



NEWBORN SCREENING (NBS)

Newborn screening (NBS)

- Procedure to determine if the newborn infant has a heritable congenital metabolic disorder that may lead to serious physical health complications, mental retardation, and even death if left undetected and untreated

History of NBS in the Philippines

- **1996 – Initiated in the Philippines through PPS/ POGS “Philippine Newborn Screening Project” with 24 accredited hospitals**
- **1998 – G6PD was added to the list of disorders and homocystinuria was deleted**
- **1999 – DOH included NBSP in the CHILD 2025 Program**
- **2001 – DOH created the National Technical Working Group for the nationwide implementation of NBSP**
- **2004 – NBS was integrated into the public health delivery system with the enactment of RA 9288 or “Newborn Screening Act of 2004**
 - 6 Congenital Metabolic Disorders
- **2014 – Expanded newborn screening was implemented**
 - 22 more disorders were added (hemoglobinopathies and additional metabolic disorders)

Objectives of NBS



- **Newborn has access to newborn screening**
- **Sustainable newborn screening system**
- **All health practitioners are aware of the advantages**
- **Parents recognize their responsibility**

Newborn Screening Act of 2004 (RA 9288)

- Protect the rights of children to survival and full and healthy development as normal individuals
- Provide for a comprehensive, integrative and sustainable national newborn screening system to ensure that every baby born in the Philippines is offered the opportunity to undergo newborn screening and be spared from heritable conditions

Components of Comprehensive NBS System

- **Education of relevant stakeholders**
- **Collection and biochemical screening of blood samples taken from newborns**
- **Tracking and confirmatory testing to ensure the accuracy of screening results**
- **Clinical evaluation and biochemical/ medical confirmation of test results**



Components of Comprehensive NBS System

- Drugs and medical/
surgical management
and dietary
supplementation to
address the heritable
conditions**
- Evaluation activities to
assess long term
outcome, patient
compliance and quality
assurance**



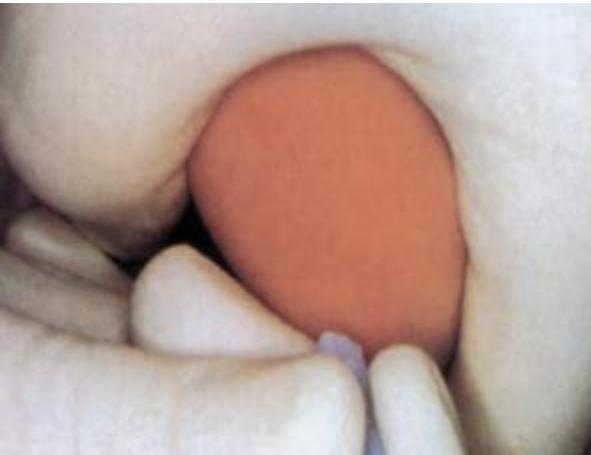
Performance of Newborn Screening

- Ideal time: 48 hours to 72 hours after birth
- May also be done 24 hours after birth
- High risk newborn in NICU may be exempted from the 3-day requirement but must be tested by 7 days

