

# Methodological and policy issues related to the cost effectiveness of newborn screening in severe pediatric genetic disorders

*Presentation at ISPOR Educational Symposium*

Peter Neumann, ScD,

Director, Center for the Evaluation of Value & Risk in Health, Tufts Medical Center, Boston

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# Landscape for newborn genetic screening

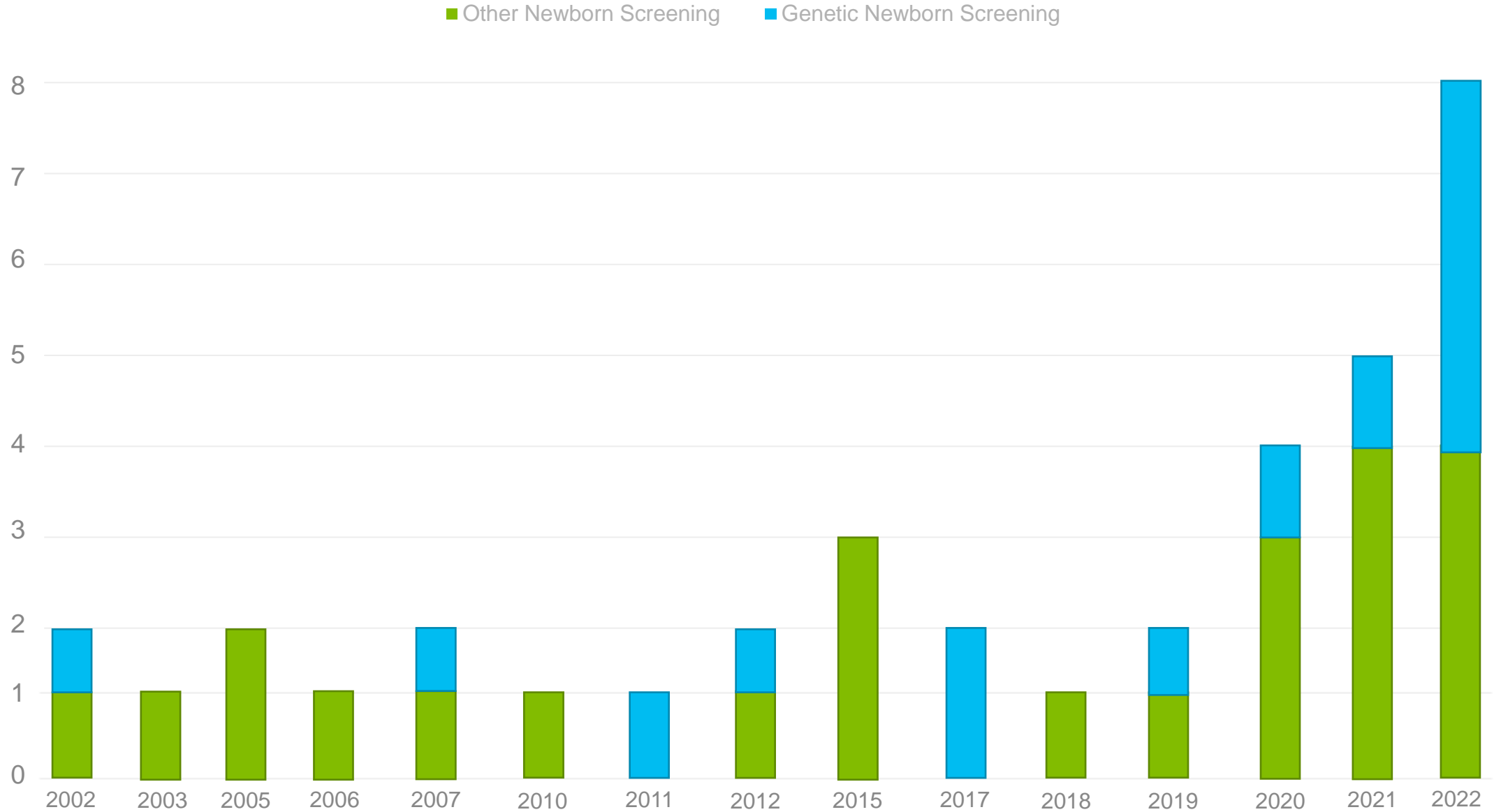
- Increasing ability to detect serious genetic conditions in newborns
- New therapies available/coming
- Which screening tests to prioritize?
- Role of the RUSP (Recommended Uniform Screening Panel)
- Delays in adding screening to the RUSP
- Even if added, delays in adoption by states – why?
- Underfunded/overworked public health labs
- How to identify benefits and harms of introducing a new program?

