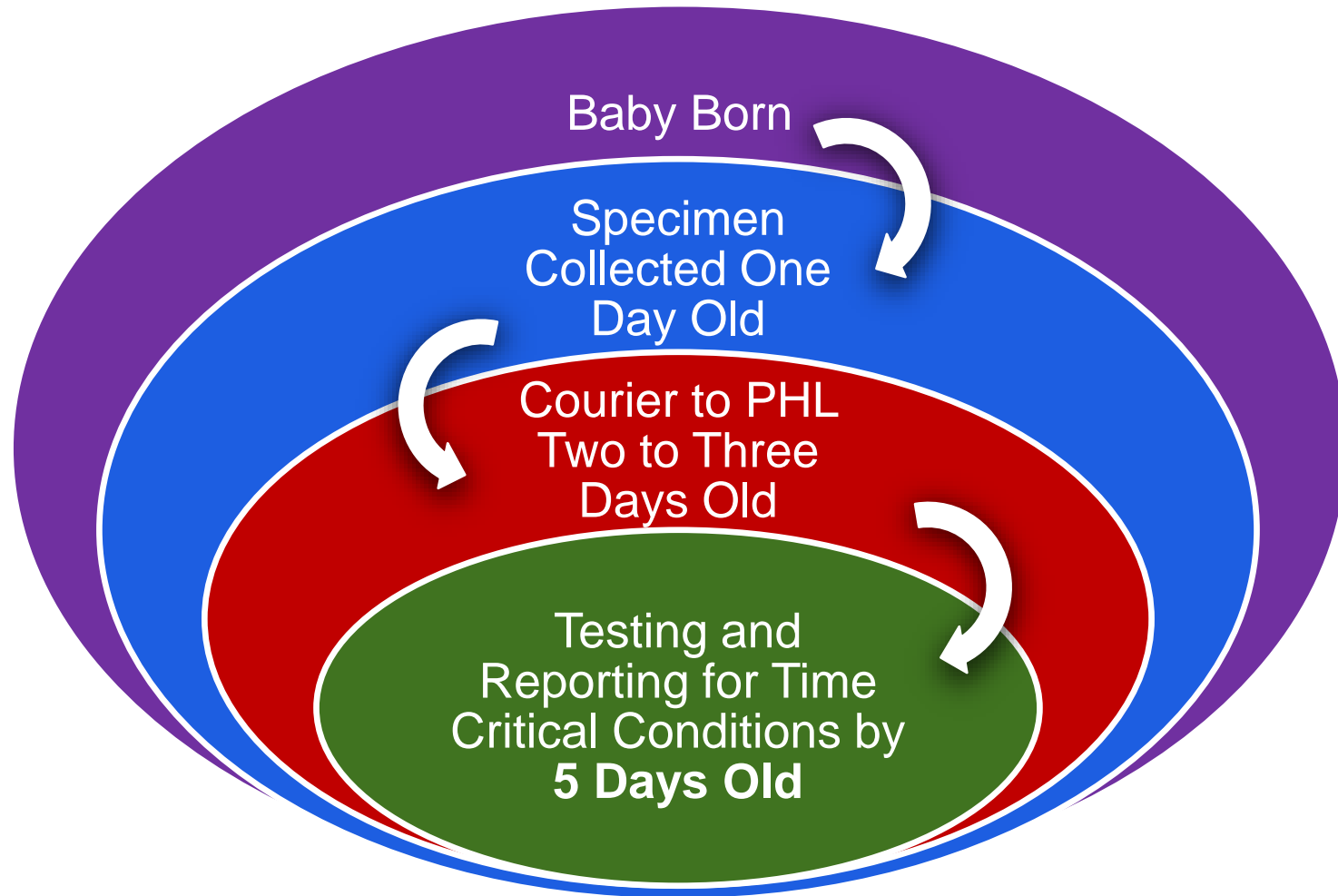


Life Saving Goals of Newborn Screening

Federal recommendations say to save babies we must meet this timeline:



- 54 different conditions or hidden disorders
 - Baby will usually look normal and healthy at birth
 - Newborn screen is only way to know if they may get sick or die
 - Once baby starts to get sick – damage may not be reversible
- Some babies only have a few hours before they start to get very sick
- Some will die within the first few days of life without our help
- 90 to 100 babies identified with condition each year
- It takes all of us working together as a team to save lives
 - Hospital staff - collect the specimen and have it ready
 - Courier staff - pick up specimen and take to PHL ASAP
 - Lab staff – test and report
 - Nurses at Health Department – contact Pediatrician & Specialist
 - Pediatrician & Specialist – confirm disorder & get baby on treatment

Amino Acid Disorders

Time Critical:
Baby can die at
3 days old

Signs and Symptoms:
seizures, low muscle tone, lethargy, coma, developmental delay, ketosis, too much ammonia in blood, low blood sugar, and metabolic acidosis

Biotinidase Deficiency

Treatment will make baby feel a lot better

Signs and Symptoms: no energy, low muscle tone, seizures, lack of control of body movements, and patchy red skin

Classic Galactosemia

Time Critical:
Every day untreated increases amount of mental and/or physical disability

Signs and Symptoms: vomiting, diarrhea, yellow skin and whites of eyes, cataracts, liver failure, and wide spread infection

Congenital Adrenal Hyperplasia

Time Critical:
Days matter - especially boys

Signs and Symptoms: infant's external genitals don't appear male or female, salt loss, and blood pressure issues

Congenital Hypothyroidism

Need treatment within 2 weeks of life

Signs and Symptoms: larger than normal baby, enlarged head, skeletal delay, hoarse cry, dry skin, and constipation

Cystic Fibrosis

Early treatment can help with lung problems and digestion

Signs and Symptoms: coughing, wheezing, respiratory infections, and poor weight gain

Fatty Acid Disorders

Time Critical: Babies can die in their sleep within a few days of life

Signs and Symptoms: low ketones and low blood sugar during fasting, liver failure, coma, and low muscle tone

Sever Combined Immuno-deficiency

Need bone marrow transplant by 3 months of age - takes a while to find a match

Signs and Symptoms: Infections that won't go away, yeast infection in the mouth and throat

Hemoglobinopathies

Treatment to prevent infections and pain is needed

Signs and Symptoms: blocked or damaged blood vessels, respiratory failure, stroke, fever, low # of red blood cells, wide spread infections, temporarily stop making red blood cells

Organic Acid Disorders

Time Critical: Babies can die in their sleep within a few days of life

Signs and Symptoms: vomiting, coma seizures, low blood sugar, low muscle tone, loss of control of body movements, and death while sleeping.