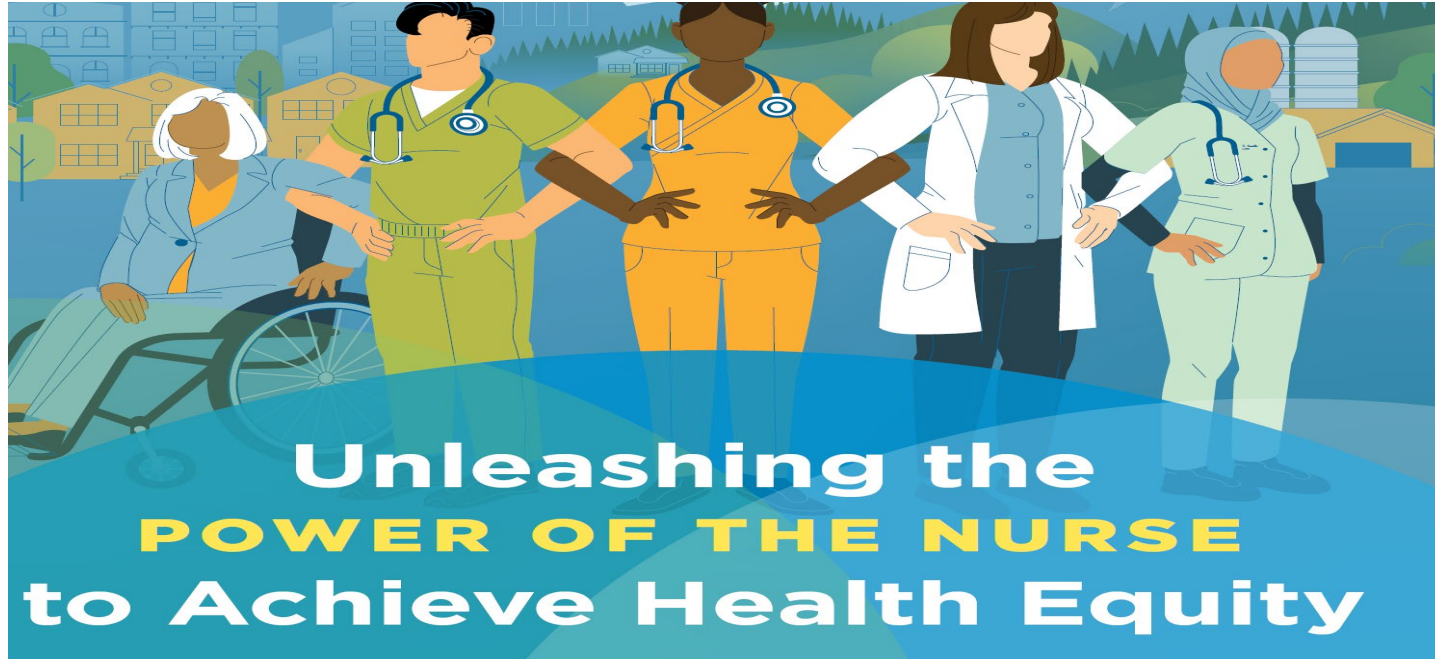


Safeguarding Newborn Screening in the Realm of Unbridled Genomic Possibilities



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Associate Professor

Disclosures



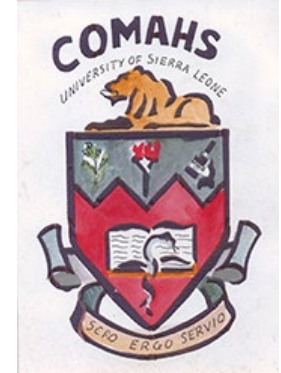
- There is no potential conflict of interest or financial interest to be disclosed

Back drop: From Ebola to Sickle Cell Disease





Sierra Leone Sickle Cell Disease Project



Next-Generation (NGS) Sequencing Newborn Screening Program: Ready for Prime Time?

