WASHINGTON STATE BOARD OF HEALTH

Washington State Department of HEALTH

NOTICE OF PUBLIC MEETING

Friday, September 8, 2023
9:00 a.m. – 4:00 p.m.

Note: This is a hybrid meeting held via Zoom and in-person at the Washington State Public Health Laboratory at 1610 NE 150 St, Shoreline, WA, 98155. Meeting access and instructions are provided below. Language interpretation available.

Newborn Screening Technical Advisory Committee (TAC) Agenda
Guanidinoacetate methyltransferase (GAMT) Deficiency and Arginase 1 Deficiency (ARG1-D)

Time	Agenda Item	Speaker
9:00 a.m.	1. Welcome & Introductions	Molly Dinardo, State Board of Health Melanie Ogleton, Cardea Services Amanda Winters, Cardea Services
9:20 a.m.	2. TAC Overview & Meeting Norms	Kelly Oshiro, TAC Co-Chair, State Board of Health Nirupama Shridhar, TAC Co-Chair, Department of Health Melanie Ogleton, Cardea Services Amanda Winters, Cardea Services
9:30 a.m.	3. Newborn Screening Program Overview	John Thompson, Department of Health
9:35 a.m.	4. Newborn Screening Criteria Review	Molly Dinardo, State Board of Health
9:40 a.m.	5. Family Perspective – GAMT Deficiency	Kim Tuminello
9:55 a.m.	6. Natural History of GAMT Deficiency – Diagnostic Testing & Available Treatment	Dr. Emily Shelkowitz, Seattle Children's

Time	Agenda Item	Speaker
10:25 a.m.	7. Available Screening Technology – GAMT Deficiency	Michael Katsuyama, Department of Health
10:35 a.m.	Break	
10:45 a.m.	8. Cost Benefit Analysis – GAMT Deficiency	Makena Chandra, University of Washington Michael Katsuyama, Department of Health
11:15 a.m.	9. Application of Criteria & Discussion	Melanie Ogleton, Cardea Services Amanda Winters, Cardea Services
11:45 a.m.	Working Lunch (as needed)	
12:15 p.m.	10. GAMT Deficiency Vote #1 – Criteria	Melanie Ogleton, Cardea Services Amanda Winters, Cardea Services
12:30 p.m.	11. GAMT Deficiency Vote #1 – Results & Discussion	Kelly Oshiro, TAC Co-Chair, State Board of Health Melanie Ogleton, Cardea Services Amanda Winters, Cardea Services
12:40 p.m.	12. GAMT Deficiency Vote #2 – TAC Recommendation	Melanie Ogleton, Cardea Services Amanda Winters, Cardea Services
12:55 p.m.	13. GAMT Deficiency Vote #2 – Results and Next Steps	Kelly Oshiro, TAC Co-Chair, State Board of Health Melanie Ogleton, Cardea Services Amanda Winters, Cardea Services
1:00 p.m.	Break	
1:10 p.m.	14. Family Perspective – ARG1-D	Christine Zahn

Time	Agenda Item	Speaker
1:25 p.m.	15. Natural History of ARG1-D – Diagnostic Testing & Available Treatment	Dr. Emily Shelkowitz, Seattle Children's
1:55 p.m.	16. Available Screening Technology – ARG1-D	Michael Katsuyama, Department of Health
2:05 p.m.	Break	
2:10 p.m.	17. Cost-Benefit Analysis – ARG1-D	Makena Chandra, University of Washington Michael Katsuyama, Department of Health
2:40 p.m.	18. Application of Criteria & Discussion	Melanie Ogleton, Cardea Services Amanda Winters, Cardea Services
3:00 p.m.	19. ARG1-D Vote #1 – Criteria	Melanie Ogleton, Cardea Services Amanda Winters, Cardea Services
3:15 p.m.	20. ARG1-D Vote # 1 – Results & Discussion	Nirupama Shridhar, TAC Co-Chair, Department of Health Melanie Ogleton, Cardea Services Amanda Winters, Cardea Services
3:25 p.m.	21. ARG1-D Vote # 2 – TAC Recommendation	Melanie Ogleton, Cardea Services Amanda Winters, Cardea Services
3:40 p.m.	22. ARG1-D Vote #2 – Results and Next Steps	Nirupama Shridhar, TAC Co-Chair, Department of Health Melanie Ogleton, Cardea Services Amanda Winters, Cardea Services
3:50 p.m.	23. Meeting Closeout	Nirupama Shridhar, TAC Co-Chair, Department of Health Kelly Oshiro, TAC Co-Chair, State Board of Health Melanie Ogleton, Cardea Services Amanda Winters, Cardea Services

Time	Agenda Item	Speaker
4:00 p.m.	Adjournment	

Zoom Meeting Information:

- **To access the meeting online and to register:**
  https://us02web.zoom.us/meeting/register/tZcodumvpzMuGNb95iClMBbVIPHT-7YGkOXm

  After registering, you will receive a confirmation email containing information about joining the meeting.

- **You can also dial-in using your phone for listen-only mode:**
  Call in: +1 (253) 205-0468
  Webinar ID: 835 1079 8422
  Passcode: 2023

Important Meeting Information to Know:

- This meeting is open to the public. The public can observe the meeting online.
- The Technical Advisory Committee will not take formal action or receive public comment. If you have comments or materials you would like to share with the full Board, please send them to wsboh@sboh.wa.gov.
- Times are estimates only. We reserve the right to alter the order of the agenda.
- If you have any technical difficulties accessing the meeting via Zoom, please contact Molly Dinardo at 564-669-3455 or molly.dinardo@sboh.wa.gov.
- Every effort will be made to provide Spanish interpretation, and American Sign Language (ASL). Should you need confirmation of these services, please email wsboh@sboh.wa.gov in advance of the meeting date.
- If you would like meeting materials in an alternate format or a different language, or if you are a person living with a disability and need reasonable modification, please contact the State Board of Health at (360) 236-4110 or by email wsboh@sboh.wa.gov. Please make your request as soon as possible to help us meet your needs. Some requests may take longer than two weeks to fulfill. TTY users can dial 711.