Note: (+) result, repeat test 14 days after

Blood Specimen Collection Procedure

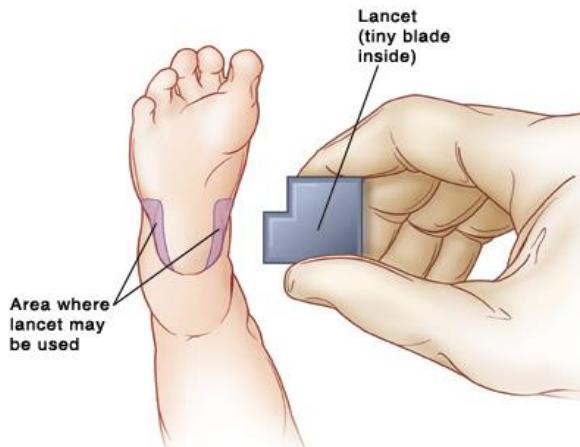
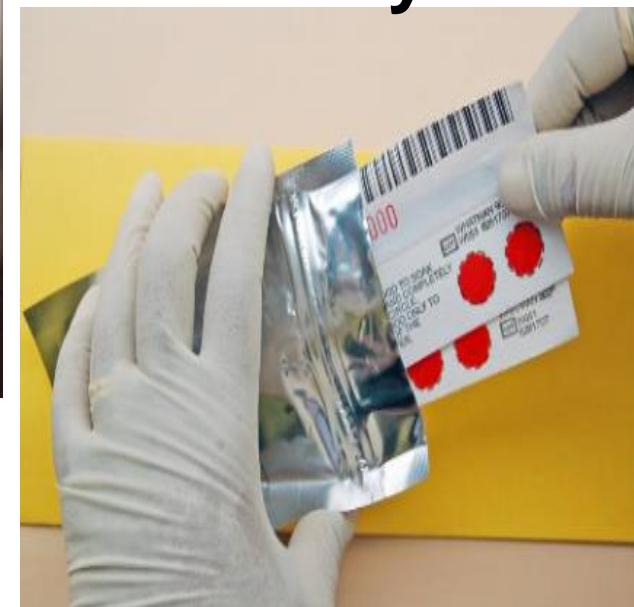
Puncture heel



