Newborn Screening Advisory Committee

First Biannual Advisory Committee Meeting
April 19, 2022
1:00 pm -3:00 pm

MONTANA DPHHS
Healthy People. Healthy Communities.
Department of Public Health & Human Services

CSHS
CHILDREN'S SPECIAL HEALTH SERVICES

Introductions

1. Name
2. Organization
3. Role
4. Physical Description

Agenda

1:00p - 1:20p	**Introductions** ● Yarrow Facilitators ● Voting & Non-Voting Committee Members ● Ground Rules
1:20p - 1:50p	**Montana Newborn Screening Program Overview** ● Montana Program Details ● Lab and Lab Processes ● SMA Example
1:50p - 2:10p	**Newborn Screening Advisory Committee Background** ● General NBS Committee Background ● Committee Roles & Responsibilities
2:10p - 2:40p	**Newborn Screening Advisory Committee Next Steps** ● Committee Regulations ● Condition Criteria ● Website ● Meeting Schedule
2:40p - 3:00p	**Review & Wrap Up** ● Question and Answer Period ● Public Comment Period

Non-Voting Advisory Council Members

Name	Organization	Role
Adam Meier	DPHHS	Non-voting member
Mackenzie Petersen	DPHHS/CSHS	Internal workgroup
Amber Bell	DPHHS/CSHS	Internal workgroup
Angela Dusko	DPHHS/Lab	Internal workgroup
Crystal Fortune	DPHHS/Lab	Internal workgroup
Kirsten Krane	Yarrow	Contracted facilitator (non-member)
Anna Schmitt	Yarrow	Contracted facilitator (non-member)
Krystal Bosenbark	Yarrow	Contracted facilitator (non-member)

Voting Advisory Council Members

Name	Organization	Role
Sara Sullivan		Person affected by or family members of a person affected by a disorder
Kotie Dunmire	Butte High School	Person affected by or family members of a person affected by a disorder
Dr. Allison Young		Physician / Nurse practitioners who are board-certified in obstetrics, pediatrics, family medicine, or neonatology
Jennifer Banna	University of Montana	Physician / Nurse practitioners who are board-certified in obstetrics, pediatrics, family medicine, or neonatology

Voting Advisory Council Members

Name	Organization	Role
Amanda Osborne	Helena Birth Studio	Representative of a birthing center
Miranda Prevel	DPHHS	Representative of medicaid or the insurance industry
Shelley Eagan	Billings Clinic	Representative of an advocacy association regarding newborns with medical conditions or rare disorders
Dr. Abdallah "Abe" Elias	Shodair	Medical geneticist or who has at least 5 years of experience working in a testing laboratory
Marion Rudek	IHS	Representative who works in a tribal health care system

Ground Rules

- Mute
- Video
- Questions in the chat
- Clarifying questions
- Avoid interrupting
- Avoid acronyms
- Use specific examples
- Focus on the collective interests and goals
- Additional meetings or communications may be scheduled
- Next steps assigned to ensure accountability
- Facilitators may call on attendees for input
- Safe space

Ground rules adapted from Strategy Management Consulting "*Ground Rules for Effective Meetings and Strategic Planning Offsites*"

Mackenzie’s Slides

Newborn (Bloodspot) Screening Background

Newborn screening has been named one of the Top 10 Great Public Health Achievements by the Centers for Disease Control and Prevention (bloodspot screening is the third component)