Welcome

Newborn Screening Technical Advisory Committee (TAC) PUBLIC MEETING

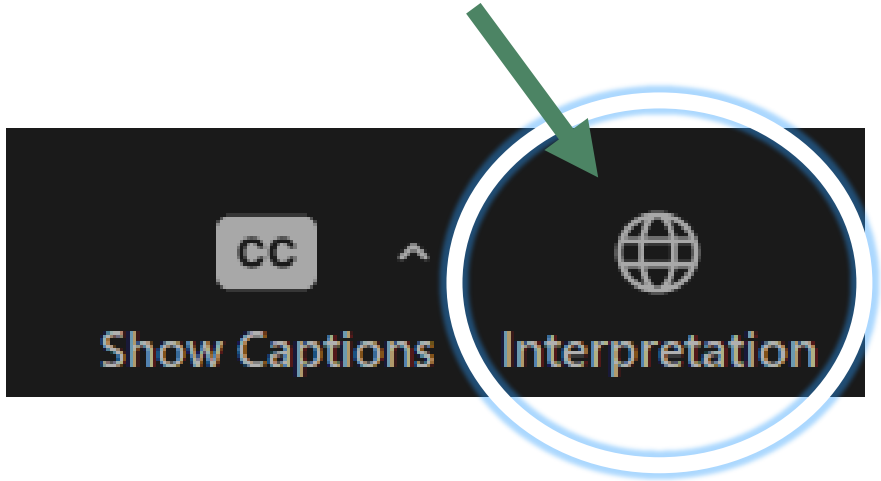
Meeting starts at 9:00 a.m.

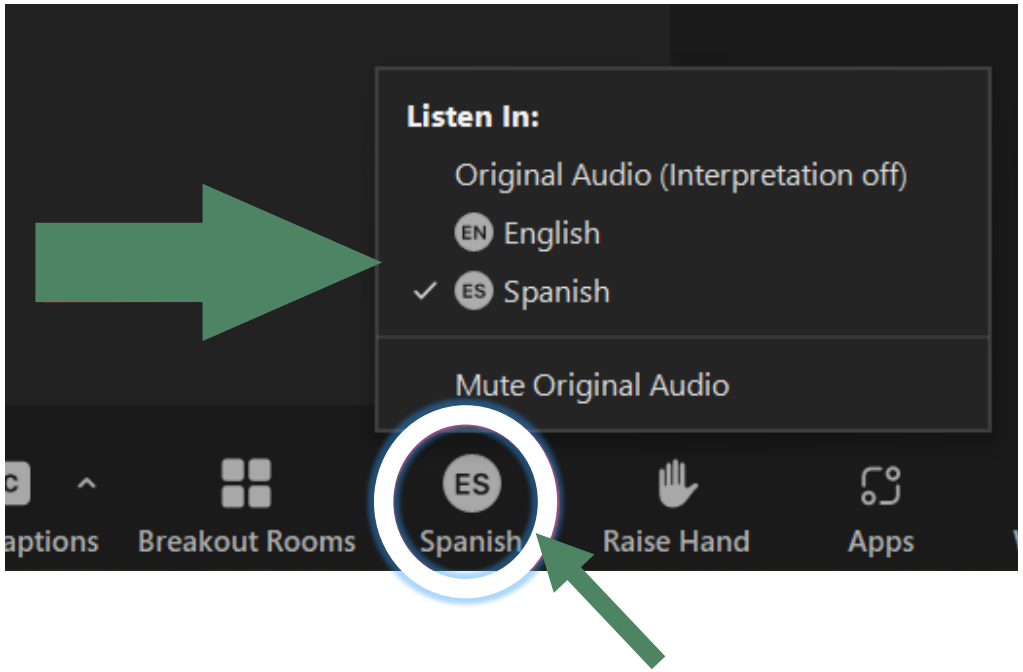
Washington State Department of HEALTH

WASHINGTON STATE BOARD OF HEALTH

Canales de Idioma de Zoom
Zoom Language Channels

Canales de idioma
Language channels





Elige un idioma
Choose a language

GAMT Deficiency & ARG1-D Technical Advisory Committee

Welcome, Expectations, Meeting Norms

- Notice of this meeting and meeting materials were posted to the Board's website in advance of the meeting.

- The TAC will not take formal action or receive public comment. If you have comments or materials you would like to share with the State Board of Health, please send them to wsboh@sboh.wa.gov.

- Times are estimates only. We reserve the right to alter the order of the agenda.

Today's Meeting Materials

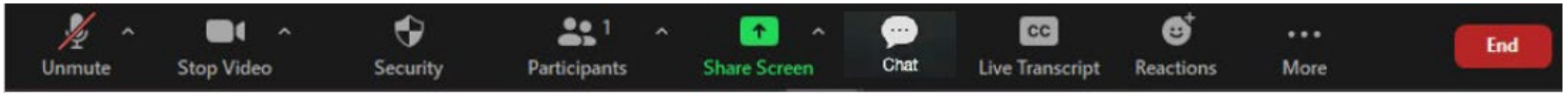
SCAN

https://sboh.wa.gov/meetings/meeting-information/meeting-information/materials/2023-09-08

Zoom Meeting Functions

Mute/
unmute mic

View participants,
change your name



Turn webcam
on/off

Leave
meeting

Note: Depending on your role, you may not have access to all functions identified on this slide.

Introductions

Meeting Purpose

Determine whether GAMT Deficiency and ARG1-D meets the Washington State Board of Health's criteria for inclusion in the list of conditions for which all Washington-born newborns must be screened.

Plan for the Day

- Presentations from families impacted by the conditions, Board staff, Department staff, and subject matter experts.
- Discussion and evaluation of GAMT Deficiency & ARG1-D against the Board's 5 criteria.
- Two rounds of voting and discussion of results for each condition to inform recommendations and next steps.

Meeting Norms

- Be here now and stay purpose-oriented
- Listen for understanding; seek clarification and resist assumptions
- Appreciate the strength of diverse cultures and perspectives
- Engage respectfully; see with new eyes and hear with new ears
- Move up into a speaking role; move up into a listening role
- Stay on topic and mind the time
- Assume positive intent; acknowledge and repair harms

Questions?