Lightly touch filter paper to **LARGE** blood drop

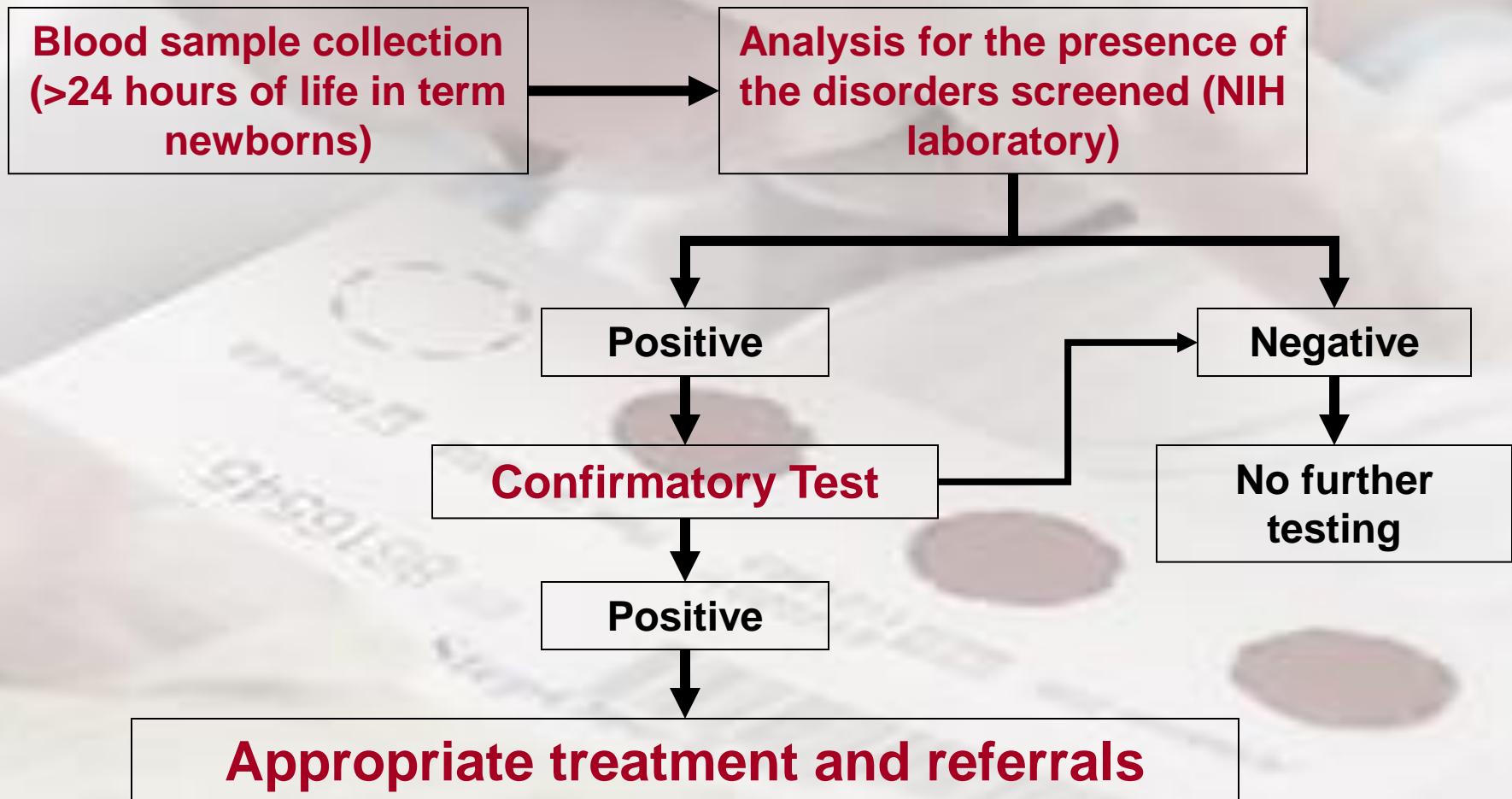


Dry the sample & send to the laboratory



Area where
lancet may
be used

NBS Screening Procedure





Obligation of healthcare provider

- Parents and practitioners have joint responsibility to ensure that NBS is performed
- Refusal of testing on grounds of religious belief shall be written for



HERITABLE CONDITIONS:

CONGENITAL HYPOTHYROIDISM

- Endocrine disorder also referred to as cretinism or dwarfism
- Results from the absence or lack of development of thyroid gland causing absence or lack of thyroxine needed for metabolism and growth of the body and the brain;
- Is not initiated within 4 weeks
- The baby's physical growth will be stunted and he may suffer from irreversible mental retardation



Untreated Congenital Hypothyroidism



- Jaundice
- Poor feeding
- Hypotonia
- Macroglossia (large tongue)
- Large fontanelles, delayed closure
- Coarse facial features
- Mental retardation
- Short stature

CONGENITAL HYPOTHYROIDISM

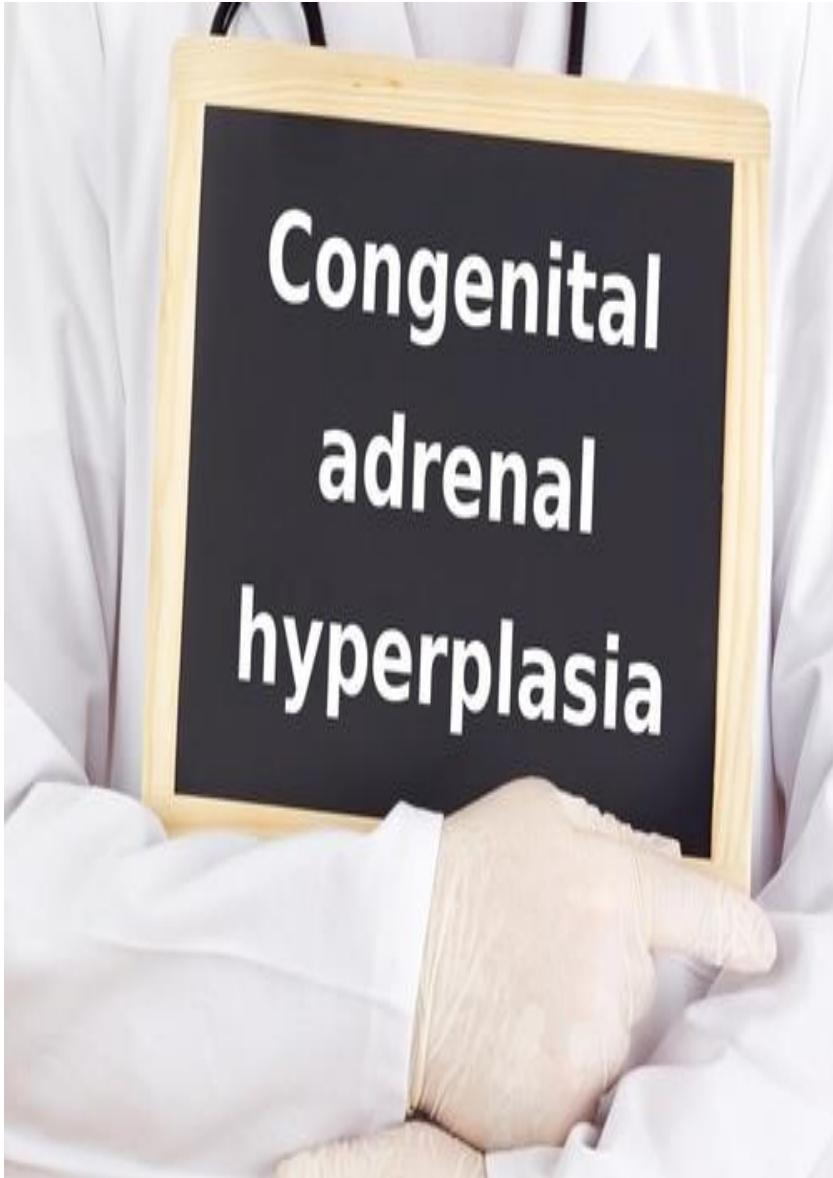
TREATMENT:

