

ACHDNC Review of Research focusing on Lived Experience Perspectives

August 8, 2024

Background

- Stakeholders expressed need for patients' lived experiences be taken into consideration in its Committee processes
 - Lived experience, such as those expressed by public comments during ACHDNC meetings, can be translated into evidence and considered by the Committee
- Committee members and HRSA staff agree with our stakeholders, and we've worked to identify and schedule speakers for presentations on research that includes patient's lived experiences at multiple meetings since May 2023

Mind the Gap

May 2023

- Beth Tarini, MD, MS, MBA, Children's National Research Institute
- Described data gaps for the impact of false positives and uncertain prognoses with newborn screening.
- Summarized active NIH-funded research projects examining the impact of false positives and uncertain prognoses in newborn screening.
 - Mixed methods approach including qualitative research
 - Follow-up on results expected at a future ACHDNC meeting

Family Outcomes of Newborn Screening, January 2024

- Don Bailey, PhD & Elizabeth Reynolds, PhD & Melissa Raspa, PhD, RTI International
- Described previous research developing a tool to measure family outcomes of early intervention
 - HRSA funding current study to adapt this tool to family outcomes from newborn screening
 - Working with families to develop key concepts (e.g. quality of life)

Families' Search for Meaning and Value in Rare Genetic Diagnoses

January 2024

- Sara Ackerman, Ph.D., M.Ed., University of California, San Francisco
- Described findings from the Program in Prenatal and Pediatric Genomic Sequencing (P3EGS) 2017-2022
 - Examined the utility of genomic information from families
 - Explored the ethical and social issues of exome sequencing
 - Data collected through clinic observations and in-depth interviews with parents

The “Value of Values”: Expanding Assessment of Net Benefits and Harms through Social Science Data, January 2024

- Aaron Goldenberg, Ph.D., M.A., M.P.H, Case Western Reserve University School of Medicine
- Described the need for more data from the family perspective on newborn screening in order to expand notions of benefits and harms for newborns, families and society.
- Provided example of ScreenPlus NBS study in New York
 - Consented pilot NBS program
 - Gathered key stakeholder opinions to guide ethically sensitive decision-making about NBS expansion

Qualitative Evidence Synthesis: GRADE-CERQual Approach for Assessing the Confidence in Synthesized Findings, May 2024

- Jane Noyes, M.D., PhD, Professor in Health and Social Services Research and Child Health, Bangor University, Wales
- Described how qualitative research can be synthesized in an evidence review and used by decision-making groups
 - GRADE-CERQual approach for assessing confidence in qualitative findings
- Provided examples of how GRADE-CERQual has been used in developed evidence-based clinical guidelines
- www.cerqual.org

Summary of Research

- Mind the Gap
- Early Intervention and Newborn Screening
- Families' Search for Meaning and Value in Rare Genetic Diagnoses
- The “Value of Values”: Expanding Assessment of Net Benefits and Harms through Social Science Data
- Qualitative Evidence Synthesis: GRADE-CERQual Approach for Assessing the Confidence in Synthesized Findings

Conclusions

- There are established methods for translating lived experience into peer-reviewed research
- Such research can be synthesized in the evidence review for a condition nominated for the RUSP
- The voices of families can be included in an evidence-based decision-making process
- NEXT STEP:
 - Panel on resources available to fund research that includes the lived experience perspective

Discussion