

# Overview of Considering OTCD for Newborn Screening in Washington

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# The Five Criteria



1. Available Screening Technology
2. Diagnostic Testing and Treatment Are Available
3. Prevention Potential and Medical Rationale
4. Public Health Rationale
5. Cost Benefit/ Cost Effectiveness

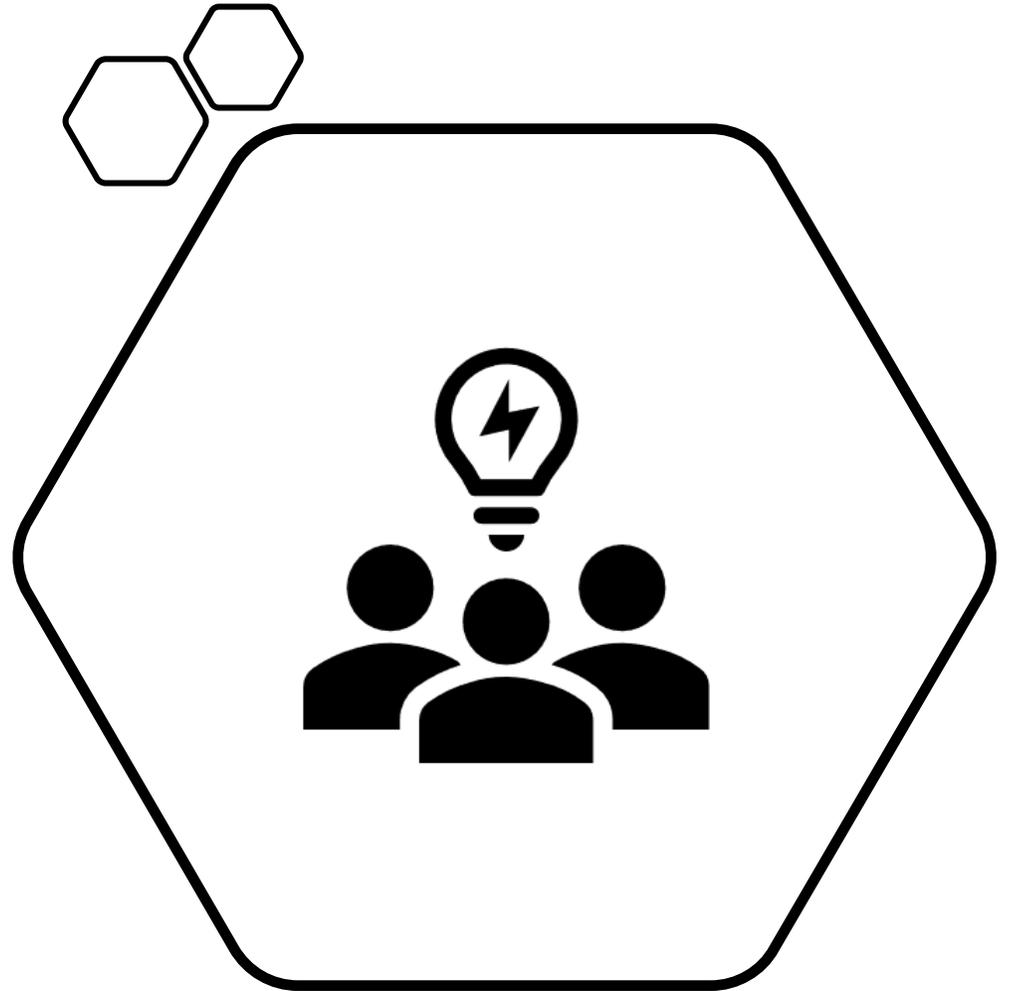
# Technical Advisory Committee



- Meeting 1 (06/16/2021): Assess and vote on the first four criteria
  - Presentations from BCG doctors, our program, and the family perspective
  - Votes:
    - Available Screening Technology: 12 Yes, 1 Unsure
    - Diagnostic Testing and Treatment Available: 12 Yes, 1 Unsure
    - Prevention and Medical Rationale: 11 Yes, 2 Unsure
    - Public Health Rationale: 12 Yes, 1 Unsure
- Takeaways:
  - Challenges of the virtual setting
  - Neary unanimous support for screening for the first 4 criteria