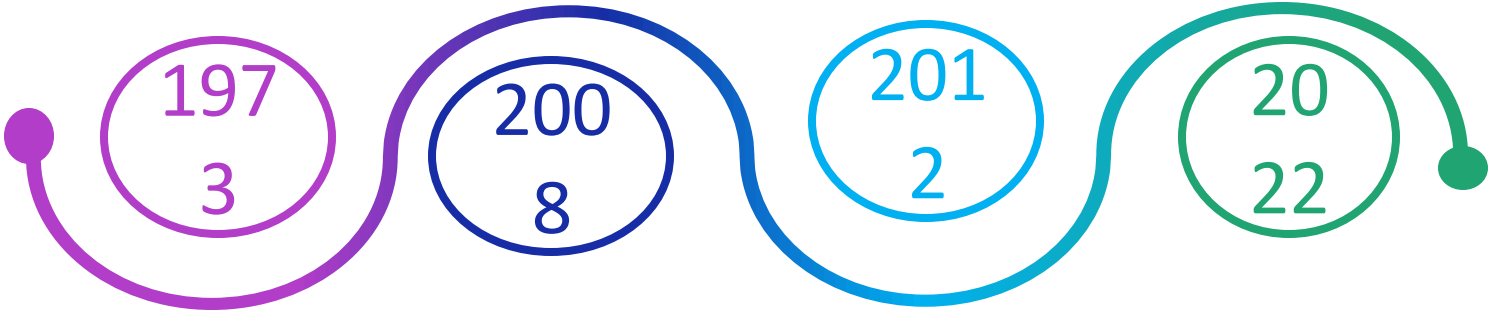
1963 saw the first PKU testing with Robert Guthrie's bacterial inhibition assay for phenylketonuria (PKU)

- Cystic fibrosis late 1990's, early 2000's
- 1990s-Tandem Mass Spectroscopy
- Severe Combined Immunodeficiency (SCID) aka "Bubble Boy syndrome"
- Lysosomal storage disorders

NEWBORN SCREENING: TOWARD A UNIFORM SCREENING PANEL AND SYSTEM (Advisory Committee on Heritable Disorders in Newborns and Children | Official web site of the U.S. Health Resources & Services Administration (hrsa.gov))

History of Montana Newborn (Bloodspot) Screening

Note: Due to Montana's low birth rate (12,000 births per year), our annual incidence rates do not reflect the statistical averages.



