

PUBLIC DECISION-MAKING ABOUT NEWBORN SCREENING IN CONTEXTS OF TREATMENT, INTERVENTION, AND BENEFIT

What does this mean for CMV?

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The goal of **newborn screening** is to identify disorders in which early identification can change the course or outcome.

Newborn screening is justified by the existence of a defined medical **treatment** that must be provided early to be effective

Newborn screening should focus on whether meaningful **benefit** accrues as a result of early identification

NBS Past & Present

- Wilson & Jungner (1968)
 - Screen when prospects for **treating** the disorder “are at least reasonable.”
- Possible treatments
 - Drug therapy
 - Dietary interventions
 - Special social, medical, or educational services
 - “Management of the patient in relation to his total social situation and his immediate family and social group.”

Current Decision-Making

- State by state guidance from SACHDNC
- Recommended disorders make up the recommended uniform screening panel (RUSP).
 - Currently includes more than 50 disorders
- Additions are evaluated with criteria that assess:
 - Characteristics of the disorder
 - Screening and diagnostic measures
 - Potential for treatment
- *Most important consideration remains to the child being screened and the benefit accrued by that individual.*

- “**Benefit**” in NBS is construed in different ways across major reports and policy statements

Screening is appropriate when “there is evidence of substantial public **benefit** and acceptance. Sub-groups that may receive this **benefit** include infant, family, and society.”

-Committee for the Study of Inborn Errors of Metabolism, (1975)

Screening should occur only when there is strong evidence of **benefit** to the newborn at the earliest possible age.

- Institute of Medicine (1994)

Other types of benefit

- Elimination of the “diagnostic odyssey”
- Provision of reproductive risk information to parents
- Fostering research with affected children
- The developmental, psychological, and social benefits that occur from early disease detection.

What about CMV?

Why CMV?

- Important public health problem
- Incidence similar to combined incidence of all metabolic or endocrine disorders in the current US core panel
- There is a presymptomatic/early symptomatic stage
- Test would be generally acceptable to the population
- Much is know about natural history

Why not CMV? Benefit?

- Cannon et al., 2014: Categorized measurable potential benefits according to most common CMV disabilities
 - Hearing loss
 - Antiretrovirals (but primarily symptomatics)
 - Cognitive deficit
 - Antiretrovirals (but primarily symptomatics)
 - Vision impairment
 - Outcomes better when diagnosed early
 - Limited RCT antiretroviral evidence

But what does *benefit* mean to the public?

What falls under the category of *benefit* or *beneficial* for a non-medical audience?

Research Aims

- To better understand the characteristics of disorders that the public perceives as most beneficial in a screening context.
- To consider some of the terminology that is used in characterizing different disorders and their perceived benefits of inclusion on the newborn screen.

Methods

Survey development

- Survey consisted of **twelve scenarios** designed to represent disorders with different characteristics
 - Treatable?
 - Adult-onset?
 - Only reproductive information available?
 - Research-benefit only?
- 2 questions
- 7-point Likert scale