- Lifelong thyroid hormone replacement therapy (as soon as possible after diagnosis) as a single morning dose
- DOC: Synthetic Levothyroxine (Synthroid, Proloid and Levothroid)
- If treatment started early: normal physical growth and intelligence

NURSING CARE MANAGEMENT

- Early identification
- Lifelong treatment - compliance with drug regimen (Levothyroid and Synthroid)
 - Tasteless, can be crushed and mixed
 - If dose missed: double next dose

OVERDOSE	INADEQUATE TREATMENT
+ Rapid pulse + Dyspnea + Irritability + Insomnia + fever, sweating Weight loss	+ fatigue + sleepiness Decreased appetite Constipation

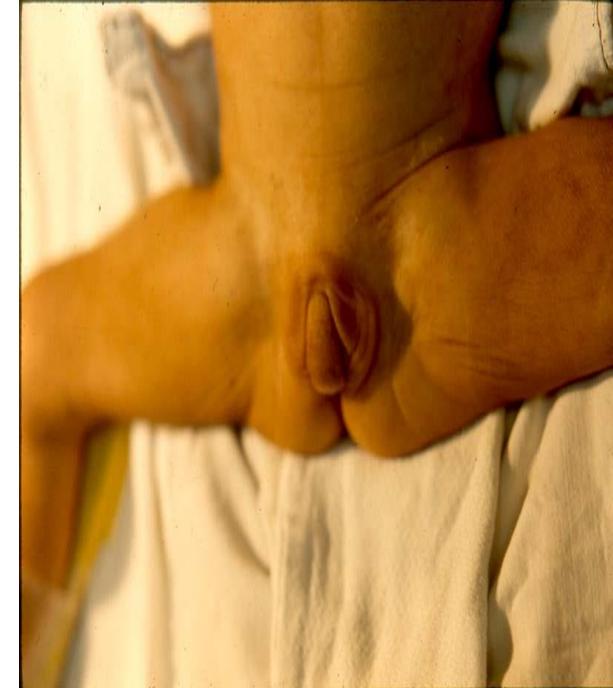


- An endocrine disorder caused by an inborn defect in the biosynthesis of adrenal cortisol that causes severe salt or sodium losses, dehydration and abnormally high levels of male sex hormones in both boys and girls

CONGENITAL ADRENAL HYPERPLASIA

Symptoms:

- Begin shortly after birth:
 - Anorexia
 - Progressive weight loss
 - Vomiting
 - Dehydration
 - Disturbances in cardiac rate and rhythm
 - Cyanosis
 - Dyspnea



Ambiguous genitalia in females

If not treated early, babies may die within a few weeks

CONGENITAL ADRENAL HYPERPLASIA

Treatment:

- Continued hormonal replacement of **HYDROCORTISONE**
 - Glucocorticoid replacement therapy
- Mineralocorticoid therapy (Salt-wasting form)



Ambiguous genitalia in females

PHENYLKETONURIA

- Inborn error of metabolism characterized by lack of enzyme phenylalanine hydroxylase (needed to breakdown phenylalanine → elevated serum phenylalanine → brain damage and mental retardation)
- Late physical signs reflect the absence of adequate melanin pigment: blond hair, fair skin and blue eyes



