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Updates on Newborn Screening in Oklahoma

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OKLAHOMA
State Department of Health

Public Health Laboratory

- Transition to Stillwater
- Effective 3/16
  - Contracted courier service picks up specimens from birthing facilities and CHDs
  - Specimens submitted via mail should be mailed to the new Stillwater address to avoid delays
  - All newborn screening specimens are being received in the PHL and then sent to PerkinElmer Genetics via FedEx for testing
  - PDF image of PerkinElmer's StepOne report is available for download
  - Abnormal results are reported to NBS Follow-up Program
    - Time critical conditions are now reported 24/7
  - After testing is complete the specimens are return to the PHL
  - Specimens are destroyed around day 42 per PHL protocol
- OK NBS Result Report updated
- Newborn Screening Portal updated

NBS Expansion

- New Conditions
  - Pompe
  - Mucopolysaccharidosis Type I (MPS I)
  - Spinal Muscular Atrophy (SMA)
  - X-Linked Adrenoleukodystrophy (X-ALD)

- Benefits
  - Identification of infants who are at risk
  - Evaluation by a specialist
  - Confirmatory testing
  - Treatment
  - Improved health outcomes

Pompe

- Pompe is a deadly, progressive, neuromuscular disorder
- Mutations in the gene that makes the enzyme α-glucosidase (GAA) which breaks down glycogen (complex sugar)
  - accumulation of glycogen in certain organs and tissues, especially muscles, impairs their ability to function normally
- Estimated 1-2 infants will be identified annually

Mucopolysaccharidosis Type I (MPS I)

- Mucopolysaccharidosis Type I is a deadly multi-system disorder
- Mucopolysaccharides or glycosaminoglycans (GAGs) function as lubricants, shock absorbers, and have bactericidal properties
  - Components of cartilages and ligaments, some are involved in support and motor function
- As GAGs age, they are broken down in lysosomes and their components are recycled
- Individuals with MPS I are missing the enzyme alpha-L-iduronidase (IDUA) needed to break down mucopolysaccharides or GAGs
- Estimated 1 infant will be identified every two years

Spinal Muscular Atrophy (SMA)

- SMA is a neuromuscular disease
  - Progressive degeneration of motor neurons in the spinal cord and brainstem
    - Weakness and atrophy of muscles used for crawling, walking, sitting up, and controlling head movement
    - Severe case—muscles used for breathing and swallowing are affected
  - Mutation in the survival motor neuron (SMN1) gene which leads to a shortage of the SMN protein
  - Motor neurons die and nerve impulses are not passed between the brain and muscles
- Estimated 5-8 infants will be identified annually

X-Linked Adrenoleukodystrophy (X-ALD)

- X-ALD is a peroxisomal disorder
  - Nervous system and adrenal glands are affected
  - Myelin, the fatty covering that insulates nerves in the brain and spinal cord is prone to deterioration or demyelination
    - Demyelination reduces the ability of nerves to relay information correctly to the brain
  - Damage to the outer layer of the adrenal gland causes a shortage of certain hormones known as adrenocortical insufficiency
    - May cause weakness, weight loss, skin changes, vomiting and coma
  - Mutations in the ABCD1 gene results in a shortage of adrenoleukodystrophy protein (ALDP)
    - Transport and subsequent breakdown of very long chain fatty acids (VLCFAs) is disrupted, resulting in high levels of these fats in the body
    - Accumulation of VLCFAs can be toxic to the adrenal cortex and myelin
      - Triggers an inflammatory response in the brain, which could lead to a breakdown in myelin
- Estimated 1-2 infants will be identified annually

Questions

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