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NBS Treatment

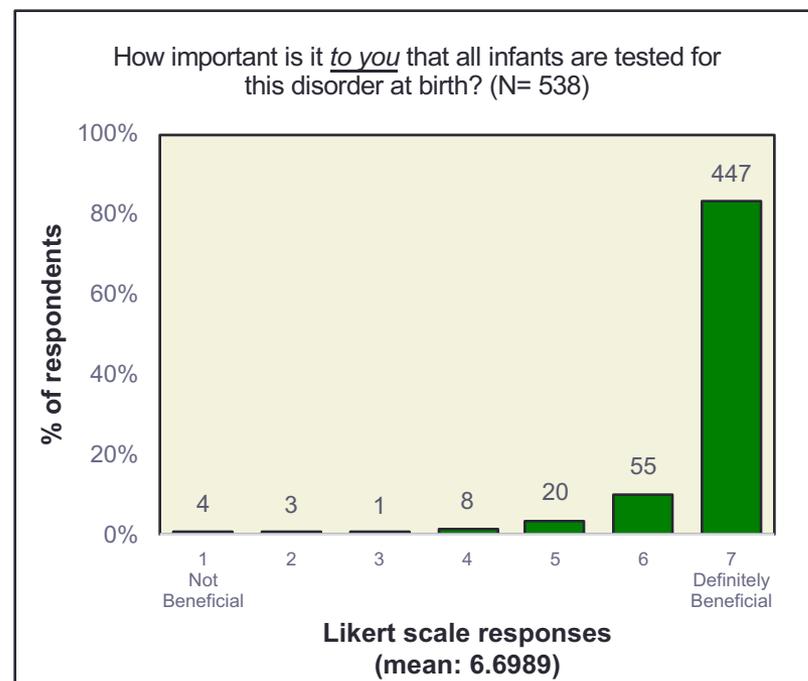
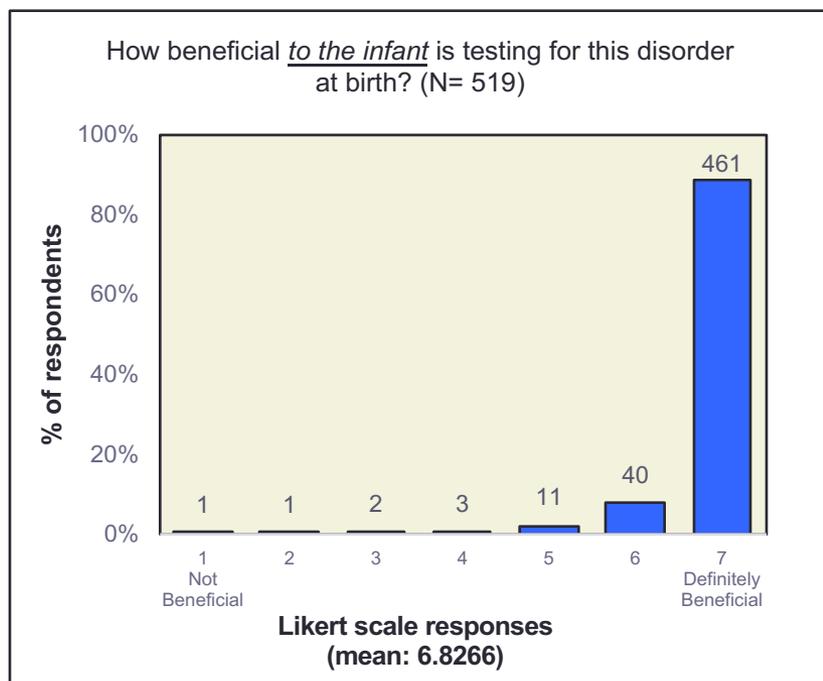
***1. In your opinion, how beneficial to the infant is testing for the following disorders at birth? Remember, in most cases, the child will *not* show signs of the disorder at birth.**