THERAPEUTIC MANAGEMENT

- Restriction of dietary protein
- Maintain safe range of phenylalanine (2-8mg/dl)
 - Brain damage: 11-15mg/dl
- Meet child optimum level for growth
- Special milk substitute + tyroxine
- + Breastmilk low protein
- Low phenylalanine diet throughout life
- 93% Mental retardation, 72% Microcephaly
- No high protein and dairy products
- Pregnant mother should be placed in low phenylalanine diet

NURSING CARE MANAGEMENT

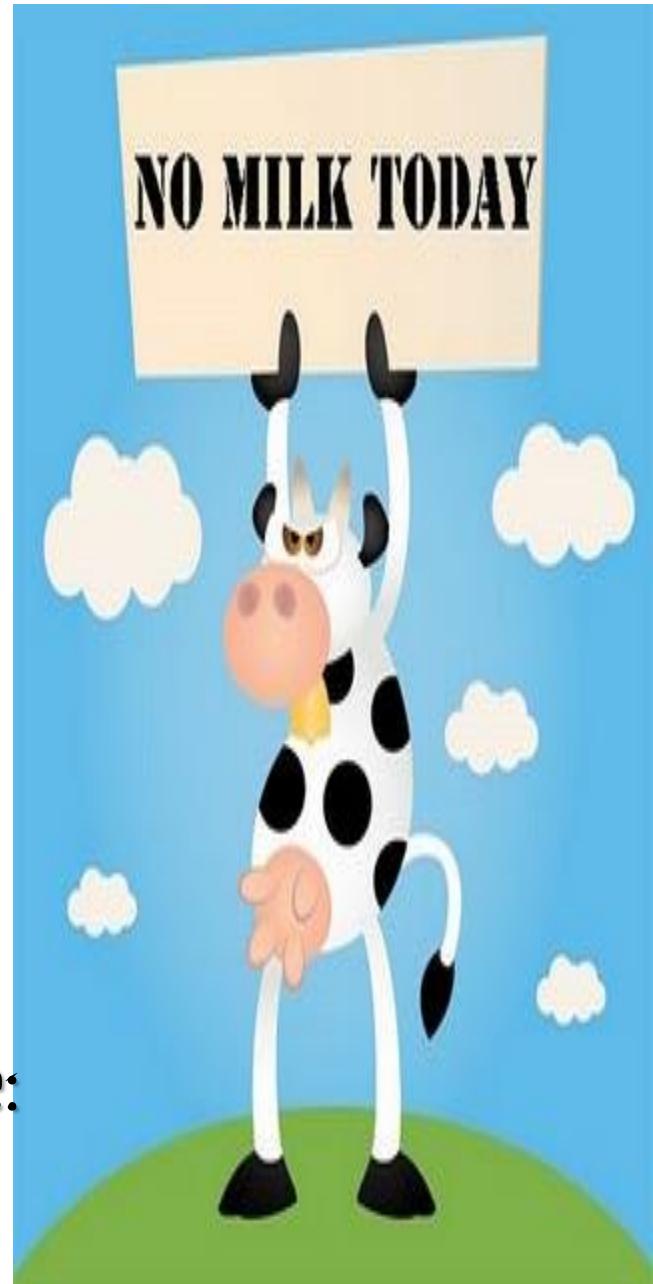
- Diet restriction
- Peer pressure - temptation in food
- Involve in menu plan, reward - child
- Support family - express feelings
- Monitor physical, neurological, and intellectual development

GALACTOSEMIA

Type 1: deficient Galactokinase → inability to convert galactose to glucose → galactosemia
→ galactosuria

Complications: mental deficiency, cataracts and death

Dietary treatment: galactose-free diet (galactose: high in milk and milk products)



GALACTOSEMIA

Type 2: "Classic" galactosemia

— Serious deficiency of Uridyl Transferase



Early symptoms: jaundice, vomiting, enlarged liver and spleen
hypoglycemia, convulsions and feeding difficulties

Complications: liver cirrhosis and irreversible mental retardation

Dietary treatment: exclusion of galactose from the diet to prevent severe liver cirrhosis, mental retardation, cataracts and recurrent hypoglycemia

