# Hearing Briefing: Newborn Screening

# Chapter 246-650 WAC

## Adding OTCD, GAMT, and ARG1-D

Kelly Kramer, Policy Advisor - August 7, 2024

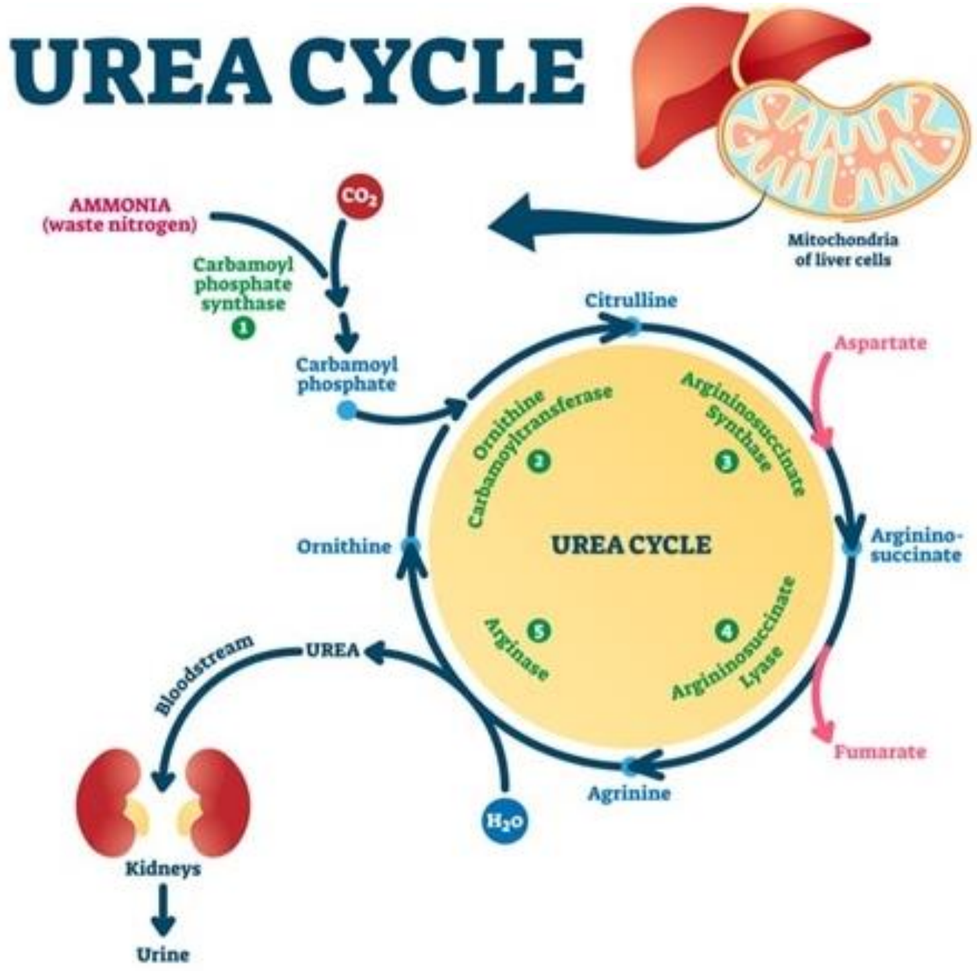
WASHINGTON STATE BOARD OF HEALTH

# Briefing Agenda

- Condition Overviews
- Rulemaking Progress
- Proposed Rule Changes
- Next Steps and Timeline