	Not beneficial	2	3	4	5	6	Definitely beneficial
If a child is found to have this disorder at birth, the child's parents would know that they themselves are at an increased chance of developing the disorder. Individuals with this disorder do not show signs of the disorder until their adult years.	<input type="radio"/>						
If a child is found to have this disorder at birth, the child may be able to receive treatment in the form of additional help from the state. This help may include, for example, monetary disability benefits or other supportive services.	<input type="radio"/>						
If a child is found to have this disorder at birth, the child can receive a treatment in the form of a special diet and will grow up with normal physical and mental abilities.	<input type="radio"/>						
If a child is found to have this disorder at birth, the child's parents could find out that future children have an increased chance of also having the disorder.	<input type="radio"/>						
If a child is found to have this disorder at birth, the child can receive treatment in the form of education that will help with his or her mental abilities later in life.	<input type="radio"/>						
Even if a child is found to have this disorder at birth, the child cannot receive any treatment until signs of the disorder appear.	<input type="radio"/>						
If a child is found to have this disorder at birth, the child's family will know of the diagnosis much sooner, but treatment cannot begin until signs of the disorder appear.	<input type="radio"/>						
If a child is found to have this disorder at birth, the child can be enrolled in a research study for experimental medication. There is no guarantee that individuals in the study will be saved by the experimental medication, but other affected individuals in the future may be saved.	<input type="radio"/>						
If a child is found to have this disorder at birth, the child can receive a risky treatment with the potential to add 3 years to the child's life. Most of this time will be spent in and out of the hospital.	<input type="radio"/>						
If a child is found to have this disorder at birth, the child can receive a treatment that will improve the child's physical abilities. The disorder's impact on the child's mental abilities cannot be reversed.	<input type="radio"/>						
If a child is found to have this disorder at birth, the child can receive a risky treatment with the potential to add 6 months to the child's life. Most of this time will be spent in and out of the hospital.	<input type="radio"/>						
If a child is found to have this disorder at birth, the child can receive a treatment that will improve the child's mental abilities. The disorder's impact on the child's physical abilities cannot be reversed.	<input type="radio"/>						

The Scenarios (a snapshot)

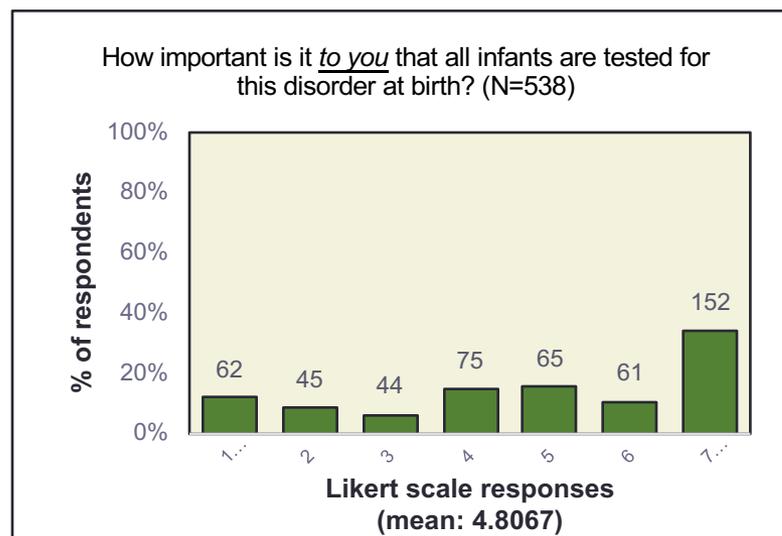
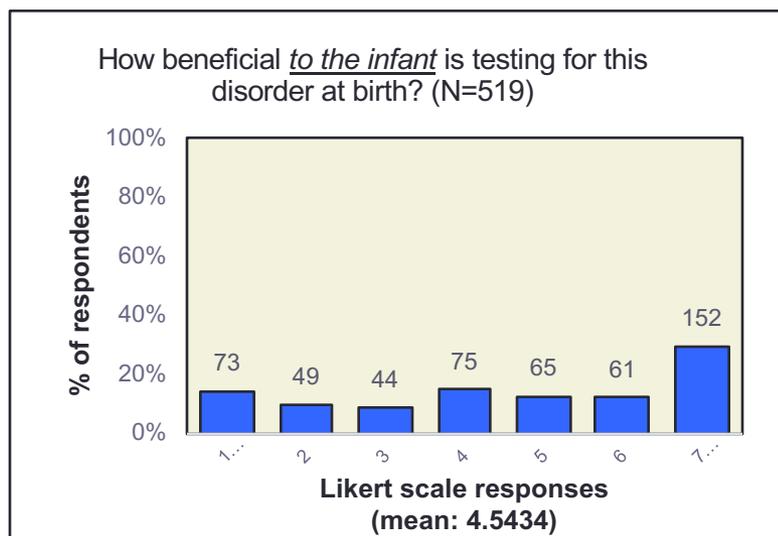
Scenario #1

- If a child is found to have this disorder at birth, the child can receive an **intervention/treatment** in the form of a special diet and will grow up with normal physical and mental abilities.