1973 — **PKU and Congenital hypothyroidism**

2008 — **MT mandated all primary conditions on the RUSP**

2012 — **MT added SCID**

2022 — **First MT NBS Advisory Panel meeting**

Bloodspot Screening in Montana

Figure 4. **Phases of the NBS Blot Spot Process**

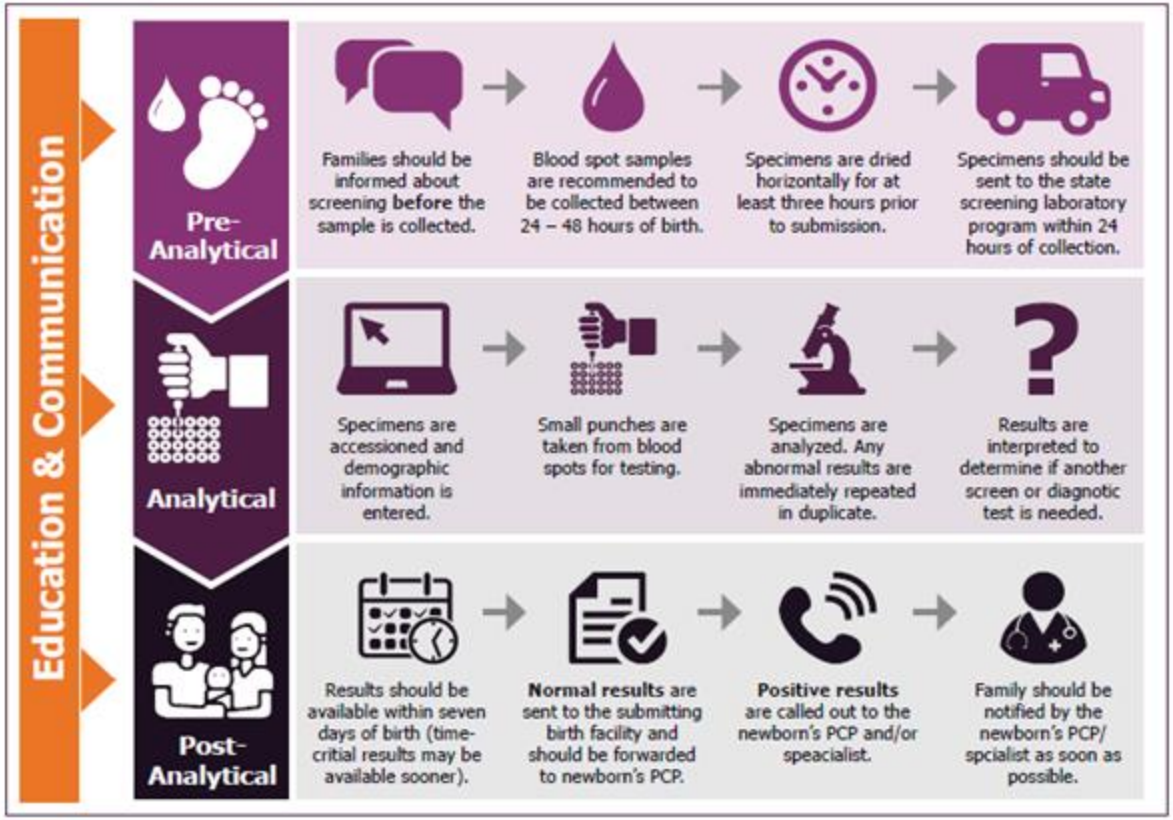


Image taken from NewSTEPS-SMA-Toolkit (June 2021)

The newborn screening program is authorized in State Statute (Montana Code Annotated 50-19-203) and by Administrative Rule (ARM 37.57.3: Infant Screening Tests and Eye Treatment)

MTPHL NBS website:

https://dphhs.mt.gov/publichealth/LaboratoryServices/NewbornScreening

Spinal Muscular Atrophy (SMA)

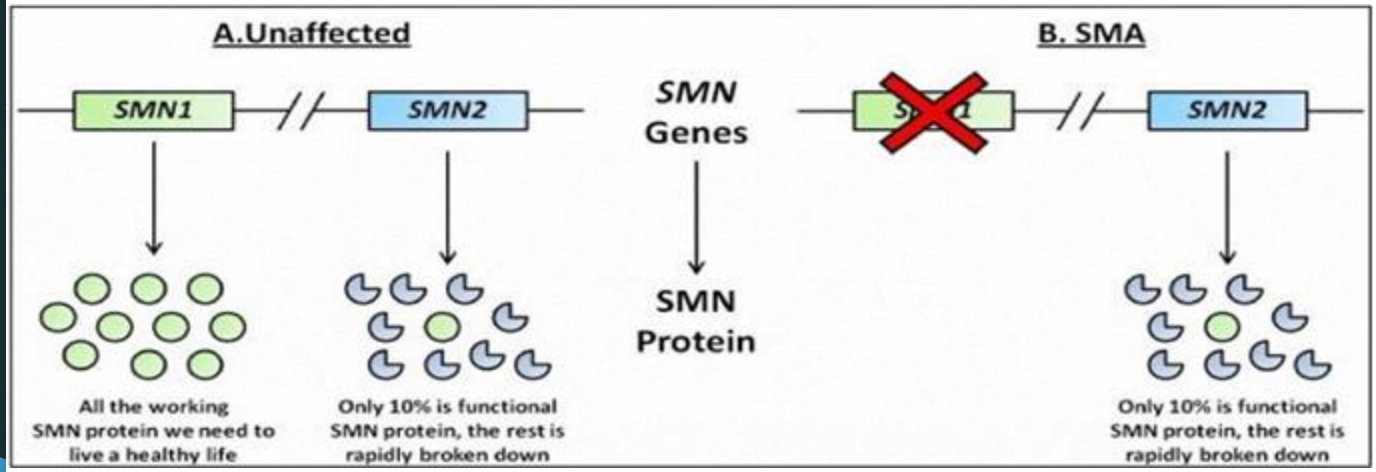


Image from New STEPS-SMA-Toolkit (June 2021)

[Summary of Nominated Conditions to the Recommended Uniform Screening Panel (RUSP) (hrsa.gov)]

Survival Motor Gene (SMN)

Second nomination in 2017 and added to the recommended uniform screening panel (RUSP) in July 2018