# Ornithine Transcarbamylase Deficiency (OTCD)

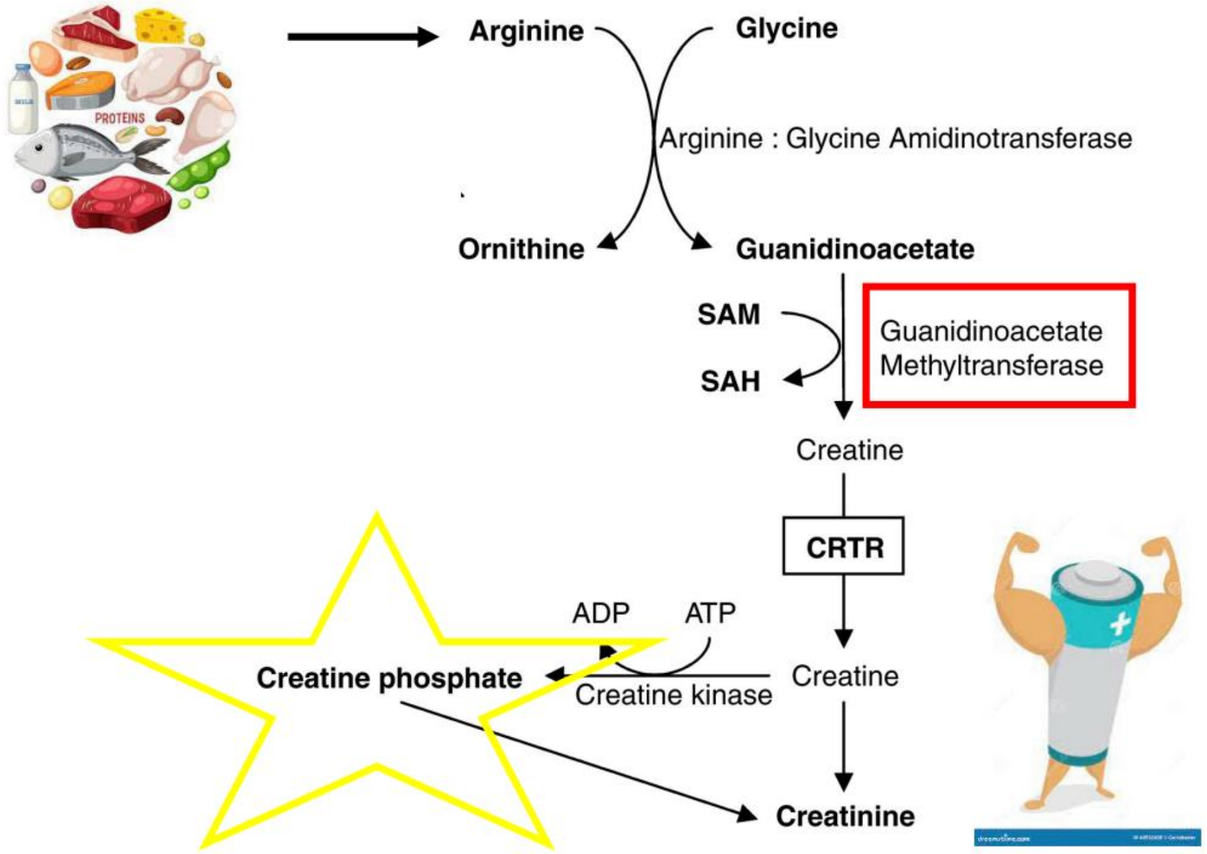
- Inherited urea cycle disorder.
- X-linked recessive inheritance pattern (more likely to affect male babies).
- Prevalence: ~1:56,000.
- Interferes with the body's ability to process ammonia (or nitrogen waste).
- High levels of ammonia are toxic to the brain and nervous system, and can lead to:
  - Tiredness/sluggishness
  - Sepsis-like symptoms
  - Comas
    - Developmental delays
    - Death