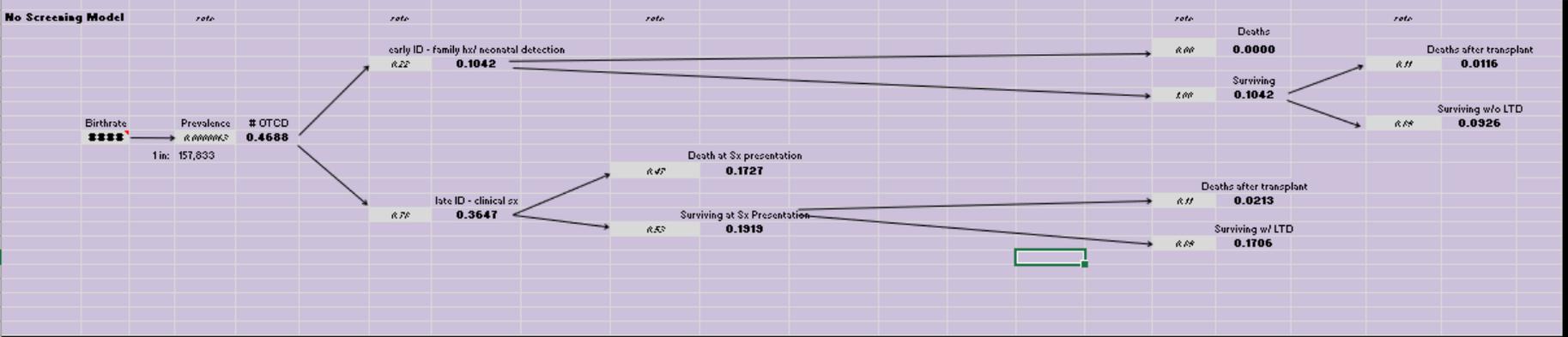
# Technical Advisory Committee

- Meeting 2 (07/07/2021): Vote on the 5<sup>th</sup> criteria and then overall
  - Presentation of Economic Analysis
  - Voting on criteria 5 and overall
  - Data from literature, existing NBS programs, expert opinion