THE JOURNAL OF PEDIATRICS • www.jpeds.com

EDITORIALS

Large Scale Next Generation Sequencing and Newborn Screening: Are We Ready?



Newborn screening (NBS) is a public health program that aims to identify children with medically actionable conditions prior to becoming symptomatic and provide interventions that result in improvement of long-term outcomes. Over the past decades, a number of conditions have been gradually added to the screening panels. Significant expansion of NBS happened in the mid-1990s to early 2000s when tandem mass spec-

See related article, p 68

method to help facilitate decisions about the potential use of genomic sequencing for NBS as well as assisting parents and physicians in making informed decisions about the disclosure of results. The authors convened a multidisciplinary committee and used previously published methods for assessing clinical actionability and adjusted the scoring rubric by incorporating age of onset and age at which interventions would begin. The scoring system was vali-

“Despite the increasingly attractive usage of NGS, its implementation as a general practice is therefore still premature with substantial challenges to be addressed, and it is unlikely to – and should not – replace present screening methods”.

Ding S, *Pediatr Investig* 2022

Review

> *Front Pediatr.* 2021 Jul 19;9:663752. doi: 10.3389/fped.2021.663752. eCollection 2021.

The Use of Whole Genome and Exome Sequencing for Newborn Screening: Challenges and Opportunities for Population Health

Audrey C Woerner¹, Renata C Gallagher², Jerry Vockley^{1,3}, Aashish N Adhikari^{4,5}

NGS presents the possibility of NBS for a wider set of disorders, many of which are rare and lack effective treatment

it will be difficult, if not impossible, to implement genomic newborn **screening** in a way that treats the population equitably.”

NBS – Communication Gap Problem



International Journal of
Neonatal Screening



Review

Psychosocial Issues Related to Newborn Screening: A Systematic Review and Synthesis

Audrey Tluczek ^{1,*}, Anne L. Ersig ¹ and Shinyo Lee ²

Psychosocial Issues

1. Parent Knowledge of and Attitudes about NBS and/or Dried Blood Spots
2. Parent Reactions to NBS Results
3. Parent Understanding of NBS Results
4. Parent Education Following Confirmed Diagnosis
5. Communication
6. Parent Decisions about Carrier Testing and Future Pregnancies
7. Child and Family Outcomes
8. Disparities

Recommendations

1. Improve parents' general knowledge of NBS
2. Improve parents' understanding of NBS positive results and next steps
3. Reduce parents' emotional distress and ensure that their understanding of diagnostics test results and related implications.
4. Support families beyond the NBS process

Sustaining NGS-NBS will Require Public Health Communication Adjustments and NBS Champions

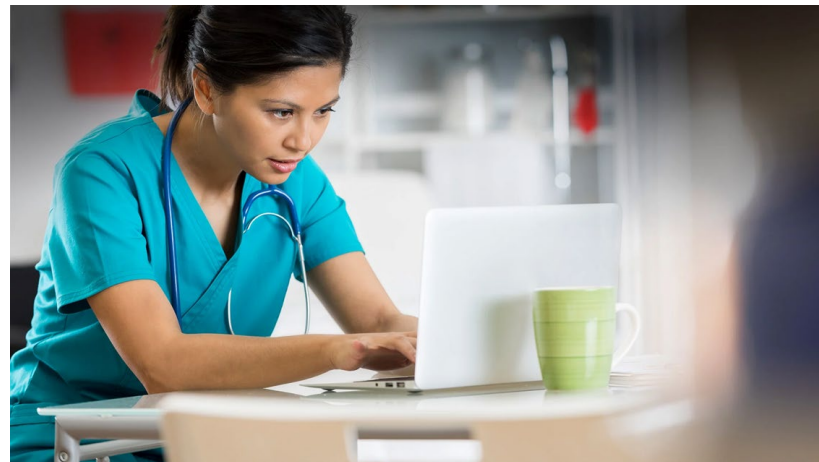
- 1. On a public level – NGS-NBS would require a **structure** that would facilitate careful and considerable communication efforts to engage future new parents and stakeholders in conversation about **engagement** in the current opt-out system used for NBS.
- 2. On an individual level – NGS-NBS communication efforts would need to address parental expectations and understanding before testing, and interpretation of the newborn's genome sequencing findings. Conversation efforts should **empower** parents and engage them in decision-making and ensure parents have support on deciding how to act on genomic results.