Scenario #2

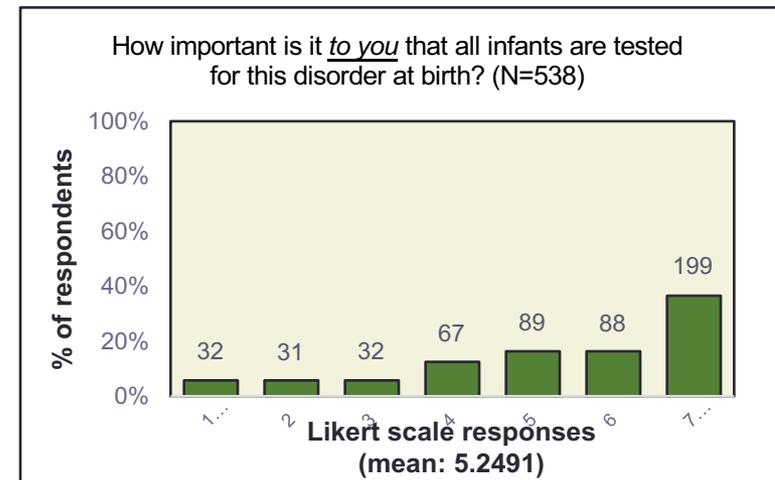
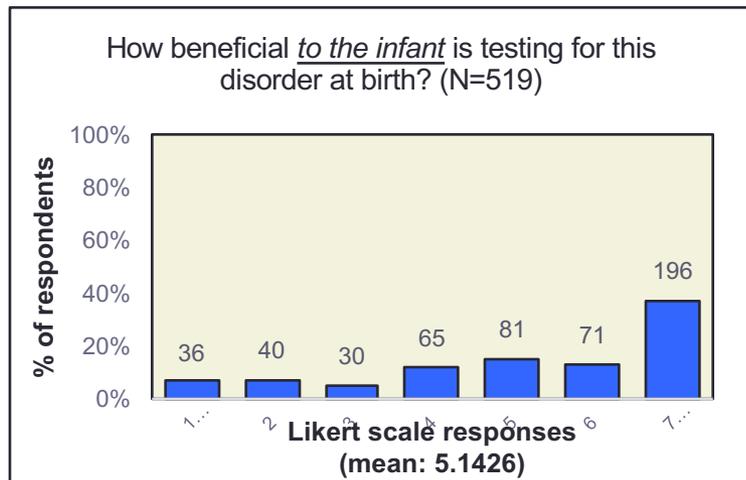
- *Even if a child is found to have this disorder at birth, the child cannot receive any **intervention/treatment** until signs of the disorder appear.*



- Versus usual identification
 - Parents less supportive (but still generally supportive) if you could not do something right away (Lipstein et. al., 2010)

