cause the rapid hepatic conversion of galactose to glucose following the ingestion of lactose</td>
</tr>
<tr>
<td>• Enzymes GALK, GALT, Galactose 4, UDP</td>
</tr>
<tr>
<td>• Usually it is a GALT deficiency</td>
</tr>
<tr>
<td></td>
</tr>
<tr>
<td></td>
</tr>
<tr>
<td></td>
</tr>
<tr>
<td></td>
</tr>
</tbody>
</table>
Galactosemia Treatment

Lactose free diet: soy formula as infant and no lactose for life

- MSG, soy sauce
- Butter
- Ice cream
- Pizza
- Dry milk protein, whey, casein, yogurt
- Lactose (many medications are cut with lactose)

Nursing Care Galactosemia

- Coordinate Care
  - Dietary
  - Genetics
  - Developmental intervention
- Educate
- Call the doctor with
  - Poor Appetite
  - Behavioral change
  - Vomiting
  - Infection or illness
- Emotional Support
- Galactosemia Foundation
  - http://galactosemia.org

Nursing Care Galactosemia Ambulatory

- Follow up labs
- Make sure pt has follow up appointment
Urea Cycle Defects
Hyperammonemia

Pathophysiology
• Missing enzymes to convert ammonia and other organic acids to urea

Presentation
• Abnormal newborn screen
• Refusal to eat
• Vomiting
• Drowsiness
• Seizure
• Rapid breathing

Inheritance Pattern
• Inherited autosomal recessive trait
• OTC (Ornithine transcarbamylase deficiency) is inherited as X linked disease
• Incidence: 1: 70,000 births

Urea Cycle Disorder Treatment
• Dietary restriction of protein
• Pharm treatment with sodium benzoate and phenylacetate
  - Reduce ammonia level
  - Arginine or Citrulline
• Dialysis for high ammonia levels
• Liver transplant

Nursing Care Urea Cycle Disorder
• Coordinate Care
  Dietary
  Genetics
  Developmental intervention

Call the doctor with
• Poor appetite
• Behavioral change
• Vomiting
• Infection or illness

Educate
• Emotional support
  National Urea Cycle Disorders Foundation
  www.nucdf.org
Nursing Care Urea Cycle Disorder

During illness the ammonia level can be elevated. Check the labs!!!

Case Study

Birth History: 33 year old mother, G3, P2; insulin dependent due to gestational DM, good BS control, GBS negative. Birth weight 8 lbs.

Family History: Positive for maternal uncle died on the 3rd day of life from unknown cause.

Assessment: 2 day old newborn; rooming with mom. Unable to nurse, sleepy and unresponsive with blood draw. Mild jaundice. Resp reg / rapid 80. Temp 97 Ax, HR 122, Non-reactive to stimuli; Decreased tone; no suck.

Labs: Normal Total Bilirubin, CBC, differential and blood culture.

Treatment: Transfer to NICU-IV D10W; amp and gent given. Within 24 hours he began having seizures.

Differential: Sepsis or IEM

Diagnostic Studies: Ammonia level 1901 mcg / L (less than 50)
- Glutamine 1632 mcg / L (376-709)
- Citrulline Trace (10-45)
- Urine orotic acid 852 mcg / mol creatinine (0.12-3.07)

Diagnosis: Ornithine Transcarbinase Deficiency (OTC deficiency)

Treatment: Remove Protein; Administer Sodium Benzoate, Phenylacetate and Arginine; Hemodialysis

Outcome: Ammonia 70 mcg / L after 36 hours. Neurological status improved.

First Year of Life: 2 metabolic crisis required hospitalization before Liver transplant.
I wanted to write just to thank you so much for everything, every word, every effort, every email...you made this period of my life easier...I don't have enough words to thank you.  

C.C., Mexico

We were so scared and lost before we contacted NUCDF. They personally spent hours on the phone with us answering our questions. My husband and I learned more from a single NUCDF member than our eight month hospitalization did!  

Thank you for always being there for us and giving us the knowledge we need to help our daughter live every day with UCD.