We Need NGS-NBS Nurse Champions

Department of Health and Human Services

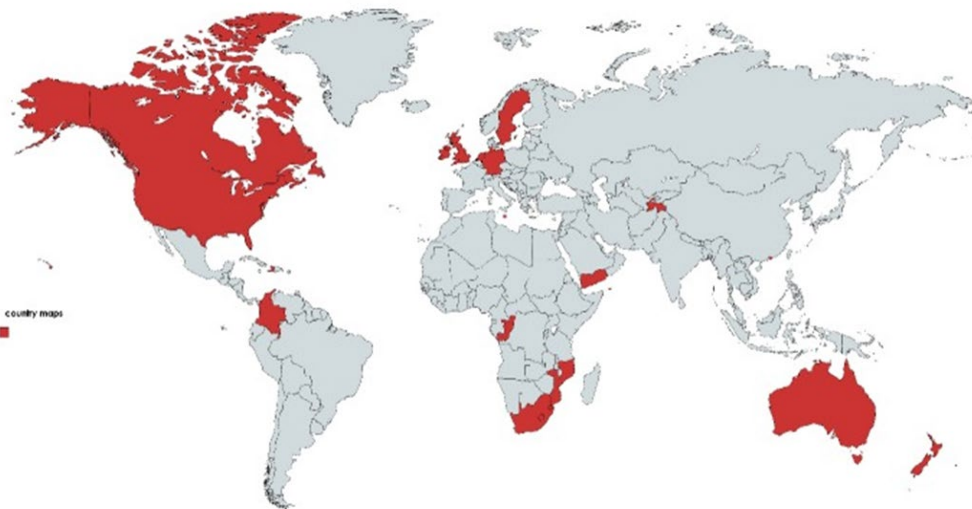
Part 1. Overview Information

Participating Organization(s)	National Institutes of Health (NIH)
Components of Participating Organizations	National Cancer Institute (NCI) National Human Genome Research Institute (NHGRI)
Funding Opportunity Title	Investigator-Initiated Research on Genetic Counseling Processes and Practices (R01 Clinical Trial Optional)
Activity Code	R01 Research Project Grant
Announcement Type	New
Related Notices	• September 01, 2020 - Notice of Applicant Information Webinar and Frequently Asked Questions (FAQs) for Investigator-Initiated Research on Genetic Counseling Processes and Practices Request for Applications (RFAs): RFA-HG-20-048 and RFA-HG-20-049. See Notice NOT-HG-20-057 .
Funding Opportunity Announcement (FOA) Number	RFA-HG-20-048



- ☐ In the USA, nurses are uniquely placed to reach large segments of the population. With their reputation as a trusted source of health information, they could provide pre-test counseling, testing, and treatment across the care spectrum.
- ☐ Upskilling – **Equipped nurses with technical assistance, context-specific training, and resources to provide care and coordinate follow-up care for prompt management of disease presentations.**
- ☐ “Task –shifting” – **provide specific clinical service previously provided by other clinicians or laboratory technicians.**

Nurse Champion Model – Evidence Based Model



The Nurse Champion Model for Advancing Newborn Screening of Critical Congenital Heart Disease

Implementation of the Champions for Skin Integrity model to improve leg and foot ulcer care in the primary healthcare setting

HEALTHCARE DELIVERY

A nurse-led intervention to improve management of virological failure in public sector HIV clinics in Durban, South Africa:
A pre- and post-implementation evaluation