Scenario #3

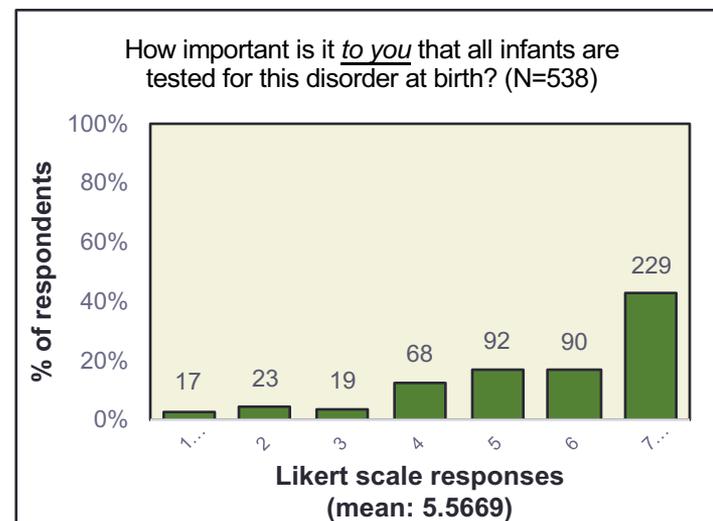
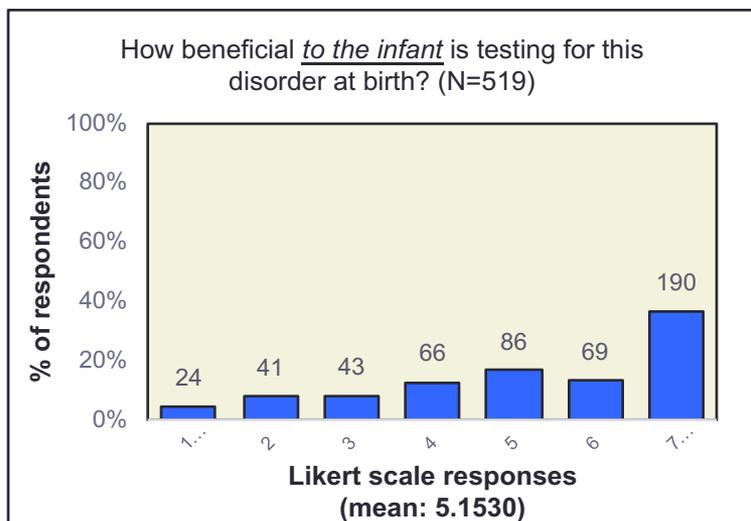
- *If a child is found to have this disorder at birth, the child's family will know of the diagnosis much sooner, but **intervention/ treatment** cannot begin until signs of the disorder appear.*



- *Same as prior scenario, emphasis on diagnosis time*
 - Lipstein et al., 2010
 - Early identification of disease in the absence of curative therapies was still seen as a benefit

Scenario #8

- *If a child is found to have this disorder at birth, the child can be enrolled in a research study for experimental medication. There is no guarantee that individuals in the study will be saved by the experimental medication, but other affected individuals in the future may be saved.*



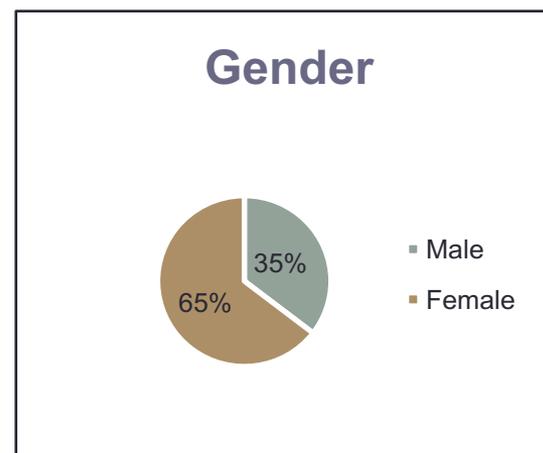
- Higher rating of importance to oneself ($p < 0.0005$)
 - Opportunity for further research