---

### Fatty Acid Metabolism

**Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD)**

<table>
<thead>
<tr>
<th>Pathophysiology</th>
<th>Presentation</th>
<th>Inheritance</th>
</tr>
</thead>
<tbody>
<tr>
<td>MCADD enzyme missing</td>
<td>Abnormal newborn screen</td>
<td>Inherited autosomal recessive trait</td>
</tr>
<tr>
<td>Cannot use certain fat for energy-uses glucose</td>
<td>Extreme sleepiness</td>
<td>Incidence is 1:15,000 babies born in US</td>
</tr>
<tr>
<td></td>
<td>Irritability</td>
<td>1 in every 70 Caucasians is a carrier</td>
</tr>
<tr>
<td></td>
<td>Vomiting</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Diarrhea</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Hypoglycemia</td>
<td></td>
</tr>
</tbody>
</table>

---

**Fatty Acid Metabolism: MCAAD**

[Diagram showing metabolic pathway]
**Treatment of Fatty Acid Metabolism: MCADD**

- Treat and prevent hypoglycemia; avoid fasting; frequent small feeds; IV glucose during treatment
- Restricted LOW fat, high protein, high CHO
- L-Carnitine
- High mortality with the initial episode. In retrospect, some SIDS cases are probably a result of non-ketotic hypoglycemic seizure
- Genetic counseling
- Emotional support

**Nursing Care MCAAD**

**Coordinate Care**
- Dietary
- Genetics
- Developmental Intervention

**Call the doctor with**
- Poor Appetite
- Behavioral Change
- Vomiting
- Infection or illness

**Educate**
- Avoid going long periods without food

**Emotional Support**
- Fatty Oxidation Disorders (FOD) Family Support
  - [http://www.fodsupport.org](http://www.fodsupport.org)
Neonatal Screening

Newborn Screening History

- **Mid 1950's**
  - PKU was the first metabolic disorder known to benefit from early dietary therapy
  - Detecting PKU in all affected infants before irreversible brain damage occurred became the challenge

- **1962**
  - Guthrie developed a simple bacterial assay for phenylalanine that required only a small amount of blood soaked into filter paper
  - Infants in newborn nurseries were routinely checked for PKU

- **2003**
  - As late as 2003—only 4 states tested for only six disorders
  - By April 2011, all states were testing for at least 26 disorders

Federal Newborn Screening Today

- **2014**
  - On December 18, 2014, President Obama signed the Newborn Screening Saves Lives Reauthorization Act of 2014
  - The Act includes new timeliness and tracking measures to ensure newborn babies with deadly, yet treatable disorders, are diagnosed quickly
  - The Act mandates a consent by parents for blood spots used in federally funded research
  - Most states test for 31 disorders
Federal Newborn Screening Today

2015

- Expand newborn screening into Next Generation Sequencing (NGS)
- The National Institute of Child Health and Development is currently funding four 5-year research projects to examine the application of NGS to newborn screening.
- Examine technical feasibility
- Test the medical effectiveness of sequencing in neonatal setting
- Address the ethical, legal and social implications

State Neonatal Screening Programs

- Federally mandated but state run
  While there are recommendations, each state may choose what tests will be included
- Know your state law
  Who is responsible for follow up of abnormal results
  Timing of testing, how it is to be drawn
- False positives and false negatives

Neonatal Endocrine Emergencies

- Thyroid disease
  Hypothyroidism
  Thyrotoxicosis
- Adrenal disorders
  Congenital Adrenal Hyperplasia
  Adrenal insufficiency
- Hypopituitarism
- Hypoglycemia
### Neonatal Thyroid Disease

<table>
<thead>
<tr>
<th>Hypothyroidism</th>
<th>Thyrotoxicosis</th>
</tr>
</thead>
<tbody>
<tr>
<td>Permanent or Transient</td>
<td>Thyrotoxicosis</td>
</tr>
<tr>
<td>Primary or Central</td>
<td>Neonatal Grave's Disease</td>
</tr>
<tr>
<td>Congenital or Acquired</td>
<td></td>
</tr>
</tbody>
</table>

### Question 1

A 12-week old female was born at home and has received no medical care. She has a coarse face, with puffy eyelids, thickened protruding tongue, and thick hair. Her cranial sutures are easily palpable, and posterior and anterior fontanels are open. Her abdomen is protuberant and an umbilical hernia is present. Her skin is cool to touch and mottled. No masses are palpable in the neck.

Of the following, the MOST likely long-term sequelae of this infant’s condition is