Courtesy with permission of the author

Impact of nurse champion on quality of care and outcomes in type 2 diabetes patients

Madison Wilson BSN, Huey-Shys Chen PhD, RN, MCHES, FAAN and Marilynne Wood PhD, MSN, RN
College of Nursing, The University of Toledo, Toledo, Ohio, USA



Cochrane Database of Systematic Reviews

Nurses as substitutes for doctors in primary care (Review)

Barriers and facilitators to the implementation of doctor-nurse substitution strategies in primary care: a qualitative evidence synthesis (Review)

Why Nurse Champions for Expanded Newborn Screening



ANA  ENTERPRISE

The International Society of Nurses in Genetics (ISONG) (International Society of Nurses in Genetics, 2012) issued a position statement on the role of nurses in the newborn screening process in 2012, which recommends the following for nursing:

- Active participation in current knowledge of NBS as an advocate for patients and families,
- Provide patient education prior to testing for NBS,
- Offer counseling to families post testing,
- Explain use of results, and if being utilized in research studies, explain their benefits, and
- Include information about NBS in newborn care curriculum for patient education.

Review > J Nurs Scholarsh 2013 Mar;45(1):25-33. doi: 10.1111/jnu.12005. Epub 2013 Jan 31.

Implications of newborn screening for nurses

Jane DeLuca ¹, Karen L Zanni, Natasha Bonhomme, Alex R Kemper

Affiliations + expand

PMID: 23368429 DOI: 10.1111/jnu.12005

> Nurs Res. 2017 Mar/Apr;66(2):198-205. doi: 10.1097/NNR.0000000000000211.

Genome Sequencing Technologies and Nursing: What Are the Roles of Nurses and Nurse Scientists?

Jacquelyn Y Taylor ¹, Michelle L Wright, Kathleen T Hickey, David E Housman

Affiliations + expand

PMID: 28252579 PMCID: PMC5334658 DOI: 10.1097/NNR.0000000000000211

Free PMC article



Expanded Newborn Screening

Challenges to NICU Nurses

Rachel A. Joseph, PhD, CCRN



Genomic NBS Education Curriculum for Nurse Champions

> *Genet Med.* 2019 Mar;21(3):525-533. doi: 10.1038/s41436-018-0101-x. Epub 2018 Aug 13.

Including ELSI research questions in newborn screening pilot studies

Aaron J Goldenberg¹, Michele Lloyd-Puryear², Jeffrey P Brosco³, Bradford Therrell⁴, Lynn Bush⁵, Susan Berry⁶, Amy Brower⁷, Natasha Bonhomme⁸, Bruce Bowdish⁷, Denise Chrysler⁹, Angus Clarke¹⁰, Thomas Crawford¹¹, Edward Goldman¹², Sally Hiner¹³, R Rodney Howell¹⁴, David Orren¹⁵, Benjamin S Wilfond¹⁶, Michael Watson⁵;
Bioethics and Legal Workgroup of the Newborn Screening Translational Research Network



Advancing Genomic Research and Reducing Health Disparities: What Can Nurse Scholars Do?

Cheedy Jaja, PhD, MPH, MN, RN¹, Robert Gibson, PhD, MSOTR/L², Shirley Quarles, EdD, RN, FAAN³

¹ Psi-ot large, Assistant Professor, Georgia Health Sciences University, College of Nursing, Augusta, GA
² Associate Professor, Georgia Health Sciences University, College of Allied Health, Augusta, GA
³ Associate Professor, Georgia Health Sciences University, College of Nursing, Augusta, GA

Discussion Paper

Whole exome or genome sequencing: nurses need to prepare families for the possibilities

Cynthia A. Prows RN, MSN CNS FAAN ✉ Grace Tran MS, Beverly Blosser MS

First published: 01 September 2014 | <https://doi.org/10.1111/jan.12516> | Citations: 8

