Newborn Screening Advisory Committee

Wednesday, August 26, 2024
1:00 pm - 4:30 pm

Role Call / Introductions

1. Name
2. Organization
3. Role
4. Physical Description (e.g. age, skin color, gender, hairstyle and hair color, clothes description, any distinctive accessories)*

*Please include a physical description of yourself for meeting participants who may be visually impaired. Share only those attributes you feel comfortable sharing. Thank you!

Agenda

Time	Agenda Item
1:00p - 1:20p	**Welcome & Roll Call** ● Icebreaker ● Voting & Non-Voting Members
1:20p - 1:40p	**Unfinished Business**
1:40p - 1:50p	**Vote on Pompe**
1:50p - 2:05p	**Gaucher Nomination Packet Review**
2:05p - 2:20p	**Gaucher Packet Discussion**
2:20p - 2:30p	Break

Agenda

2:30p - 3:20p	**Gaucher Presentations** ● SME: Dr. Ozlem Goker-Alpan, MD ● Family: Aviva Rosenberg and Chris Heredia ● Montana State Laboratory: Jeanne Lee
3:20p - 4:00p	**Gaucher Discussion** ● Question & Answer with presenters
4:00p - 4:10p	**Public Comment Period**
4:10p - 4:30p	**Newborn Screening Advisory Committee Next Steps** ● Schedule next meeting
4:30p	**Meeting Close**

Public Comment Period (10 minutes)

- Moderator will announce comment period
- Use “raise hand” feature or dial *9
- Moderator will call your name
- Unmute yourself (if calling in dial *6)
- 2 minute max per comment
- Please email additional comments up to 1 hour after meeting ends to: HHSNewbornAdvisoryCommittee@mt.gov

Ground Rules

- Mute
- Video
- 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*"

Voting

- Only voting members who have submitted their COI statement can vote on Pompe
- Quorum = simple majority

Unfinished Business

- Voting Meeting
- Membership

Voting Meeting Procedure

Voting Considerations

- Voting members only
- Voting Options:
  - Recommend
  - Do not recommend
  - Do not have enough information to make a decision at this time

“Do not have enough information to make a decision at this time”

What does this mean?

Your final decision depends on specific information that you know is coming. The conversation is expected to continue at the next / upcoming meeting.

Quorum Confirmation

Vote on Pompe

Gaucher Nomination Packet Review

Packet Response Overview

Symptoms and age of onset

- Often diagnostic delays and misdiagnoses
- Low platelet count (thrombocytopenia) and enlarged spleen
- Complications include avascular necrosis, severe bleeding, chronic bone pain, sepsis, pathologic fractures, growth failure, and chronic liver disease

How is this disorder currently identified?

- Symptomatic presentation followed by a blood test

Why should it be screened at birth

- Early detection and management, can help mitigate some of these serious health risks and improve the quality of life for individuals with Gaucher disease.

Packet Response Overview

How is this disorder treated?

- Is there a treatment available?
  - Yes - FDA approved
- Is the treatment in the experimental phase?
  - No

Proposed screening test method

- Dried blood spot

Packet Response Overview

Status of condition in the United States

- States currently screening for the condition: 6 (IL, MO, NJ, NM, TN, OR)
- Condition has been reviewed by RUSP: No
- Registries or databases currently established for the condition: 2

Selection Criteria			
	True	Unsure	No
1. It can be identified at a period of time (24 to 48 hours after birth) at which it would not ordinarily be clinically detected.	X		
2. A test with appropriate sensitivity and specificity is available.	X		
3. There is a significant risk of illness, disability, or death if babies are not treated promptly (within the recommended time frame for the condition).	X		
4. Effective treatment is available and access to follow-up care and counseling is generally available.	X		
5. There are demonstrated benefits of early detection, timely intervention, and efficacious treatment.	X		
6. The benefits to babies and to society outweigh the risks and burdens of screening and treatment.	X		

Selection Criteria			
	True	**Unsure**	**No**
7. There are minimal financial impacts on the family.	X		
8. There is a public health benefit to conducting the test.	X		
9. There exist responsible parties who will follow up with families and implement necessary interventions.	X		
10. The condition's case definition and spectrum are well described.	X		
11. FOR LAB USE ONLY - The public health laboratory can support the testing resources and expertise necessary to provide accurate and timely results.			

Gaucher Nomination Packet Discussion

10 Minute Break

Clinical Background from Gaucher Subject Matter Experts:

Dr. Ozlem Goker-Alpan, MD

Gaucher Family Story:

Chris Heredia, Gaucher Advocate & Aviva Rosenberg, JD, Gaucher Community Alliance

GAUCHER COMMUNITY ALLIANCE

GAUCHER COMMUNITY
ALLIANCE

Chris Heredia

Parent of Gaucher Patient
&
Board Member

For Patients

By Patients

GAUCHER COMMUNITY ALLIANCE

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Questions?

Thank you!