WASHINGTON STATE BOARD OF HEALTH

Washington State Board of Health

PROCESS TO EVALUATE CONDITIONS FOR INCLUSION IN THE REQUIRED NEWBORN SCREENING PANEL

The Washington State Board of Health has the duty under RCW 70.83.050 to define and adopt rules for screening Washington-born infants for heritable conditions. Chapter 246-650-020 WAC lists conditions for which all newborns must be screened. Members of the public, staff at Department of Health, and/or Board members can request that the Board review a particular condition for possible inclusion in the NBS panel. In order to determine which conditions to include in the newborn screening panel, the Board convenes an advisory committee to evaluate candidate conditions using guiding principles and an established set of criteria.

The following is a description of the Qualifying Assumption, Guiding Principles, and Criteria which the Board has approved in order to evaluate conditions for possible inclusion in the newborn screening panel. The Washington State Board of Health and Department of Health apply the qualifying assumption. The Board appointed Advisory Committee applies the following three guiding principles and evaluates the five criteria in order to make recommendations to the Board on which condition(s) to include in the state's required NBS panel.

QUALIFYING ASSUMPTION

Before an advisory committee is convened to review a candidate condition against the Board's five newborn screening requirements, a preliminary review should be done to determine whether there is sufficient scientific evidence available to apply the criteria for inclusion.

THREE GUIDING PRINCIPLES

Three guiding principles govern all aspects of the evaluation of a candidate condition for possible inclusion in the NBS panel.

- Decision to add a screening test should be driven by evidence. For example, test reliability and available treatment have been scientifically evaluated, and those treatments can improve health outcomes for affected children.
- All children who screen positive should have reasonable access to diagnostic and treatment services.
- Benefits of screening for the disease/condition should outweigh harm to families, children and society.

CRITERIA

1. **Available Screening Technology:** Sensitive, specific and timely tests are available that can be adapted to mass screening.
2. **Diagnostic Testing and Treatment Available:** Accurate diagnostic tests, medical expertise, and effective treatment are available for evaluation and care of all infants identified with the condition.
3. **Prevention Potential and Medical Rationale:** The newborn identification of the condition allows early diagnosis and intervention. Important considerations:
    - There is sufficient time between birth and onset of irreversible harm to allow for diagnosis and intervention.
    - The benefits of detecting and treating early onset forms of the condition (within one year of life) balance the impact of detecting late onset forms of the condition.
    - Newborn screening is not appropriate for conditions that only present in adulthood.
4. **Public Health Rationale:** Nature of the condition justifies population-based screening rather than risk-based screening or other approaches.
5. **Cost-benefit/Cost-effectiveness:** The outcomes outweigh the costs of screening. All outcomes, both positive and negative, need to be considered in the analysis. Important considerations to be included in economic analyses include:
    - The prevalence of the condition among newborns.
    - The positive and negative predictive values of the screening and diagnostic tests.
    - Variability of clinical presentation by those who have the condition.
    - The impact of ambiguous results. For example the emotional and economic impact on the family and medical system.
    - Adverse effects or unintended consequences of screening.

WASHINGTON STATE
BOARD OF HEALTH

Guanidinoacetate methyltransferase (GAMT) Deficiency Overview
Newborn Screening Technical Advisory Committee
September 8, 2023

ABOUT THE CONDITION

- GAMT Deficiency is a rare inherited amino acid disorder that prevents the body from properly producing creatine, which helps your organs store and use energy.[1,2]
- Without enough creatine, the body's organs do not get enough energy to support its vital functions, which can cause damage.
- This damage primarily affects the brain and muscles, as these organs need the most energy.
- Without early treatment, GAMT deficiency can cause serious cognitive and neurological impairments.
- GAMT deficiency is caused by changes in the GAMT gene.

SIGNS & SYMPTOMS

- People with GAMT Deficiency may begin showing symptoms from early infancy to age three.
- Signs and symptoms can vary but may include mild to severe intellectual and developmental disabilities, delayed sitting or walking, delayed or limited speech ability, muscle weakness or low muscle tone, behavioral issues (anxiety, aggression, self-injury, hyperactivity), seizures, and uncontrollable movements.

DIAGNOSIS

- GAMT Deficiency can be detected through a newborn screening blood spot using tandem mass spectrometry.
- Diagnostic tests include testing for guanidinoacetate (GUAC) and low creatine in the blood after a positive newborn screening test.
- Molecular testing can also be helpful.

TREATMENT

- May include creatine and ornithine supplements, sodium benzoate (a medication that can reduce levels of an amino acid called glycine), medications to treat seizures, a lifelong diet low in protein, and speech, occupational, and behavior therapy.

To request this document in an alternate format or a different language, please contact the State Board of Health at (360) 236-4110 or by email at wsboh@sboh.wa.gov.

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1. National Institutes of Health, Genetic and Rare Diseases Information Center. Guanidinoacetate methyltransferase deficiency - About the Disease. Updated February 2023. Accessed August 25, 2023. https://rarediseases.info.nih.gov/diseases/2578/guanidinoacetate-methyltransferase-deficiency

2. Health Resources & Services Administration. Guanidinoacetate methyltransferase deficiency | Newborn Screening. Updated June 2023. Accessed August 25, 2023. https://newbornscreening.hrsa.gov/conditions/guanidinoacetate-methyltransferase-deficiency

Arginase 1 Deficiency (ARG1-D) Overview
Newborn Screening Technical Advisory Committee
September 8, 2023

ABOUT THE CONDITION

- ARG1-D is a rare and inherited metabolic disease that prevents the body from properly breaking down the amino acid arginine, an enzyme in the blood.[1,2]
- Arginase is one of six enzymes responsible for breaking down arginine and is part of an essential process in the body called the urea cycle.
- The urea cycle helps remove ammonia (or nitrogen) from the body, a waste product used to process protein.
- If the arginase enzyme isn't working properly, the body can't break down arginine and get rid of ammonia through the urea cycle.
- Irregularities in the urea cycle may cause levels of ammonia in the blood to increase.
- When ammonia levels become too high, it has toxic effects and can cause serious damage to the nervous system and other parts of the body.