Issues Related to NBS Results
<ol style="list-style-type: none">1. What are the potential ELSI of positive screening results related to a new condition?2. What are the potential ELSI implications of false-positive screening results related to a new condition?3. What are the potential ELSI of false negative screening results related to a new condition?4. What are the potential ELSI of obtaining and reporting carrier status related to a new condition?5. What are the potential ELSI of indeterminate results related to a condition?
Issues Related to NBS System
<ol style="list-style-type: none">6. What are the cost or resource allocation implications for adding a new condition to the RUSP or a state panel?7. What are the health disparities or equity considerations related to adding a new condition to the RUSP or a state panel?8. What are the potential implications for public/parental trust in the NBS system or health department that might arise because of adding a new condition?9. Does a condition raise any concerns regarding parental permission or challenges to the ethical or social justification for requiring population-based screening?



Precision Medicine Framework for NBS Genomic Communication

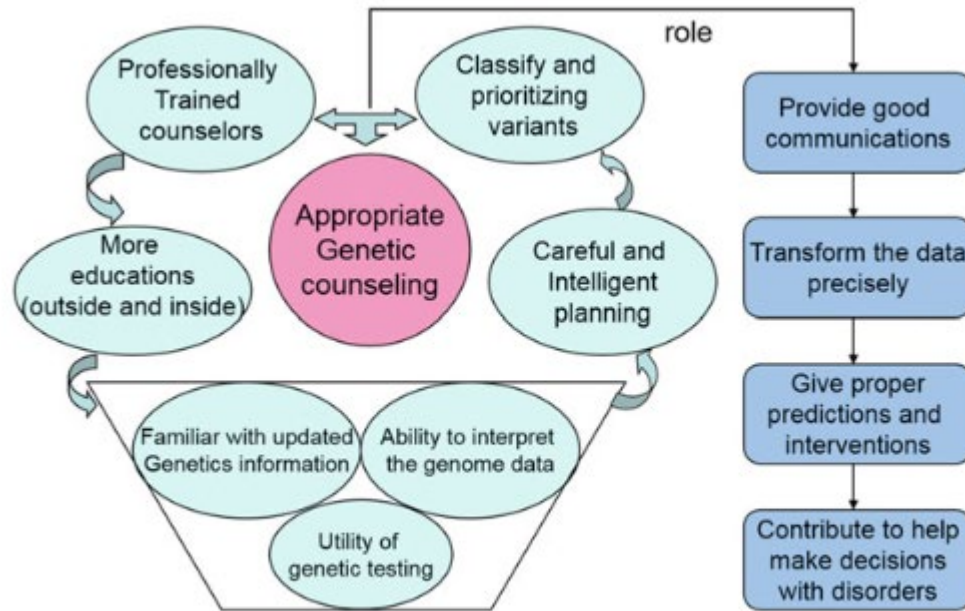


Fig. 3.2 The role of genetic counseling and education in NBS in the era of precision medicine



Expanding Access to Sickle Cell Disease Care in Sierra Leone –

- 1. Nurse Champion Model for Sickle Cell Newborn Screening.**
- 2. Nurse Champion Model for SCD Trait Genetic Counseling**
- 3. Nurse Champion Model for SCD Primary Care**

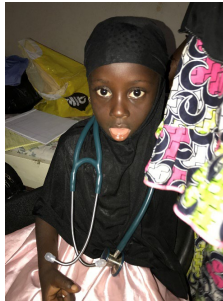
First Newborn Cohort Screened for Sickle Cell Disease in Sierra Leone



Sickle Cell Disease Presentation in Primary Care Cohort



Establishing routine wellness care for children with SCD



Nurses' Mission – Advancing Equity, Just Society, Good Health & Dignified Lives

The Future of Nursing 2020-2030

Charting a Path to
Achieve Health Equity

Our mandate is to unleash
our nursing prowess to
advance just societies, health
equity, and dignified lives

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“A nation cannot fully thrive until everyone - no matter who they are, where they live, or how much money they make - can live their healthiest possible life.”