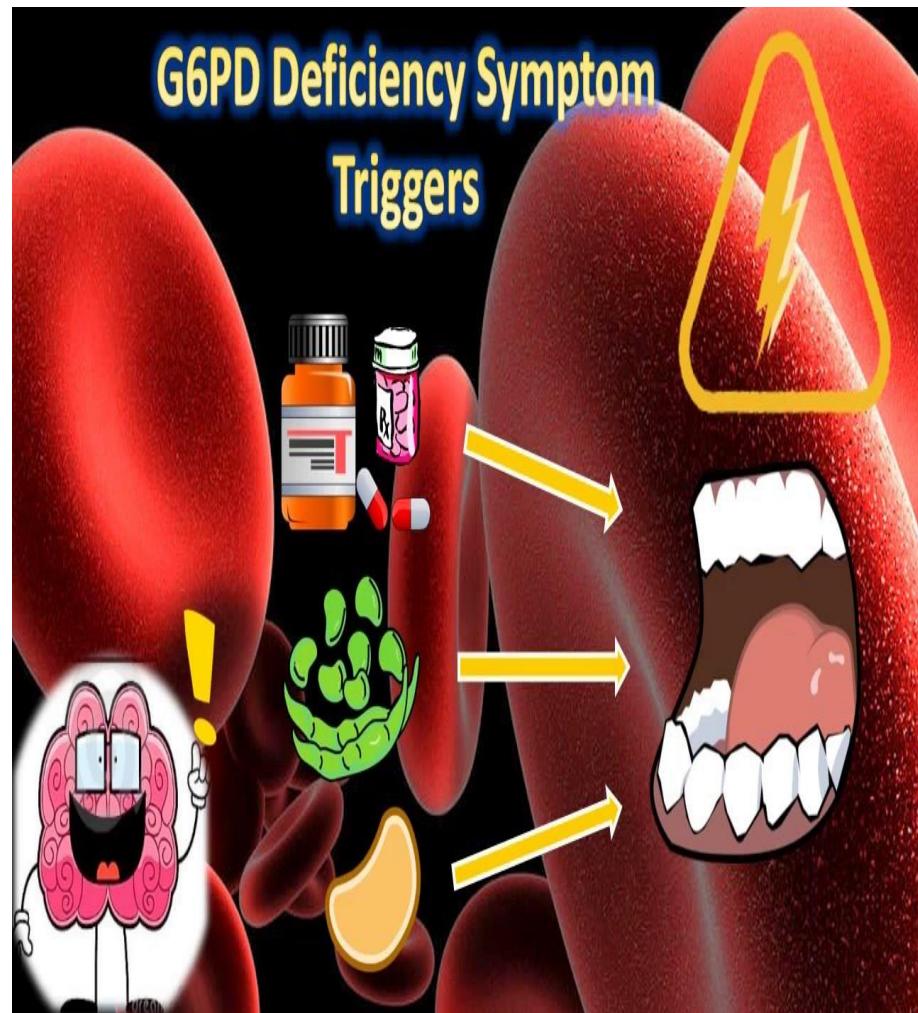
GLUCOSE 6 PHOSPHATE DEHYDROGENASE (G6PD) DEFICIENCY

- Deficiency in G6PD
- Red blood cells lack protection from the harmful effects of oxidative substances found in drugs, foods, beverage
- Severe anemia and hyperbilirubinemia → kernicterus (jaundice of the brain) and mental retardation, convulsion, coma and even death

Without G6PD, RBC's undergo HEMOLYSIS when exposed to oxidative stress!

OXIDATIVE AGENTS LEADING TO HEMOLYSIS IN G6PD DEFICIENCY

- Drugs
 - Sulfonamides, quinolones, chloramphenicol, Vitamin K
- Chemicals
 - Mothballs
- Food
 - Fava beans
- Infection



MAPLE SYRUP URINE DISEASE (MSUD)