A. Cerebral palsy
B. Corneal Opacities
C. Deafness
D. Hydrocephalous
E. Developmental Delay
**Neonatal Hypothyroidism**

<table>
<thead>
<tr>
<th>Pathophysiology</th>
<th>Presentation</th>
<th>Incidence</th>
</tr>
</thead>
<tbody>
<tr>
<td>• Thyroid dysgenesis—85%</td>
<td>• Abnormal Newborn Screen</td>
<td>• 1: 3000 to 4000</td>
</tr>
<tr>
<td>• Sometimes radionuclide</td>
<td>• Birthweight &gt; 4 kg. Gestation &gt; 42 wk.</td>
<td>• Most common congenital endocrine disorder</td>
</tr>
<tr>
<td>scanning studies using I-123</td>
<td>• Umbilical hernia</td>
<td></td>
</tr>
<tr>
<td>determine ectopic, lingual or</td>
<td>• Hypothermia</td>
<td></td>
</tr>
<tr>
<td>sublingual or missing gland</td>
<td>• Jaundice</td>
<td></td>
</tr>
<tr>
<td>• Transient hypothyroidism</td>
<td>• Bradycardia</td>
<td></td>
</tr>
<tr>
<td></td>
<td>• Poor muscle tone</td>
<td></td>
</tr>
<tr>
<td></td>
<td>• Poor feeding</td>
<td></td>
</tr>
<tr>
<td></td>
<td>• Constipation</td>
<td></td>
</tr>
</tbody>
</table>

**Presentation**

- Abnormal Newborn Screen
- Birthweight > 4 kg. Gestation > 42 wk.
- Umbilical hernia
- Hypothermia
- Jaundice
- Bradycardia
- Poor muscle tone
- Poor feeding
- Constipation

**Incidence**

- 1: 3000 to 4000
- Most common congenital endocrine disorder

**Treatment of Neonatal Hypothyroidism**

- Confirm positive diagnosis by newborn screen with serum thyroid levels
- Thyroxine replacement—Initial evaluation and treatment should be done within 2-5 days
- Regular thyroid level blood tests essential—monthly with newborn
- Newborns diagnosed and treated in the first month of life usually have normal intelligence

**Question 2**

A term infant was born GA and develops irritability, jitteriness, and tremors at 7 days of life. Physical exam reveals flushed cheeks, sweating, prominent eyes and hepatosplenomegaly. Axillary temperature is 100.4 F, HR 210 bpm, RR is 48 and BP 88/56. Muscle tone is normal.
**Question 2**

- Of the following the most likely diagnosis is
- A. Congenital heart disease
- B. Familiar dysautonomia
- C. Intrauterine infection
- D. Neonatal Thyrotoxicosis
- E. Neonatal withdrawal

---

**Neonatal Grave's Disease**

<table>
<thead>
<tr>
<th>Pathophysiology</th>
<th>Presentation</th>
<th>Inheritance Pattern</th>
</tr>
</thead>
<tbody>
<tr>
<td>Transplacental passage of TSH receptor stimulating antibody (TSA) from a mother with active or inactive Graves disease</td>
<td>Fetal tachycardia above 160 bpm should be suspicious for fetal Graves disease. In the newborn Grave's disease is manifested by irritability, flushing, tachycardia, hypertension, poor weight gain, thyroid enlargement and exophthalmos</td>
<td>Uncommon due to the low incidence of thyroid toxicosis in pregnancy</td>
</tr>
</tbody>
</table>

---

**Treatment of Fetal / Neonatal disease**

- PTU (Propylthiouracil) and Lugol's Solution (Potassium iodide) decrease the thyroid hormone secretion
- Methimazole and Carbimazole
- Corticosteroids
- Beta blockers for cardiac protection—such as Propanolol to counter the effects of the excessive free T4
- A therapeutic response should be observed within 24-36 hours
- Neonatal Grave's disease resolves spontaneously as the maternal thyroid receptor antibody in the newborn is degraded
- Clinical course is 3-12 weeks
Nursing Care Neonatal Thyroid Disease

Neonatal Adrenal Gland Disorders

- Glucocorticoids (e.g., cortisol)
- Mineralocorticoids (e.g., aldosterone)
- Sex steroids (e.g., testosterone)

Cortex
Adrenal gland
Epinephrine
Norepinephrine

- Adrenal Hypoplasia
  - In the absence of pituitary gland function, the adrenal glands fail to develop normally
  - Severe hypoglycemia can result in death in the first 48 hours of life

Neonatal Adrenal Disorders

- Classic Congenital Adrenal Hyperplasia
  - Salt Losing*
  - Simple Virilizing*
  - Severe hypoglycemia can result in death in the first 48 hours of life

- Adrenal Hypoplasia
  - In the absence of pituitary gland function, the adrenal glands fail to develop normally
  - Severe hypoglycemia can result in death in the first 48 hours of life
American Association for Clinical Chemistry (2015)

**Congenital Adrenal Hyperplasia**

Autosomal recessive. Defect in the synthesis of cortisol and sometimes aldosterone that results in increased ACTH and adrenal hyperplasia. Enzyme 21-hydroxylase (21-OH) is missing or not working.