# The role of economic evaluation

- Shed light on costs and health outcomes of screening strategies
- Characterize long-term outcomes and uncertainty
- Generate evidence to support decision making at public health level



# Published cost-effectiveness analyses of newborn screening



Source: Tufts Medical Center CEA Registry. [www.cearegistry.org](http://www.cearegistry.org)



# CEAs of genetic screening

<b>By country</b>	<b>Number of CEAs</b>
USA	8
UK	3
Netherlands	3
Other	11
<b>By disease</b>	<b>Number of CEAs</b>
Endocrine, nutritional and metabolic	16
Nervous system	3
Other	5
<b>Inclusion of societal elements?</b>	<b>Number of CEAs</b>
Yes	6
No	18

Source: Tufts Medical Center CEA Registry. [www.cearegistry.org](http://www.cearegistry.org)

# Methodological challenges

- **What perspective and time horizon?**
- **Measuring benefits (potential harms) of screening:**
  - What outcomes?
  - Timing of screening
  - Assumptions about short and long-term consequences
  - Family “spillover” effects
  - “Value of knowing”
- **Measuring costs of screening**
  - What costs to include? (Medical and nonmedical)
  - Costs of false positives/false negatives
  - Consequence of not screening (costs of diagnostic odyssey)
  - Differences in costs across regions/countries
- **Data limitations**
- **Discount rate**
- **Equity/ethical considerations**

# Policy challenges

- How to prioritize conditions for screening?
- How to speed adoption of effective screening?
- How (when) to incorporating economic evaluation into decision processes?

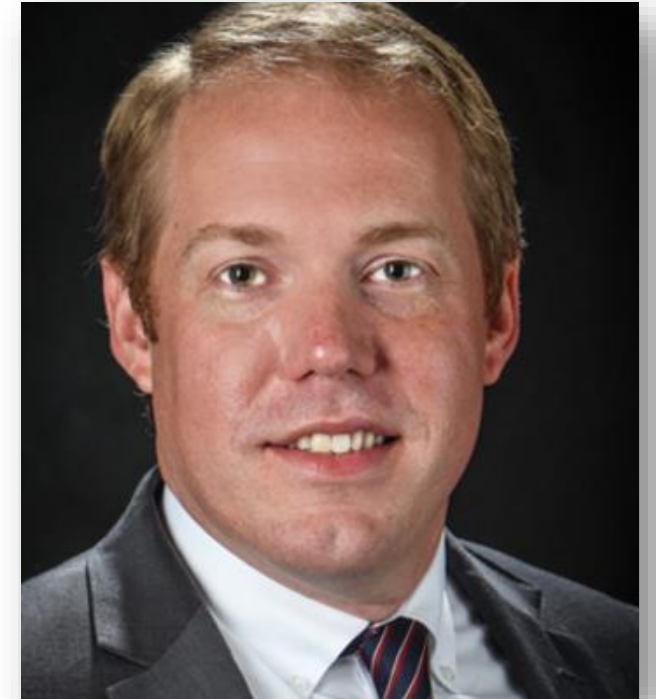
# Today's panelists



Jamie Sullivan  
*Vice President of Policy*  
EveryLife Foundation



Scott Grosse  
*Research Economist*  
Centers for Disease Control and Prevention



Bryan Amick  
*Associate Vice President  
of Government Contracts*  
Molina Healthcare