SIGNS & SYMPTOMS

- Signs of ARG1-D can vary widely and may appear anytime from infancy to early childhood.
- Symptoms of ARG1-D include seizures, muscle tightness or stiffness, difficulty eating, vomiting, and trouble breathing.
- People with ARG1-D might also experience delays in both physical and cognitive development, loss of developmental milestones, and intellectual disabilities.

DIAGNOSIS

- ARG1-D can be detected through a newborn screening blood spot using tandem mass spectrometry.
- Diagnostic tests include testing for ammonia levels, amino acids, and urine organic acids (specifically orotic acid) after a positive newborn screening test.

TREATMENT

- May include a diet low in protein, special foods or formulas, eating regularly and avoiding missing meals, and medications to get rid of extra arginine and ammonia in the body.

To request this document in an alternate format or a different language, please contact the State Board of Health at (360) 236-4110 or by email at wsboh@sboh.wa.gov.

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1. Morales A, Sticco KL. Arginase Deficiency - NIH Bookshelf. In: *StatPearls*. StatPearls Publishing; 2023. Accessed August 25, 2023. http://www.ncbi.nlm.nih.gov/books/NBK482365/

2. Health Resources and Services Administration. Arginase deficiency | Newborn Screening. Updated June 2023. Accessed August 25, 2023. https://newbornscreening.hrsa.gov/conditions/arginase-deficiency

SAVING LIVES WITH A SIMPLE BLOOD SPOT

NEWBORN SCREENING

Washington State Department of Health

What is Newborn Screening?

Newborn screening is a public health system that detects infants with serious but treatable conditions that may not be apparent at birth.

There are 3 types of newborn screening programs:

1

Blood-spot

2

Hearing

3

Pulse Oximetry

Why is Newborn Screening Important?

- It prevents death and disability for **thousands** of infants every year in the USA by providing early treatment
- The public benefits through savings in health care and disability support costs

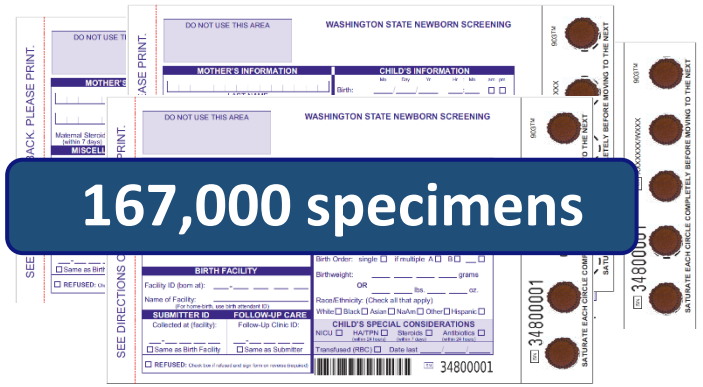
Two 6 year old girls with Congenital Hypothyroidism
Healthy 18 year old with CH, detected through Washington Newborn Screening as a baby

Washington Screens for… 32 disorders!

Amino Acid Disorders (6)	Fatty Acid Oxidation Disorders (5)	Organic Acid Disorders (8)
Phenylketonuria Homocystinuria Maple syrup urine disease Citrullinemia type I Argininosuccinic acidemia Tyrosinemia type I	Medium-chain acyl-CoA dehydrogenase deficiency Long-chain L-3-hydroxy acyl-CoA dehydrogenase deficiency Trifunctional protein deficiency Very long-chain acyl-CoA dehydrogenase deficiency Carnitine uptake defect	Isovaleric acidemia Glutaric acidemia type I Methylmalonic acidemias (CblA/B and MUT) Propionic acidemia Multiple carboxylase deficiency Beta-ketothiolase deficiency 3-hydroxy-3-methylglutaric aciduria
Endocrine Disorders (2)	**Lysosomal Storage Disorders (2)**	**Other Disorders (10)**
Congenital hypothyroidism Congenital adrenal hyperplasia	Mucopolysaccharidosis type I Glycogen storage disorder type II (Pompe)	Galactosemia Biotinidase deficiency Cystic fibrosis Sickle Cell Diseases & Hemoglobinopathies Severe combined immunodeficiency X-linked adrenoleukodystrophy Spinal muscular atrophy

Washington State Numbers

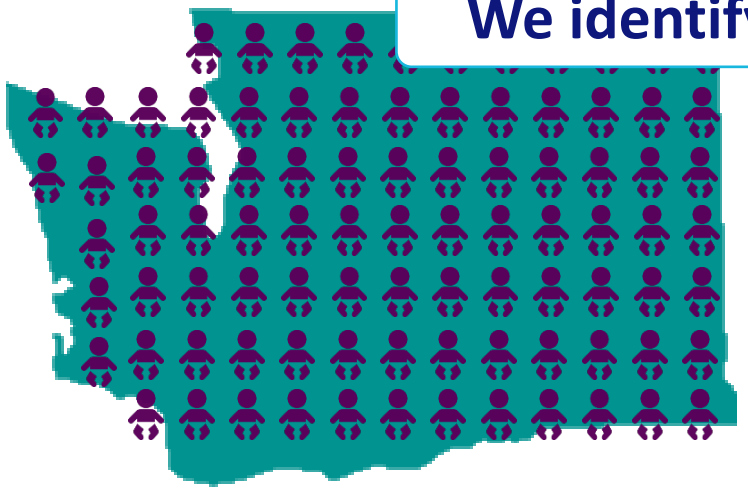
84,000 newborns



167,000 specimens

12 million tests

We identify:



200 infants

every year who benefit from early diagnosis and treatment

1,300 infants with a hemoglobin trait (not disease)

What happens when a baby has abnormal results?