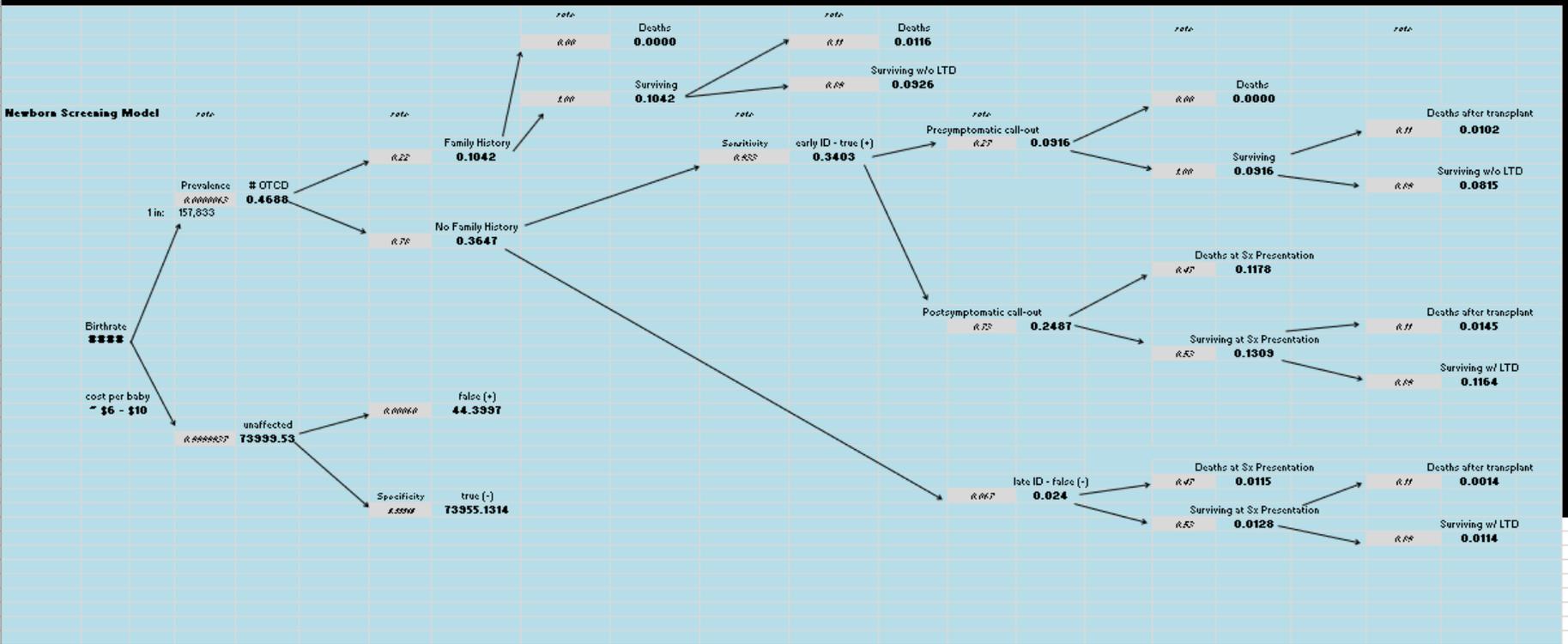


# Decision Tree Model

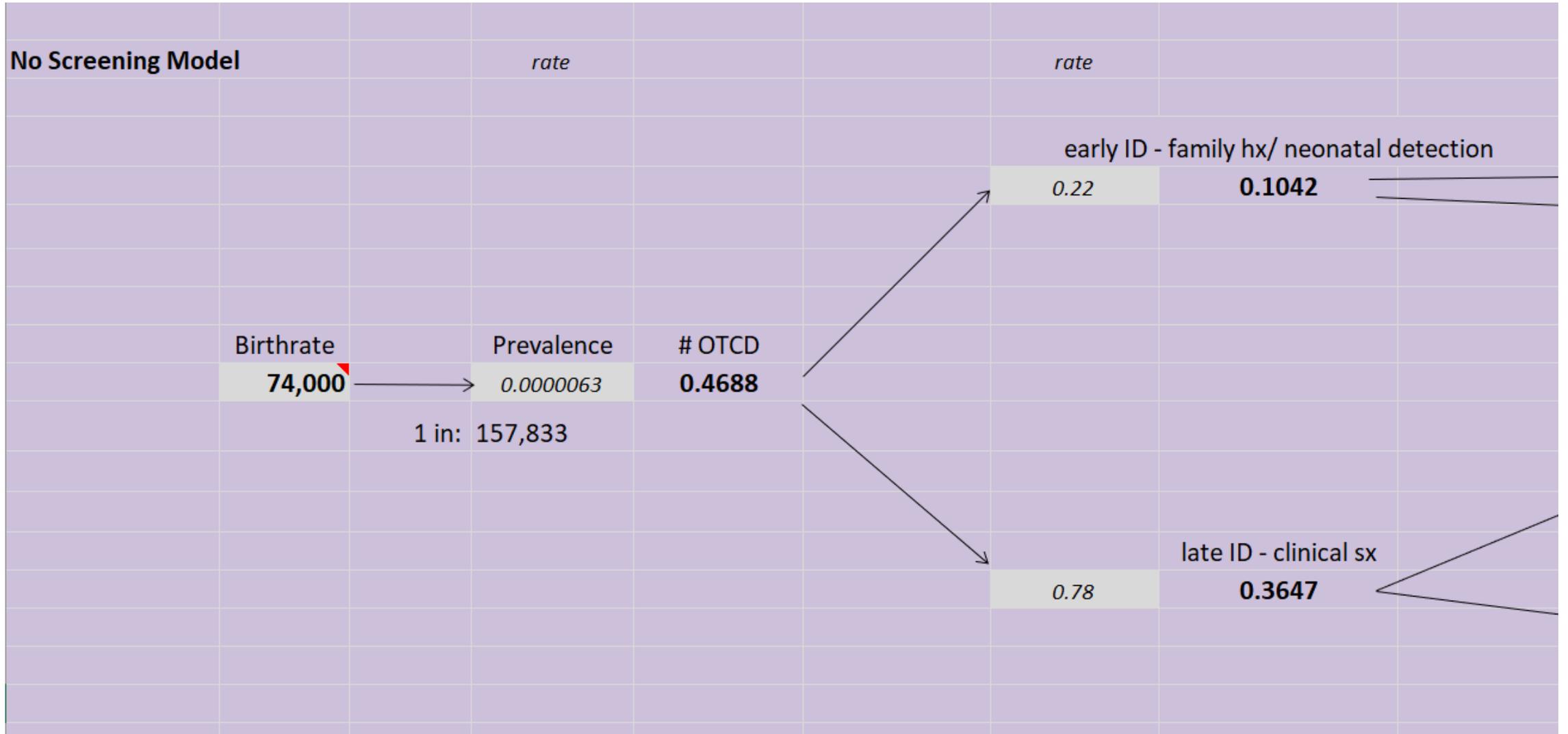
No Screening



Screening



# No Screening Model



# No Screening Model

<i>rate</i>			<i>rate</i>		<i>rate</i>		<i>rate</i>		<i>rate</i>
						Deaths			
	early ID - family hx/ neonatal detection				0.00	<b>0.0000</b>			Deaths after transplant
0.22	<b>0.1042</b>							0.11	<b>0.0116</b>
					1.00	Surviving			Surviving w/o LTD
						<b>0.1042</b>		0.89	<b>0.0926</b>

The flowchart illustrates the progression of a 'No Screening Model'. It starts with an initial rate of 0.22 for 'early ID - family hx/ neonatal detection', which results in a rate of 0.1042. This rate then branches into two paths: one leading to 'Deaths' with a rate of 0.00 and a count of 0.0000, and another leading to 'Surviving' with a rate of 1.00 and a count of 0.1042. The 'Surviving' path further branches into 'Deaths after transplant' with a rate of 0.11 and a count of 0.0116, and 'Surviving w/o LTD' with a rate of 0.89 and a count of 0.0926.

# Updated Shift: Benefits vs. Costs

SHIFT		
Benefits	Deaths averted	0.039
	LTD averted	0.043
	Value of LTD prever	\$ 64,303.58
	Value of a life	\$11,600,000.00
	Value of lives saved	\$447,552.89
	less tx costs	(\$75,580.28)
	TOTAL Benefits	\$436,276.18

Costs	costs of screening	\$ 594,241.00
	costs: false(+) dx	\$ 38,938.55
	TOTAL costs	\$ 633,179.55

Benefit/Cost ratio	0.69
net benefit	(\$196,903.37)

# Variability of Clinical Presentation

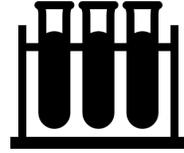


- We don't know if childhood or later onset forms of OTCD will be detected by NBS.