Source: News Medical, The Urea Cycle Step-by-Step

# Guanidinoacetate Methyltransferase (GAMT) Deficiency

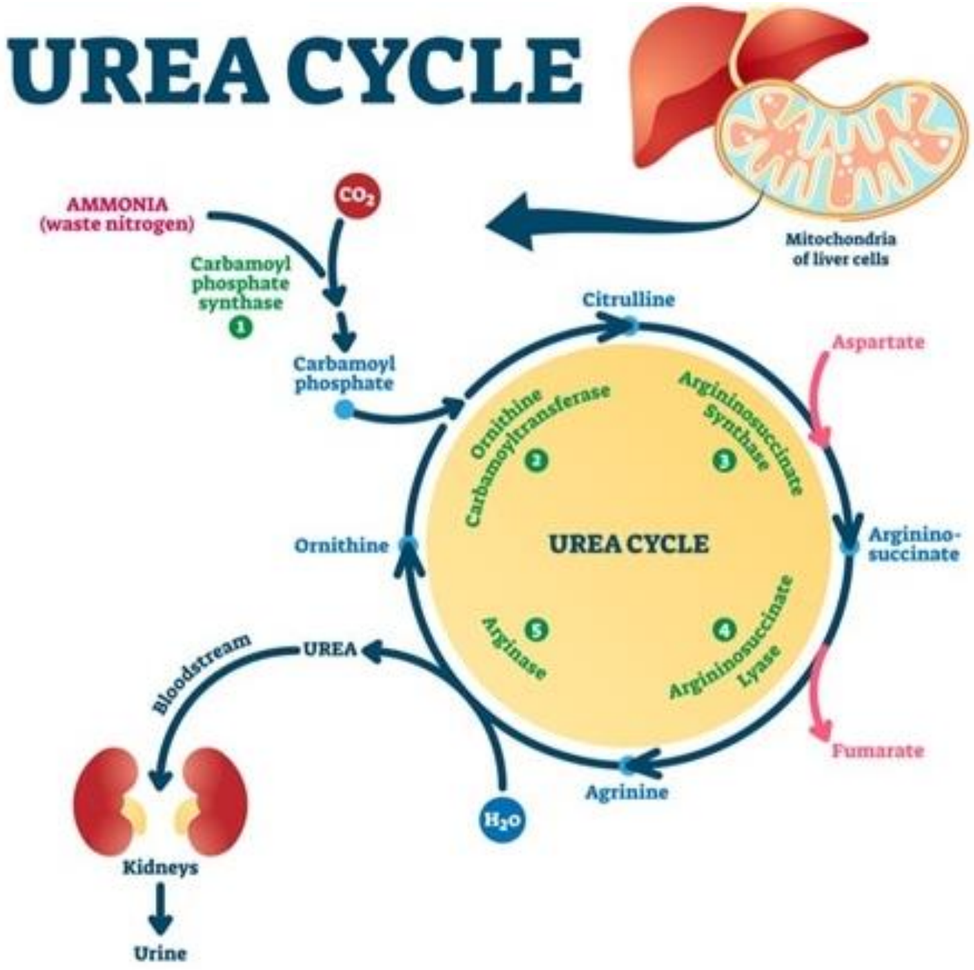
- Inherited disorder that prevents the body from metabolizing and transporting creatine.
- Autosomal recessive inheritance pattern (affects male and female babies equally).
- Prevalence: About 130 individuals diagnosed worldwide.
- Signs and symptoms of the condition vary; can start anywhere from 3 months to 2 years of age.
- If untreated, GAMT Deficiency leads to developmental delays and cognitive impairments (can be moderate or severe).




Source: Shelkowitz, E. GAMT Deficiency: Natural History, Diagnostic Testing and Treatment. SBOH NBS TAC Meeting. September 2023.

# Arginase 1 Deficiency (ARG1-D)

- Inherited urea cycle disorder that causes the amino acid arginine and ammonia to accumulate in the blood.
- Autosomal recessive inheritance pattern (affects male and female babies equally).
- Prevalence: Less than 260 individuals diagnosed worldwide.
- Symptoms start off subtle and do not become apparent until early childhood (1-3 years of age). May include:
  - Spasticity in lower extremities (most common, affects 80 -90% of those diagnosed)
  - Slowed cognitive development
  - Seizures
  - Stunted growth
  - Challenges with eating
  - Liver problems




Source: News Medical, The Urea Cycle Step-by-Step

# Screening and Treatment Available

## Screening

- All three conditions can be screened using tandem mass spectrometry technology (MS/MS).
  - Uses one 1/8” hole punch from a dried blood spot to test for 19 congenital disorders at the same time.
  - Technology used in the WA Newborn Screening Program since 2004.

## OTCD Treatment

- Emergency treatment of high blood ammonia levels
- Dietary modifications
- Liver transplant

## GAMT Treatment

- Dietary modifications and oral supplements (creatine, sodium benzoate, and/or ornithine).
- Physical, occupational, speech, and behavioral therapy.

## ARG1-D Treatment

- Dietary modifications and oral supplements.
- Ammonia diversion therapy: nitrogen scavenging medications (sodium benzoate, sodium phenylbutyrate).
- Referrals to neurology and physical, occupational, and/or speech therapy.