Dedicated team ensures the baby gets the care they need

- Depends on what the results are and which condition is suspected

Can include:

- Call baby's health care provider to check clinical status, recommend immediate evaluation and diagnostics for life-threatening conditions
- Ensure repeat specimen is submitted to resolve borderline results
- After confirmed diagnoses, ensure baby is linked into specialty care

Specialty Care Partners

Consultants:

- Seattle Children's Hospital
  - Endocrinologist
  - Pediatric Hematologist
  - Biochemical Geneticists
  - Immunologists
  - Pulmonologists
- Mary Bridge Children's
  - Biochemical Geneticists

Community:

- Northwest Sickle Cell Collaborative

Specialty Care Clinics:

- University of Washington
  - PKU Clinic
  - Congenital Hypothyroidism Developmental Evaluation Clinic
  - Neuropsych Evaluation Program
  - Biochemical Genetics Clinic
- Seattle Children's Hospital
  - Biochemical Genetics Clinic
  - Odessa Brown Sickle Cell Clinic
- Mary Bridge Sickle Cell Clinic

Mary Bridge Children's Hospital · Clinics · Foundation
1955 · CELEBRATING 60 YEARS · 2015

UW Medicine
DEPARTMENT OF MEDICINE
DEPARTMENT OF PEDIATRICS
UNIVERSITY of WASHINGTON

Seattle Children's
HOSPITAL · RESEARCH · FOUNDATION

northwest sickle cell COLLABORATIVE

ODESSA BROWN CHILDREN'S CLINIC

Cristine M. Trahms Program for Phenylketonuria
ΦKU Seattle
A Member Forever

Recommended Uniform Screening Panel (RUSP)

Federal Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC)

- Nomination to RUSP
- Full evidence review and voting

- GAMT added in January 2023
- ARG1 is considered a Secondary Condition

~Thank you~

Together we protect the lives of
Washington's youngest citizens.

Washington State Department of Health

Process to Evaluate Conditions for Inclusion in the Required Newborn Screening Panel

September 08, 2023

Department and Board Authority – Relevant Laws and Rules for Newborn Screening

Chapter 70.83 Revised Code of Washington (RCW)

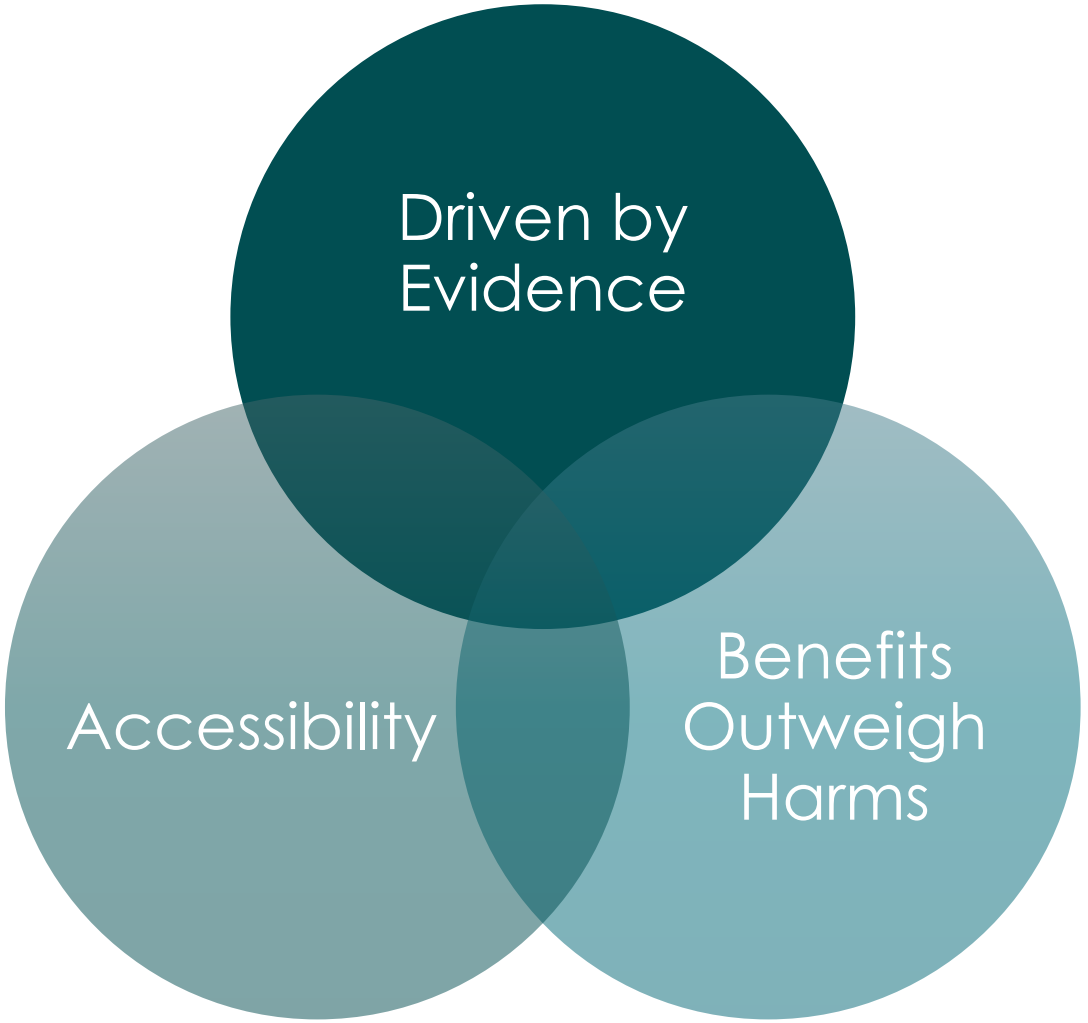
- RCW 70.83.020, Department authority to require screening tests for all babies in Washington (not just babies born in hospitals).
- RCW 70.83.050, Board authority to define and adopt rules for screening Washington-born infants for heritable conditions.