Demographic Effects & Analysis

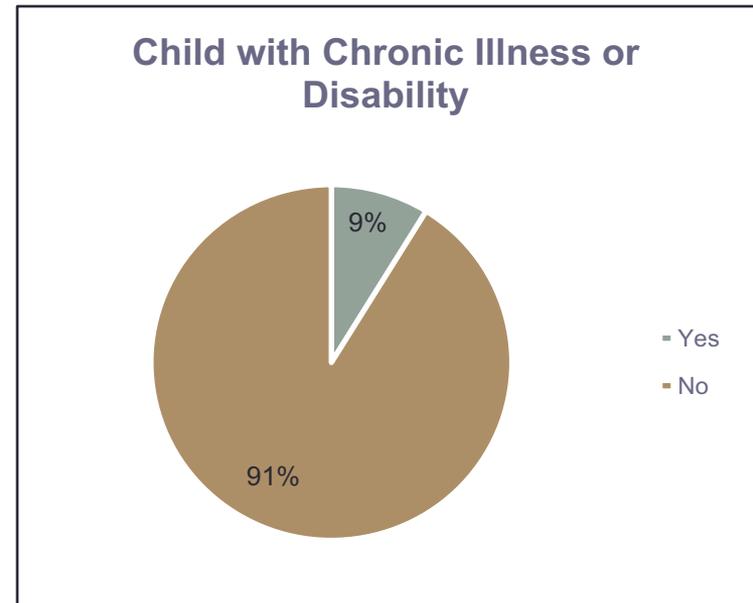
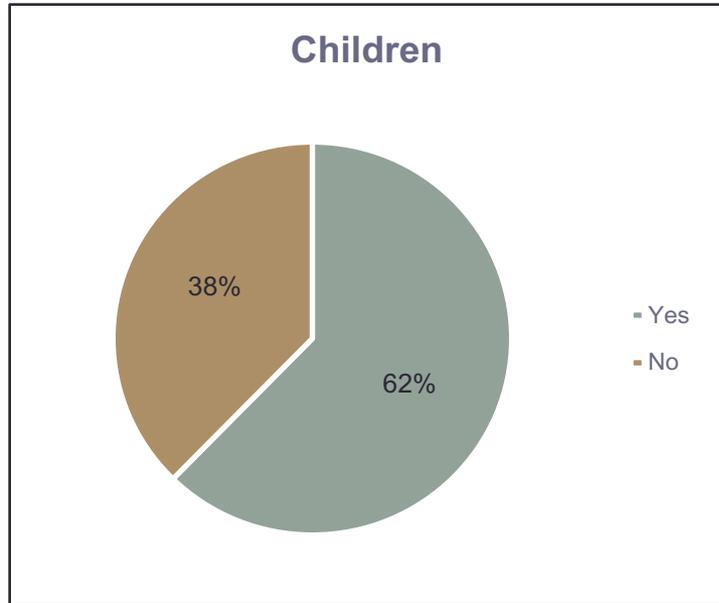
- Which demographic characteristics significantly affected how participants rated benefit and importance for each scenario?

Gender

- Females rated significantly higher benefit and importance in 12 of 24 scenario/question pairings
 - In every case, mean rank was higher for the female population
 - Consistent with other literature suggesting greater female support for expanded NBS
 - Goldenberg et al., 2013
 - Christie L., 2013



Children & Chronic Illness



Child/Child with chronic illness or disability

- Effect of “having a child” in this study dropped out when regression preformed with cohort with chronic illness or disability.
- Documented that those with children are more in favor of expanded screening
 - Plass et al., 2010
 - Most studies do not ask about health status of children
 - Families with a child with 2+ health conditions more likely to support WGS/NBS (Goldenberg et al., 2013).
- Important that future studies assess these nuances

Limitations

- Generalizability
 - Response rate 555/5840 (9.5%)
- Amount of information provided
 - Logistical limitations
 - Scenario & question interpretation
- Likert-scale assessment
 - Seven-point scale
 - Floor & ceiling effects
- Allowing for weighing of harms of testing

Thoughts, conclusions & future directions

- Cannon et al., 2014 conclusion → benefit from NBS CMV screening, early detection/intervention
 - Maximal benefit achieved with rigorous follow-up
 - Note that targeted screening may be better/more successful approach
 - Reduce potential harms (false positives)
 - Potentially reduce costs
- Targeted screening may be an easier pass
 - Screening success, methodologies, general cost can be better understood and early data can be gathered
 - Pave way for routing NBS, while helping individuals along the way