Readiness

- Laboratory
- Follow-up
- Information technology

NBS Committees - Generally

- TX
- AZ
- MN
- WI

NBS Committee Member Roles & Responsibilities

- Waiting on summary document from lawyer

NBS Advisory Committee Regulations

- Waiting on summary document from lawyer

NBS New Condition Criteria

1. Screening is needed to identify all babies who may need treatment

2. There is a significant risk of illness, disability, or death if babies are not treated promptly,

3. Effective treatment and access to follow-up care and counseling is widely available

4. Treatment is more beneficial in the newborn period than later

5. The benefits to babies and to society outweigh the risks and burdens of screening and treatment.

6. The public health laboratory can support the testing resources and expertise necessary to provide accurate and timely results.

NBS New Condition Criteria

7. Consider the financial impacts on the family

8. There is a public health benefit to conducting the test

9. There exist responsible parties who will follow up with families and implement necessary interventions

10. Case Definition. Are the condition's case definition and spectrum well described? Can they predict the phenotype or range of symptoms in newborns and children who will be identified through population-based screening?

11. There is a screening test available now or expected within 12 months that can be done quickly and is successful in finding affected newborns.

NBS New Condition Criteria Comparison

Common Themes
1. Seriousness of the condition 2. Screening is necessary to detect condition 3. Availability of treatment if screen is positive for the condition 4. Availability of specialists who are equipped to care for patient with the screened condition [this is tricky with the limited # of specialists in Montana, but maybe the specialists are in other states so it shouldn't be a limiting factor? - MP] 5. Labs [any lab – our lab or a lab we can contract with-MP] have the equipment necessary to test for the condition 6. Public health/societal benefit to screening for condition 7. Available screening for the condition is successful (sensitive and specific) in detecting newborns with the condition

Condition Criteria Comparison - RUSP

1. *Condition Seriousness*
2. *Case Definition*
3. *Analytic Validity*
4. *Clinical Utility*
5. *Treatments*
6. *Prospective Pilot Data*

Condition Criteria Comparison - MN

1. Support from an appropriate screening facility and the nominated condition is considered feasible to add
2. Clinical specialist(s) are available, ready to accept referrals, and willing to manage patients found through screening.
3. Can be found between 24 and 48 hours of life through screening but cannot be identified clinically in that time frame.
4. There is a screening test available now or [soon] that can be done quickly and is successful in finding affected newborns.
5. There is safe and effective treatment and/or intervention available…
6. There is an infantile onset form of this condition.

Condition Criteria Comparison - MN

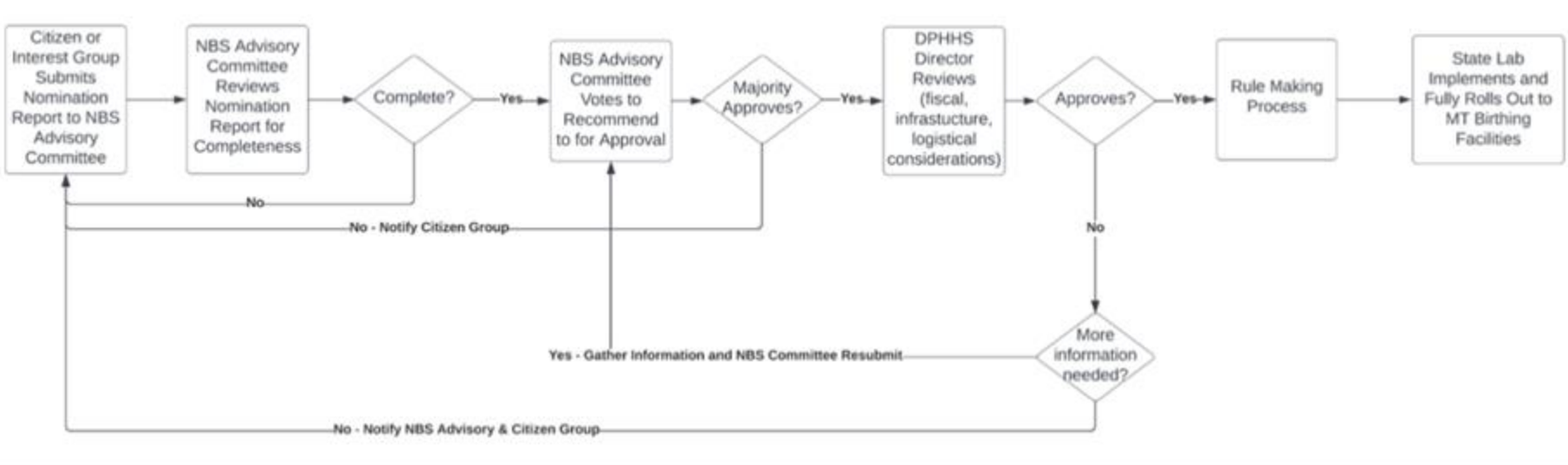
1. Screening is needed to identify all babies who may need treatment.
2. There is a significant risk of illness, disability, or death if babies are not treated promptly
3. Effective treatment is available
4. Treatment is more beneficial in the newborn period than later
5. Resources and access to treatment and counseling are widely available
6. The benefits to babies and society outweigh the risks and burdens of screening and treatment