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GAUCHER COMMUNITY
ALLIANCE

Aviva Rosenberg, JD
Co-Founder
Co-President

For Patients

By Patients

NEWBORN SCREENING FOR GAUCHER DISEASE

Aviva Rosenberg, JD

Co-Founder/Co-President

Gaucher Community Alliance

aviva@gauchercommunity.org

www.gauchercommunity.org

What is Gaucher Disease?

Gaucher disease is a rare genetic disorder caused by the lack of an enzyme call glucocerebrosidase (GCase), an enzyme that breaks down fatty substances in the body called glucocerebroside. Cells mostly accumulate in the spleen, liver, and bone marrow, but they may also collect in other tissues, including the lymphatic system, lungs, skin, eyes, kidney, heart, and in rare instances, the nervous system, or the brain.

Case for Newborn Screening

When GD manifests in childhood and remains untreated, it is often more severe and progresses more rapidly than disease that manifests in adulthood.

This emphasizes the importance of an early and definitive diagnosis followed by a comprehensive assessment of disease burden and regular monitoring of affected children.

Punita Gupta & Gregory Pastores (2018) Pharmacological treatment of pediatric Gaucher disease, Expert Review of Clinical Pharmacology, 11:12, 1183-1194

Case for Newborn Screening

- GD is a multisystem disease that leads to a spectrum of disease severity.
- Treatment is safe, effective and FDA approved.
- The disease burden and stage at treatment initiation can have a significant impact on the clinical outcomes. Despite extensive screening of symptomatic and at-risk individuals, patients with GD1 still present with irreversible complications such as osteonecrosis. Thus, asymptomatic screening and early intervention offer the best outcomes and possibility of “normalcy” rather than the expectation of living with the residual disease for the GD patients.

Ozlem Goker-Alpan, Margarita Ivanova, **Neuronopathic Gaucher disease: Rare in the West, common in the East,** Journal of Inherited Metabolic Disease, May 20, 2024

Case for Newborn Screening

People with GD3 who had received imiglucerase had improved symptoms after 5 years of treatment. They also had a greater chance of living longer, with 92% of people alive after 5 years of treatment. These results show that the ERT imiglucerase helps to improve blood, spleen, liver, and growth symptoms, and most importantly that it is a life-prolonging treatment.

‘Long-term hematological, visceral, and growth outcomes in children with Gaucher disease type 3 treated with imiglucerase in the International Collaborative Gaucher Group Gaucher Registry’ e journal Molecular Genetics and Metabolism. Feb. 6, 2024

States Currently Screening for Gaucher

- Illinois - 2014
- Missouri - 2013
- New Jersey
- Tennessee
- Oregon
- New Mexico
- Bill pending MA
- Applications under review PA, MT, IN, MN, GA

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Year	Babies Screened	GD Cases detected
2013	91,074	3
2015	91,551	3
2016	92,294	2
2017	90,489	2

Illinois Newborn Screening

- Initial 15 months of testing program, 219,973 dried blood spot specimens sent to Newborn Screening Laboratory of Illinois Department of Public Health in Chicago.

- 5 Gaucher disease cases detected.

J Pedatr 2017 Nov;190:130-135. doi: 10.1016/j.jpeds.2017.06.048. Epub 2017 Jul 17.

Tennessee Newborn Screening

- **From July 1, 2017 to December 31, 2018, 131,618 samples screened and 1 Gaucher case detected.**

New Jersey Newborn Screening

- **From July 1, 2019 to June 30, 2021, 188,075 samples screened and 7 Gaucher cases detected.**

New York Pilot Newborn Screening

- 65,605 samples screened by MS/MS over a 4 year period. 17 cases detected, 2 determined negative.

Wasserstein M, etal. GenetMed 2019

Thank you!

Thank you for listening and helping families learn valuable lifesaving information.

Questions?

Laboratory Background

Gaucher Cost Analysis

- Wisconsin does not test for Gaucher - but could ~$15/screen
- Montana PHL
  - Purchase Instrument:
    - Instrument $380,000.00 / 132,000 babies (10 years) = 2.88
  - Service contract $50,000/ 13,200 (1 year) = $3.79
  - Kits $6,930 x 15/year = $103,950 / 13,200 = $7.88
  - Total = $14.55/screen if we amortize the instrument over 10 years
- Gaucher can be a multi-plex with Pompe. If MTPHL begins testing Pompe in-house, the cost to add Gaucher will be minimal.
- This is an FDA-approved assay.

Gaucher Discussion

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Next Steps

- Follow Up from this Meeting
  - Meeting materials will be shared
  - Public website will be updated

- Next Meeting
  - Doodle Poll will be sent out to determine dates for Fall meeting
  - Will include Gaucher vote and Acid Sphingomyelinase Deficiency (ASMD) presentation

Follow Up & Thank You

Please email if you have any questions, comments, or need anything

HHSNewbornAdvisoryCommittee@mt.gov