Neonatal Onset	Childhood Onset (1m-16yr)	Later Onset (> 16yr)
N= 27	N= 52	N= 11

Data from Brassier et al. 2015 study of 90 OTCD patients

# Ambiguous Results



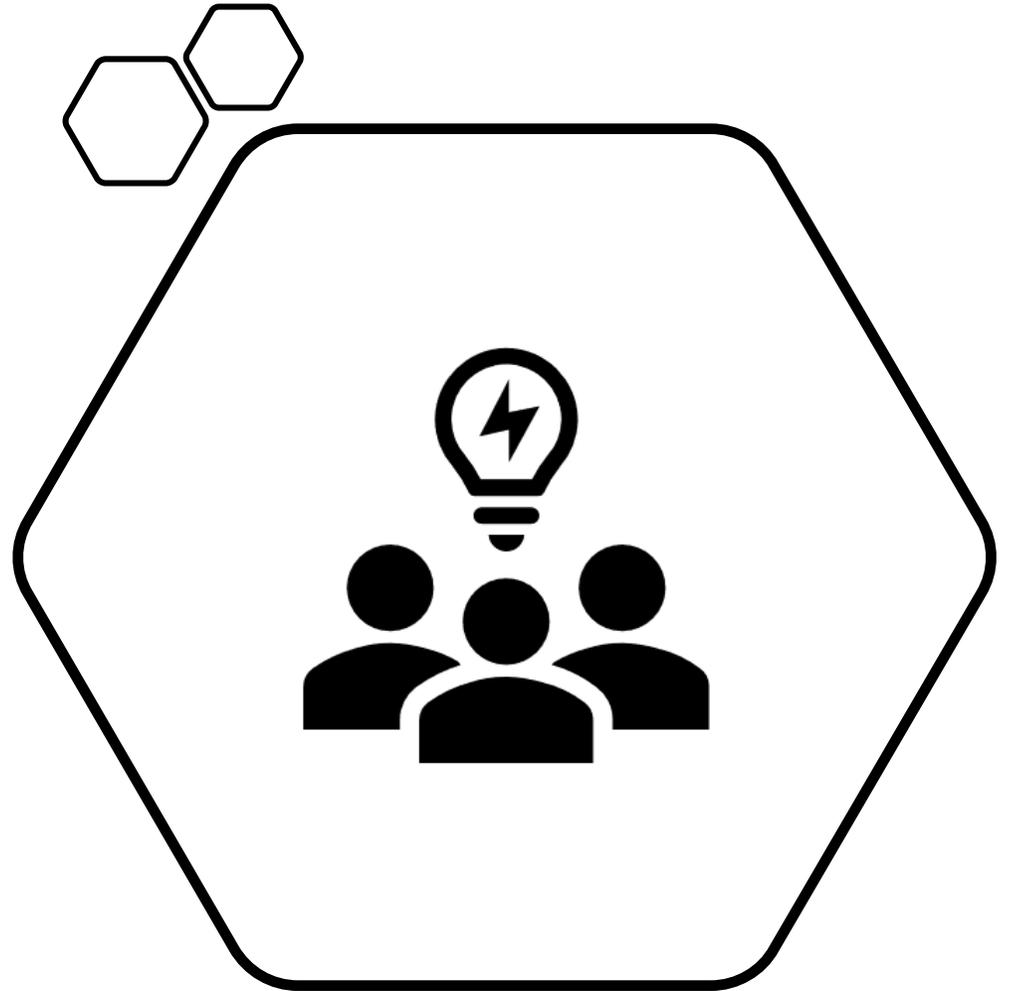
- Ambiguous results
  - Low Citrulline is not specific to OTCD
    - Other genetic disorders affecting the urea cycle (CPS1, NAGS)
    - Mitochondrial disorders
  - False positives
    - Prematurity, protein restriction, intestinal pathologies
    - Impact to families
    - Impact to the medical system

# Unintended Consequences of Screening

- Impact of the system failing
  - Expectation that screening will work
    - Families of babies with OTCD
    - Medical providers
    - Newborn screening staff
- Risk for litigation
  - What can the agency do to mitigate this risk?

# Technical Advisory Committee

- Meeting 2 (07/07/2021): Vote on the 5<sup>th</sup> criteria and then overall
  - Criterion Five votes: 7 Yes, 4 No, 2 Unsure
  - Overall (All Criteria): 9 Yes, 3 No, 1 Unsure



# SBOH Meeting

- Meeting (10/13/2021):
  - The Board heard the results of the TAC for OTCD
  - Board members engaged in discussion
- SBOH voted unanimously for the addition of OTCD to the mandatory panel



Questions  
and  
Discussion