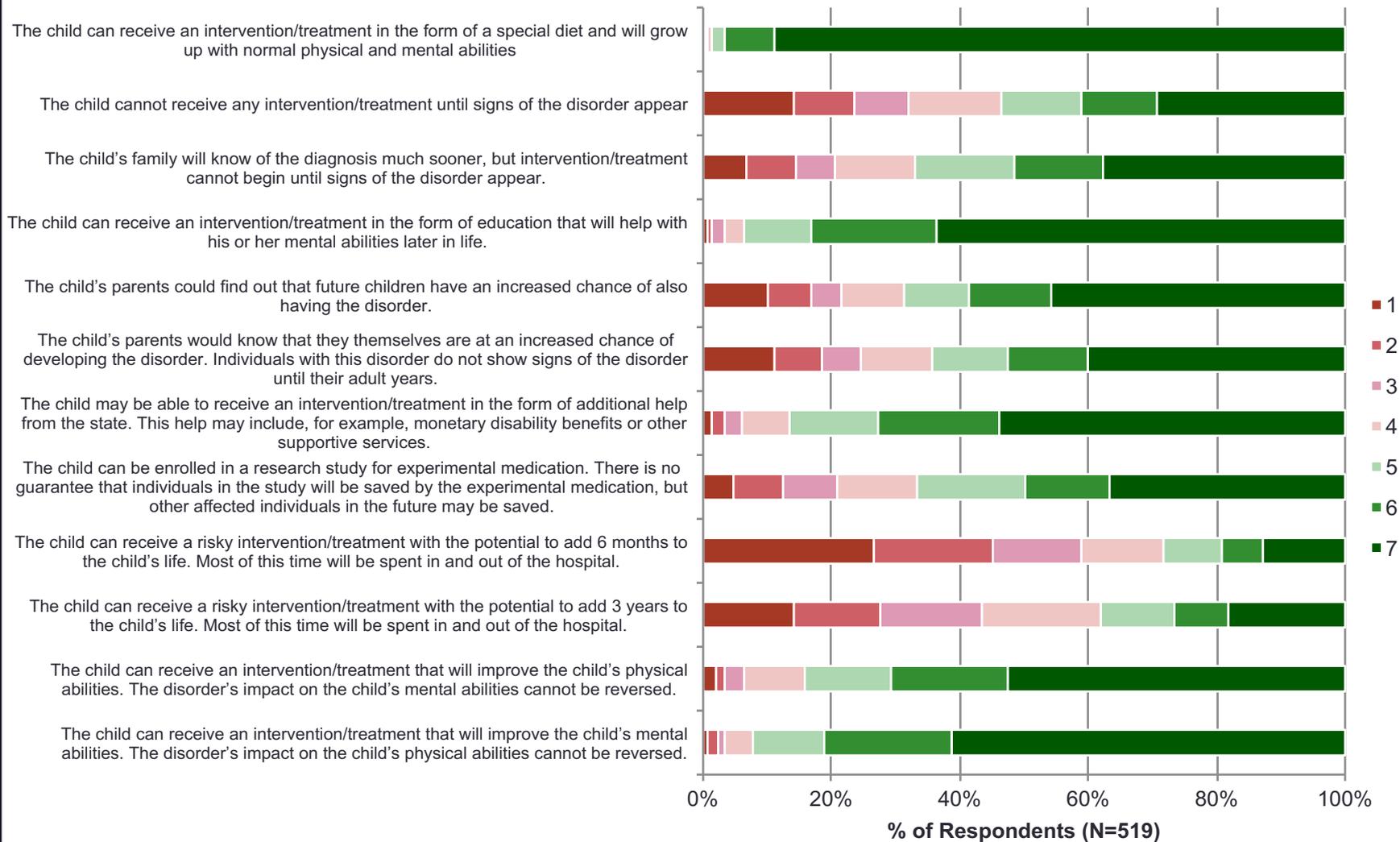
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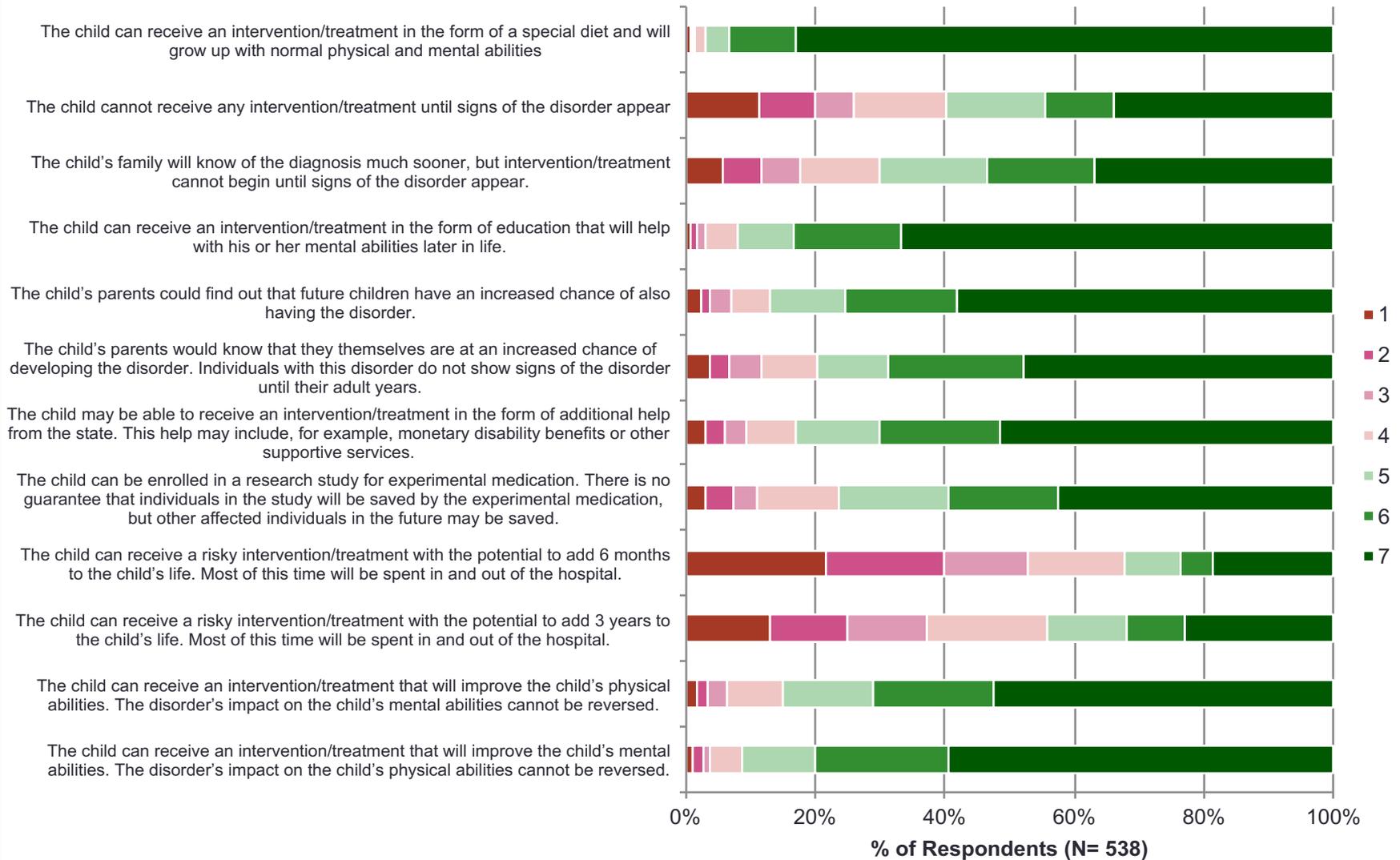
Acknowledgements

Extra Slides

How beneficial *to the infant* is testing for the following disorders at birth?



How important is it to you that all infants are tested for the following disorders at birth?



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