Chapter 246-650 Washington Administrative Code (WAC)

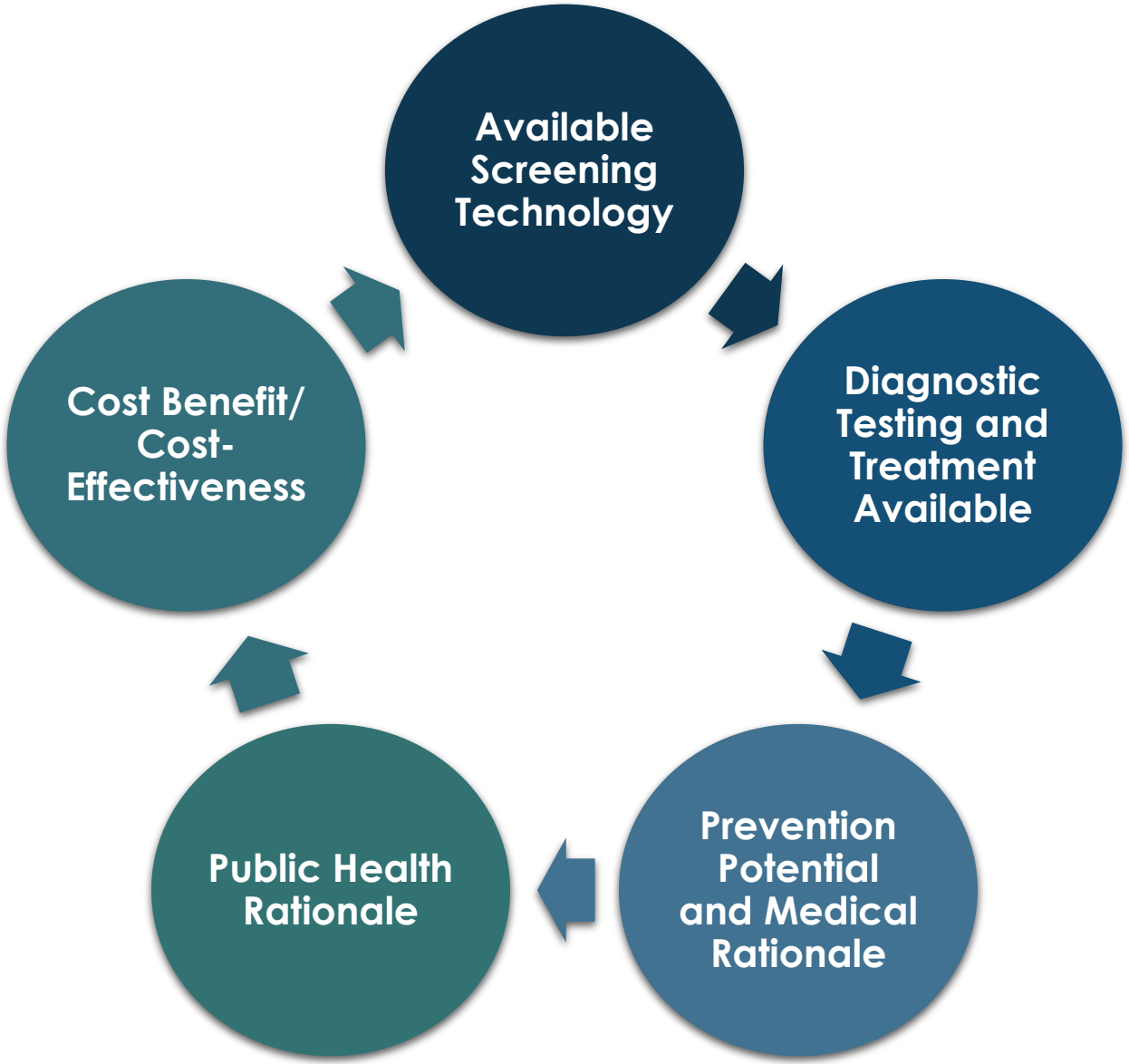
- WAC 246-650-010 and WAC 246-650-020, define and list the conditions required for the state's newborn screening panel.

Three Guiding Principles

Three guiding principles govern all aspects of the evaluation of a candidate condition for possible inclusion in Washington's Newborn Screening panel:



Five Newborn Screening Criteria



Available Screening Technology

Sensitive, specific and timely tests are available that can be adapted to mass screening.

Diagnostic Testing and Treatment Available

Accurate diagnostic tests, medical expertise, and effective treatment are available for evaluation and care of all infants identified with the condition.

Prevention Potential and Medical Rationale

The newborn identification of the condition allows early diagnosis and intervention. Important considerations:

- There is sufficient time between birth and onset of irreversible harm to allow for diagnosis and intervention.
- The benefits of detecting and treating early onset forms of the condition (within one year of life) balance the impact of detecting late onset forms of the condition.
- Newborn screening is not appropriate for conditions that only present in adulthood.

Public Health Rationale

Nature of the condition justifies population-based screening rather than risk-based screening or other approaches.

Cost-benefit / Cost-effectiveness

The outcomes outweigh the costs of screening. All outcomes, both positive and negative, need to be considered in the analysis. Important considerations to be included in economic analyses include:

- The prevalence of the condition among newborns.
- The positive and negative predictive values of the screening and diagnostic tests.
- Variability of clinical presentation by those who have the condition.
- The impact of ambiguous results. For example, the emotional and economic impact on the family and medical system.
- Adverse effects or unintended consequences of screening.

NEWBORN SCREENING TECHNICAL ADVISORY COMMITTEE OPTIONAL REFLECTION TOOL

GAMT

		Criteria				
		Available Screening Technology	Diagnostic Testing & Treatment Available	Prevention Potential & Medical Rationale	Public Health Rationale	Cost-benefit/Cost-effectiveness
Reflective Principles-Based Questions	What stands out to you about the evidence? What do you want to learn more about?	Click or tap here to enter text.	Click or tap here to enter text.	Click or tap here to enter text.	Click or tap here to enter text.	Click or tap here to enter text.
	What access & equity considerations are you thinking about?	Click or tap here to enter text.	Click or tap here to enter text.	Click or tap here to enter text.	Click or tap here to enter text.	Click or tap here to enter text.

Questions?