Condition Criteria Comparison - MT

These are not criteria per se – rather they are potential items to think about adding to the MT NBS Selection Criteria List

- Is the test financially possible for the lab to acquire necessary resources [lab needs include supplies and staffing as well as equipment-CF]?
- Financial impacts on family?
- Is safe and effective treatment available to all affected newborns, regardless of socioeconomic status?
- Public health benefit? [how do we define the attributes of a condition meeting a required level of 'public health benefit' - other than them satisfying all other requirements?-MP]
- Follow-up responsibilities and implementation

NBS Condition Criteria Decision Making



Summary of Nominated Conditions to the Recommended Uniform Screening Panel (RUSP)

CONDITION	NOMINATION SUBMITTED to HRSA mm/yy	REVIEW NOMINATION N&P WG** Review mm/yy	COMMITTEE VOTE Initiate Evidence Review mm/yy	EVIDENCE REVIEW Preliminary Report and/or Presentations mm/yy	EVIDENCE REVIEW Final Report and Presentation mm/yy	COMMITTEE VOTE Recommend Adding to the RUSP mm/yy	SECRETARY APPROVAL Add to the RUSP mm/yy
Guanidinoacetate Methyltransferase Deficiency (GAMT) *3rd Nomination	6/21	7/21	Approved 08/21	11/21; 02/22	-	-	-
Mucopolysaccharidosis II (MPS II)	12/20	02/21	Approved 05/21	08/21; 11/21	02/22	Approved 2/22	-
Spinal Muscular Atrophy (SMA) *2nd Nomination	2/17	04/17	Approved 05/17	08/17; 11/17	02/18	Approved 02/18	**07/18**
Krabbe Disease	12/07	2/08	Approved 08/08	05/09	09/09	NOT Approved 09/09	-
Fabry Disease	12/07	2/08	NOT Approved 08/08	-	-	-	-
Pompe Disease *1st Nomination	10/07	11/07	Approved 01/08	08/08	10/08	NOT Approved 10/08	-
Severe Combined Immunodeficiency (SCID)	09/07	11/07	Approved 01/08	08/08; 11/08	02/09	Approved 02/09	**02/10**

*Conditions can be nominated more than once if the Committee does not approve initiation of an evidence review or does not recommend adding the condition to the RUSP

**Nomination and Prioritization Workgroup

Website

Next Meeting

- According to HB 423: Meet twice per year
- Select a Fall month
- Doodle Poll to follow

Questions?

Public Comment Period

- Put comments in chat
- Moderator will read aloud
- 3 minute max per comment
- Unaddressed comments will be addressed via email
- Will accept public comment via email

Thanks + Next Steps

- Follow-up email will be sent soon
  - Meeting Minutes
  - Recording
  - Presentation Slides
  - Next Meeting Doodle Poll
- Email if you have questions & comments or need anything