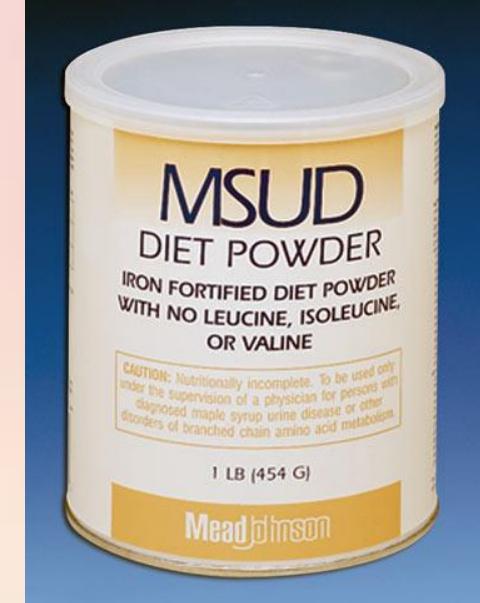
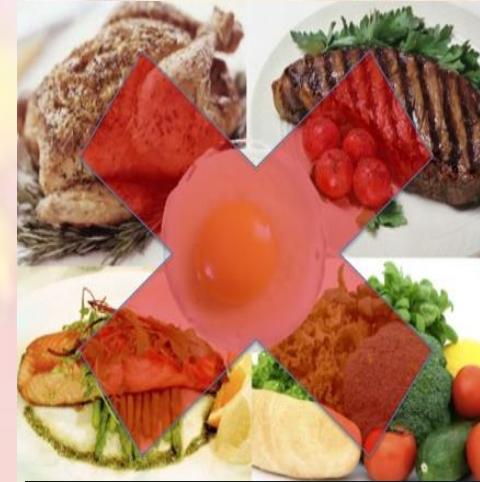
- An inherited disorder in which the body is unable to process certain protein building blocks (amino acids) properly.
- Inherited in an autosomal recessive pattern (inherit two mutated genes, one from each parent)
- Symptoms: Distinctive sweet odor of infant's urine, poor feeding, vomiting, lack of energy (lethargy), and developmental delay.
 - If untreated, will lead to seizures, coma, and death.



MAPLE SYRUP URINE DISEASE (MSUD)

Dietary treatment:

- Protein-free diet;
- Infants have a diet formula with low levels of the amino acids leucine, isoleucine, and valine;
- IV administration of amino acids that don't contain branched-chain amino acids, combined with glucose for extra calories;



STANDARD 6- TEST

Disorder Screened	Effects if NOT SCREENED	Effects if SCREENED and TREATED
CH	Severe Growth and Mental Retardation	Normal
CAH	Death	Alive and normal
GAL	Death or Cataracts	Alive and normal
PKU	Severe Mental Retardation	Normal
G6PD Deficiency	Severe Anemia, Kernicterus	Normal
MSUD	Death or Mental retardation	Alive and normal

EXPANDED NBS

Disorder Screened	Effects if NOT SCREENED	Effects if SCREENED and MANAGED
ORGANIC ACID DISORDERS	Developmental delay Breathing problems Neurologic damage Seizures Coma Early death	Alive Most will have normal development with episodes of metabolic crisis
FATTY ACID OXIDATION DISORDER	Developmental and physical delays Neurologic impairment Sudden death Seizures Coma Enlargement of the heart and liver Muscle weakness	Usually healthy in between episodes of metabolic crises Alive

EXPANDED NBS

Disorder Screened	Effects if NOT SCREENED	Effects if SCREENED and MANAGED
HEMOGLOBINOPATHIES	Painful crises Anemia Stroke Multi-organ failure Death	Alive Reduces the frequency of painful crises May reduce the need for blood transfusions
AMINO ACID DISORDER	Mental retardation Coma and death from metabolic crisis	Alive Normal growth Normal intelligence for some learning problems to others

CLINICAL MANIFESTATIONS AT BIRTH

<i>DISORDER</i>	<i>APPEARANCE AT BIRTH</i>
CAH	Hyperpigmentation
	Ambiguous Genitalia in female infants
CH	Normal
GAL	Normal
PKU	Normal
G6PD Deficiency	Normal
MSUD	Normal

When do typical signs and symptoms appear?