WASHINGTON STATE BOARD OF HEALTH

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Washington State Department of Health

NEWBORN SCREENING FOR GUANIDINOACETATE METHYLTRANSFERASE (GAMT) DEFICIENCY

Technical Advisory Committee meeting
September 8, 2023

Presenter

Michael Katsuyama

MPHc

Follow-up Lead

Newborn Screening Program

NBS Criteria

- Available Screening Technology
  - Sensitive, specific and timely tests are available that can be adapted to mass screening
- Diagnostic Testing and Treatment Available
- Prevention Potential and Medical Rationale
- Public Health Rationale
- Cost-benefit/Cost-effectiveness

Available Screening Technology

- Sensitivity – the ability of the screen to correctly identify the babies with GAMT Sensitivity = 1 – false negative rate

- Specificity – the ability of the screen to correctly identify the babies who do not have GAMT
  - Specificity = 1 – false positive rate

- Positive predictive value (PPV) – the percent of babies with a positive screen who have GAMT

- PPV = $\frac{\#\ \text{true(+)}}{\#\ \text{true(+)} + \#\ \text{false(+)}}$

Timely Tests

- Timeliness
  - Aim: Identify and treat prior to onset of symptoms
  - Each step important
    - Specimen collection
    - Specimen Transport
    - Testing
    - Result reporting
  - Goal: time-critical results reported by 5 days of life

Source: Sontag et al. PLoS ONE 15(4):e0231050 (2020 – funded by HRSA)

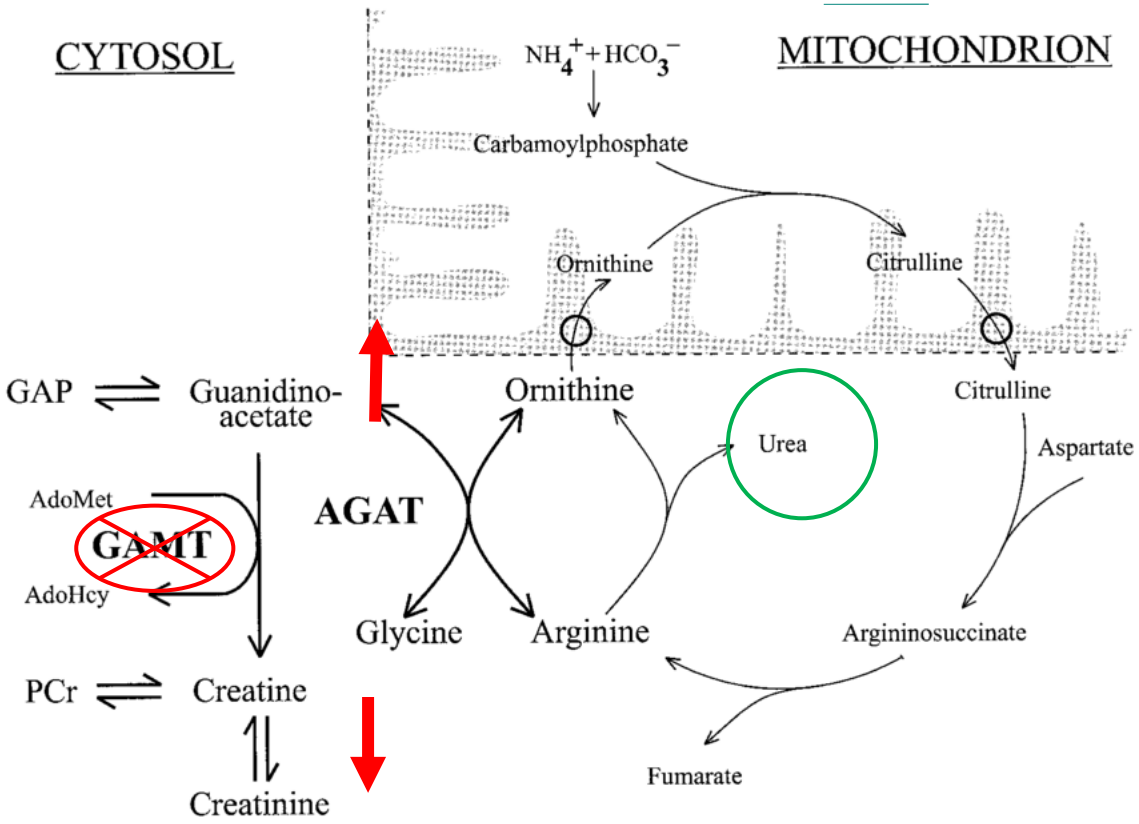
Test for GAMT
Adapted to Mass Screening

- Technology – tandem mass spectrometry (MS/MS)
- Uses one 1/8" hole punch from dried blood spot to test for 19 congenital disorders simultaneously
  - Amino acids
  - Acylcarnitines (fat transporters)
- In WA NBS Program since 2004

Test for GAMT
Adapted to Mass Screening

- Acylcarnitine/Amino acid analysis
  - Primary target: high guanidoacetate [GUAC]
  - Secondary markers may be helpful to reduce false(+) results