# Rulemaking Progress - OTCD

- In 2020, the Board approved the Department of Health (Department) to form a Technical Advisory Committee (TAC).

- June and July 2021, the TAC convened to review the condition.
  - The TAC voted to recommend the inclusion of OTCD to the NBS panel.

- October 2021, Board staff presented TAC recommendations to the Board.
  - Approved motion to initiate rulemaking

- February 2022, CR-101 filed.

- Legislature did not fund the Department's requests in the 2022 and 2023 sessions for funding to increase the NBS fee.
  - Rulemaking to add OTCD has been put on hold since.

# Rulemaking Progress – GAMT and ARG1-D

- March and April 2023 Board meetings, the Board approved Board staff and the Department to convene a TAC.
- September 2023, the TAC convened to review the conditions for inclusion on the newborn screening panel.
  - The Technical Advisory Committee voted to recommend the inclusion of GAMT and ARG1-D to the panel.
- October 2023, Board staff presented Technical Advisory Committee recommendations to the Board.
  - The Board approved a motion to initiate rulemaking for the inclusion of GAMT and ARG1-D to the panel.
- November 2023, Board staff filed CR-101.

# Newborn Screening Fee Increase

- Estimate for all three conditions is **$1.77** per baby.
- Fee increase includes salaries/benefits for lab and follow-up, cost of reagents, and biochemical genetics consulting.

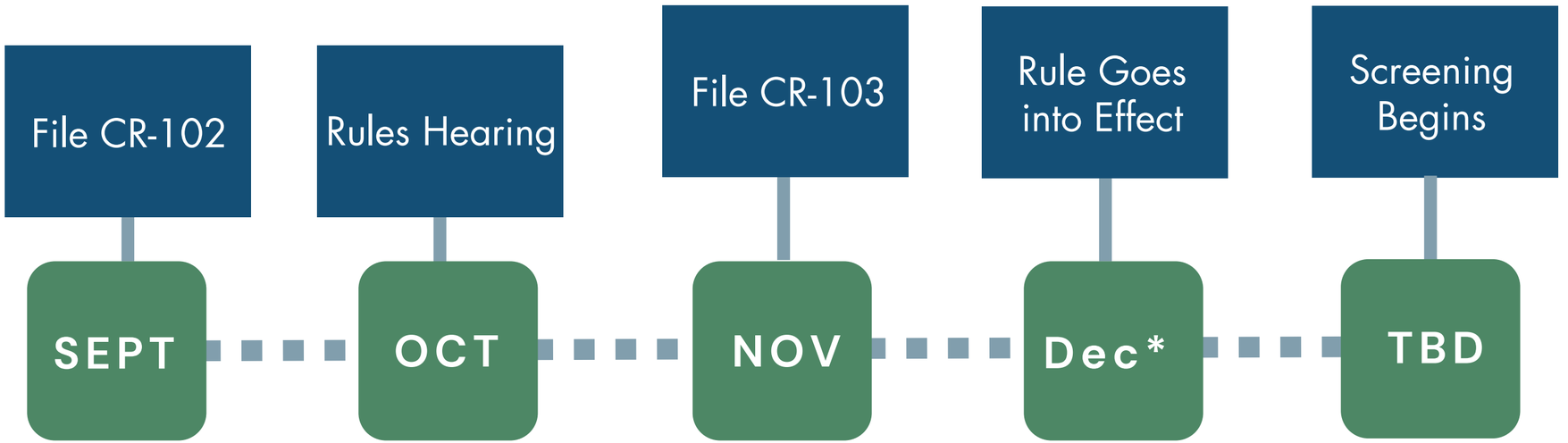
# Proposed Changes:
# WAC 246-650-010, Definitions

- Adding a definition Ornithine Transcarbamylase Deficiency (OTCD).
- Adding a definition Guanidinoacetate Methyltransferase Deficiency (GAMT).
- Adding a definition Arginase 1 Deficiency (ARG1-D).

# Proposed Changes: WAC 246-650-020, Performance of Screening Tests

- Amending the list of newborn screenings to be performed from a dried blood specimen to include OTCD, GAMT, and ARG1-D.
- Renumber to retain alphabetical nature of the list.

# Next Steps




*December or a date otherwise specified by the Department's newborn screening team. Update on screening start by the October rules hearing.

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