**Female-Classic**
- Ambiguous genitalia-virilization, large clitoris, labia may be fused and look like scrotum
- High levels of androgens does not usually affect the uterus and ovaries

**Male-Classic**
- Rarely diagnosed at birth unless they have ambiguous genitalia, are salt losers and manifest adrenal crisis, are identified in newborn screen, or have affected sibling

**Adrenal Hypoplasia or Insufficiency**

<table>
<thead>
<tr>
<th>Pathophysiology</th>
<th>Clinical manifestations</th>
<th>Diagnostic Studies</th>
</tr>
</thead>
<tbody>
<tr>
<td>Adrenal hemorrhage</td>
<td>Hyponatremia, hyperkalemia, polyuria, dehydration</td>
<td>Lytes, Glucose, Cortisol (serum and urinary)</td>
</tr>
<tr>
<td>Congenital</td>
<td>Failure to Thrive</td>
<td>Adrenal ultrasound</td>
</tr>
</tbody>
</table>

In the adrenal gland, cholesterol is converted into a precursor called pregnenolone. Then several enzymes complete the production of aldosterone, cortisol, and androgens. A deficiency in the enzyme 21-hydroxylase leads to inadequate amounts of aldosterone and cortisol (green), other substances that do not need the defective enzyme are produced in excess (blue). This enzyme deficiency is inherited and is the most common cause of congenital adrenal hyperplasia (CAH).
Treatment of Adrenal Hypoplasia and Adrenal Insufficiency

- Hydrocortisone (glucocorticoid)
- 9 alphafludrocortisone (mineral corticoid)
- Dietary sodium
- Management of ambiguous genitalia
- Genetic counseling
- Life long management
- In time of crisis-increased steroids
- Psychosocial support

Nursing Care Neonatal Adrenal disorder

- Check the baby!
  - Make sure baby keeps follow up appointments
  - Review stress done at each visit!

Neonatal Hypopituitarism
### Neonatal Congenital Pituitary Disorder

<table>
<thead>
<tr>
<th>Gene mutation</th>
<th>Pituitary agenesis</th>
</tr>
</thead>
<tbody>
<tr>
<td>Holoprosencephaly</td>
<td></td>
</tr>
<tr>
<td>Septo-optic dysplasia</td>
<td></td>
</tr>
<tr>
<td>Other midline defects</td>
<td></td>
</tr>
<tr>
<td>Infection</td>
<td></td>
</tr>
<tr>
<td>Hypovolemic shock</td>
<td></td>
</tr>
</tbody>
</table>

### Neonatal Congenital Hypopituitarism

**Anterior Pituitary**
- Cortisol, Growth Hormone, Gonadotropin, Thyroid deficiencies

**Clinical manifestation**
- Hypoglycemia
- Micropenis
- Jaundice

**Diagnosis**
- Hormone levels
- MRI

**Posterior Pituitary**
- Diabetes Insipidus

**Clinical manifestation**
- High urine output; excess of 5ml/kg/hr
- Low specific gravity
- Dehydration

**Diagnosis**
- Electrolytes
- Osmolality
- Plasma SDH level
- MRI
Treatment of Pituitary Disorder

- The immediate goals of management are to stabilize the neonate’s blood sugar and ensure that the neonate is not at risk of life-threatening cortisol deficiency
- Hypoglycemia may not resolve without growth hormone treatment
- Correct the specific hormone deficiencies

Nursing Care Neonatal Pituitary Disease

Take-away

- Competencies for non-genetics health care provider
  - Published by the National Coalition for Health Professionals Education in Genetics (NCHPEG, 2007)
  - At minimum every health care professional should be able to:
    - Examine their own competence and identify areas of strength and opportunity for growth
    - Understand that health related genetic information can have social and psychological implications for individuals and families
    - Know when to make a referral to a genetics professional
Take-away

- Knowledge
  - Basic terminology
  - Patterns of inheritance
  - Difference between diagnosis and predisposition to disease
  - The potential limitations, and risks of genetic information

- Skills
  - Family history taking, explain the benefits of genetic services, use of credible resources
  - Seek coordination and collaboration with an interdisciplinary team of health professionals
  - Provide education, care and support

Achieve the best outcome

The nurse should make continued, repeated assessments of the neonate in the acute care setting

The nurse should track the labs in the acute and outpatient setting

Coordinate care

“To watch the infant form with anxious care
The lurking symptoms of disease detect,
And with the aid of sweet nutritious food,
Or potent herb, or kindly drug, to aid
Oppressed nature in her arduous task
Be thine! And thine the grateful rich reward
Of conscious duty done -- a mead more fair
Than all the laurels which bedeck the brow
Of modern Caesar”.

Isaac Riley MD, 1811
Questions

References


References


Pola, Nicole, Mandell, Barbara. Caring for Children and their families. 3rd Edition