DISORDER	GOLDEN PERIOD
CAH	7-14 days
CH	4 weeks
Gal	2 weeks
PKU	3 weeks
G6PD deficiency	On exposure to specific agents causing hemolysis
MSUD	12-24 hours after birth or 2-3 days

SUMMARY: TREATMENT

Disorder	Treatment	
CAH	Supplementation	Glucocorticoid, mineralocorticoid, NaCl
CH	Supplementation	Thyroid Hormone
GAL	Avoidance	Galactose, Lactose
PKU	Avoidance	Protein diet
G6PD Deficiency	Avoidance	Oxidative drugs, food and chemicals
MSUD	Avoidance Supplementation	Protein diet Amino acids without branched-chain AA/ Glucose for calories



NEWBORN HEARING SCREENING

- Two different tests can be used to screen for hearing loss in newborns.
- Both tests are quick (5-10 minutes), safe and comfortable with no activity required from the newborn.



Newborn Hearing Screening:
What do the results mean?

The infographic is titled "Newborn Hearing Screening: What do the results mean?". It features two panels. The left panel shows a woman holding a baby and is labeled "Baby is born". An arrow points from this panel to the right panel, which shows a woman holding a baby and is labeled "Hearing screening is performed at 24-48 hours of age or before the baby leaves the hospital".

Baby is born

Hearing screening is performed at 24-48 hours of age or before the baby leaves the hospital

NEWBORN HEARING SCREENING

Otoacoustic Emissions (OAE) Test

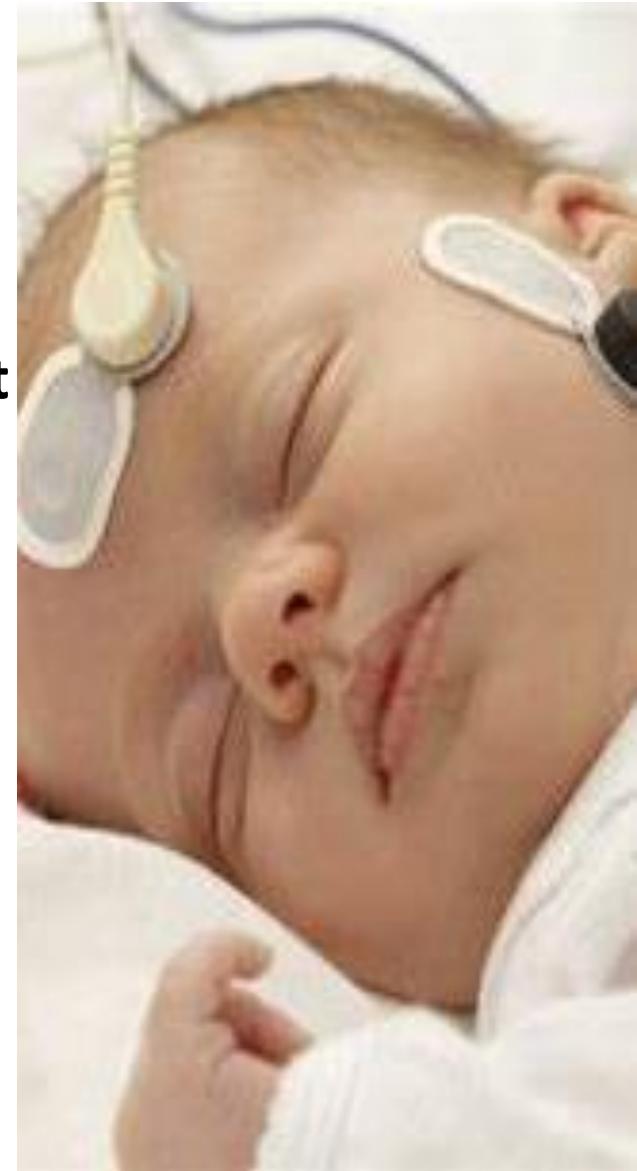
- Used to determine if certain parts of the newborn's ear respond to sound.
- During the test, a miniature earphone and microphone are placed in the ear and sounds are played. When a newborn has normal hearing, an echo is reflected back into the ear canal, which can be measured by the microphone. If no echo is detected, it can indicate hearing loss.



NEWBORN HEARING SCREENING

Auditory Brain Stem Response (ABR) Test

- Used to evaluate the auditory brain stem and the brain's response to sound.
- During the test, miniature earphones are placed in the ear and sounds are played. Band-Aid-like electrodes are placed along the newborn's head to detect the brain's response to the sounds. If the newborn's brain does not respond consistently to the sounds, there may be a hearing problem.





Thank You!