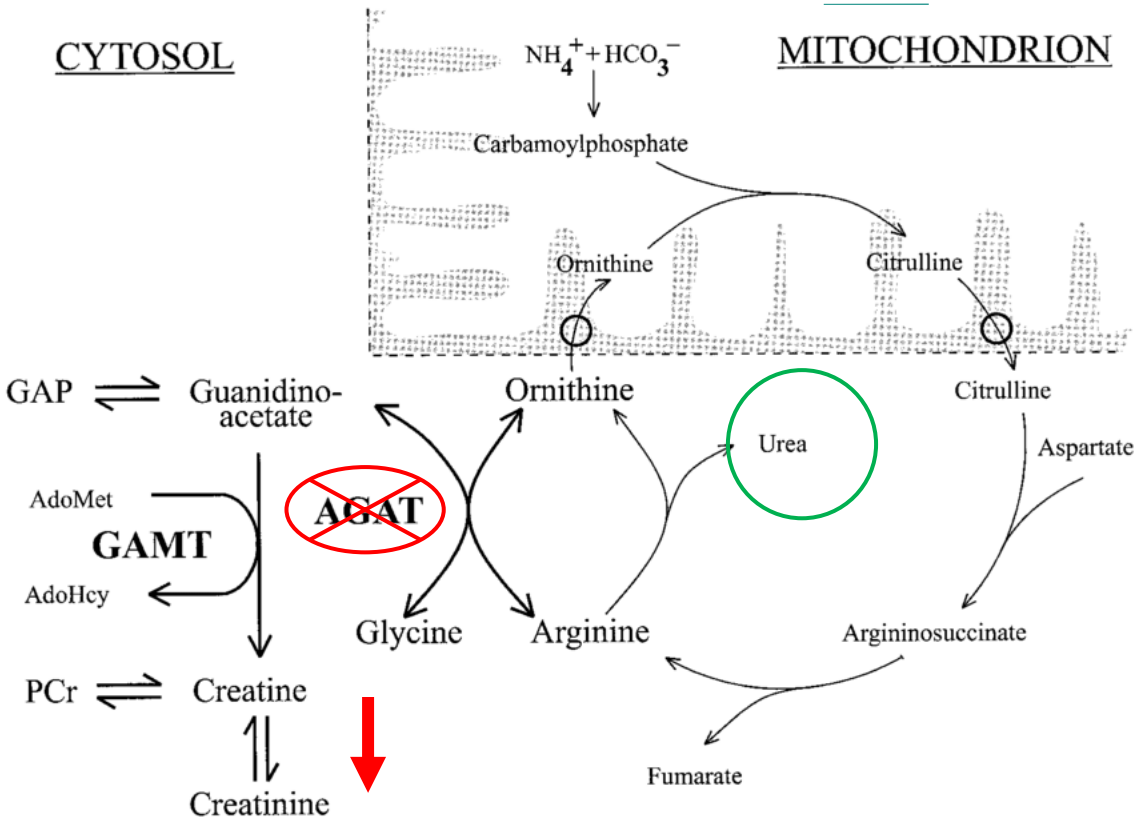
Test for GAMT
Adapted to Mass Screening



High [GUAC]

Low [Creatine]

Test for GAMT
Adapted to Mass Screening



Also has low [Creatine]

- AGAT deficiency

Only GAMT has high [GUAC]

AGAT Prevalence: less than 20 cases ever reported

CCDS caused by transporter defects = normal GUAC and Creatine in blood

Newborn Screening - GAMT

- Across 4 screening jurisdictions– 3.07 million babies
  - 3 cases of GAMT (prevalence = 1:1,000,000 births)
    - 3 true positives (sensitivity = 100.0%)
    - 0 false negatives
  - False positive rate (NY+UT) 2.1/100,000 (specificity = 99.99%)
  - PPV = 99.99%

Questions?

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Washington State Department of Health

NEWBORN SCREENING FOR ARGINASE 1 DEFICIENCY (ARG-I)

Technical Advisory Committee meeting
September 8, 2023

Presenter

Michael Katsuyama

MPHc

Follow-up Lead

Newborn Screening Program

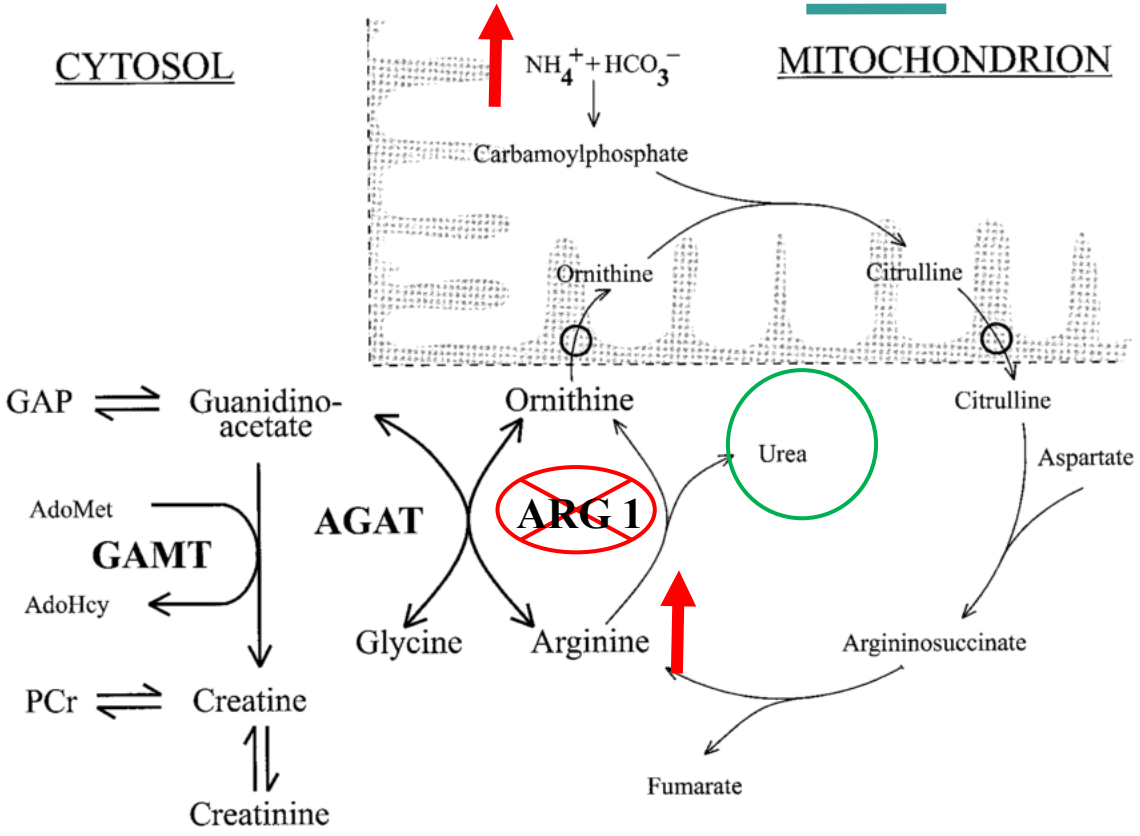
Test for ARG-I
Adapted to Mass Screening

- Technology – tandem mass spectrometry (MS/MS)
- Uses one 1/8" hole punch from dried blood spot to test for 19 congenital disorders simultaneously
  - Amino acids
  - Acylcarnitines (fat transporters)
- In WA NBS Program since 2004

Test for ARG-I
Adapted to Mass Screening

- Amino acid analysis
  - Primary target: high Arginine

Test for ARG-I
Adapted to Mass Screening



High [Arginine]

Newborn Screening – ARG-I

- US based Screening– 29 million babies
  - 22 cases of ARG-I (prevalence = 0.75:1,000,000 births)
    - 22 true positives (sensitivity = 100.0%)
    - 0 false negatives
  - False positive rate (US) 5.0/100,000 (specificity = 99.99%)
  - PPV = 99.99%

Questions?

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