

# Developing NBSChat: A Digital Education Tool for Sickle Cell Trait Follow-Up

December 10, 2025 1:00 - 2:00 pm ET

### **Moderator**

 Courtney Sumby, MS, Association of Public Health Laboratories Senior Specialist

### **Today's Webinar**

This webinar will highlight the development and evaluation of NBSChat, a scripted chatbot designed to provide accessible education and emotional reassurance for parents receiving SCT results. Learn how participatory design and mixed-methods research informed this innovative digital health intervention aimed at improving NBS follow-up care. The session will include a live demo of NBSChat, showcasing its features and usability.



### **Objectives**

- 1. Summarize the participatory design process used to develop a digital health intervention resource for newborn screening follow-up.
- 2. Describe how NBSChat addresses parental information needs and emotional reassurance following sickle cell trait results.
- 3. Discuss preliminary usability findings and implications for integrating digital tools into NBS follow-up care

### **Speakers**

- Karen Eilbeck, PhD, Professor, Biomedical Informatics, University of Utah
- Courtney Gauchel, RN-BSN, MS, APHL Newborn Screening Fellow, University of Utah

# Participatory Design of a Chatbot for Sickle Cell Trait Newborn Screening Results

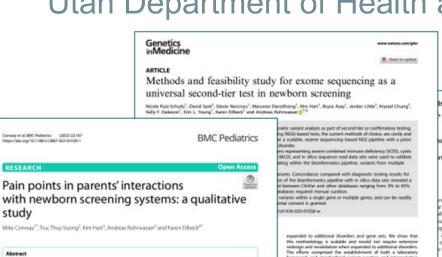
#### **University of Utah**

Courtney Gauchel, MS; Guilherme Del Fiol, MD, PhD; Kim Kaphingst, ScD; Anne Madeo, MS, Karen Eilbeck, PhD



## The initial spark for the project

### Utah Department of Health and Human Services



study

Conway et al. BMC Pediatrics (2002) 22:160 https://doi.org/10.1186/v/2887-022-03160-1

Background & Objectives: This study aims to evolves and elucidate parents experience of newborn street [HBS], with the overarching goal of identifying desiderata for the development of informatics-based educational and

Methods: We conducted four focus groups and four one-on-one qualitative inserviews with a total of 35 participants between March and September 2020. Participants were grouped into three types: parents who had received our positive newborn covering results; parents who had received folly positive results; and soon-to-be parents who had no direct experience of the screening process. Interview data were subjected to analysis using an inductive, constant

Results: Results are divided into five sections: (1) experiences related to the process of receiving NBS results and prior knowledge of the N8S program; (2) approaches to the management of a child's medical data; (3) sources of additional informational and emotional support; (4) barriers faced by parents ravigating the health system; and (5) recommendations and suggestions for new parents experiencing the NBS process.

Conclusion: Our analysis revealed a wide range of experiences of, and attitudes towards the newborn screening program and the wider newborn screening system. While parents' view of the screening process was - on the whole positive, some participants reported experiencing substantial flustration, particularly related to how results are Initially communicated and difficulties in accessing seliable, timely information. This frustration with current information management and education resources indicates a role for informatics based approaches in addressing parents' information needs.

#### Introduction & Background

Newborn Screening (NBS) allows for the early and often and medical care providers [1-3]. Newborn screening pre-symptomatic identification and diagnosis of treatable disorders in newborns. Rapid intervention prevents potentially severe complications. NBS is a system



includes a wide range of conditions including hearing

congenital heart defects, endocrine disorders, hemo

globin disorders, inhorn errors of metabolism, cystic

fibrosis, spiral muscular stropby, hysosomal storage dis-

orders, and immunodeficiencies. While the first two

screening modalities are point of care screening events, endroceine, hemoglobin, and metabolic screening entail

ized laboratories. In the United States, for metabolic screening, the newborn screen blood sample is collected

framework and standardized varient curation and interpretation processes. We developed and validated a laboratory method using two 3.2 mm diled blood spet (285) purches. We show similar turnaround time and cost impact compared with Sanger and amplicon-based NGS tests and we show that in contrast to Sanger and amplicon sequencing this methodology is highly scalable. A key component of the genomics approach is the postsequencing analysis that enables us to provide competitive turnaround times. Within the bioinformatics pipeline, we have turnacional stress. Within the bisoriformatics pipeline, we have constell multiple varient necessities to all the climical team in varient-impact interpretation. This cursiston work demonstrates the wide distribution of knowledge pertaining to disease-causing varients, and we perside a generic suite of tools that can be implemented in other disorder cases.

Detailed information for all methods is available in Supplementary materials and methods.

Days 1 sammarism the ES straigs. Supra Blooks were passented from

DNA extracted from two 3.2 mm DBS punches using Bureina's Nextens DNA First Dried Blood Spot Extraction Protocol Guide and Nextens First for

sales, Onloaning of Dulk, Salt Lake City, UK, USA, Manual: professor

is and Genomics 2021

tsMyWay: combining Fast Healthcare Interoperability Resources , Clinical Quality Language (CQL), and informational resources to create a newborn screening application

ael Watkins<sup>1</sup>, Alex Au, MD<sup>1</sup>, Truc Vuong<sup>1</sup>, Heidi Wallis<sup>2</sup>, Kim Hart, MS<sup>2</sup>, Andy Rohrwasser, Ph.D., MBA3, Karen Eilbeck, Ph.D. tment of Biomedical Informatics, University of Utah, Salt Lake City, Utah; <sup>2</sup>Utah Department of Health, Salt Lake City, Utah; 3Utah Public Health Laboratory, Taylorsville, Utah

ning (NBS) can be life-changing for the families of infants who test positive for a rare condition. While t to support these families, there can be delays in sharing these resources due to communication lag oratory, result interpreting clinician, family of the newborn, and additional care providers. This delay acerbated when additional health history is required from the mother and infant. ResultsMyWay is a et application that uses Clinical Ouality Language (COL) to automate the search for this additional It also translates the NBS results into Fast Healthcare Interoperability Resources (FHIR), increasing of exchange and the future utility of these data points. After the families are given the NBS results, then acts as a hub for several types of informational resources about the recently diagnosed condition.

ning (NBS) is a public health program that screens infants shortly after birth for diseases and disorders ly harm the infant if early detection and care management are not provided. Of the approximately four s born per year in the United States, about 12,900 of them are diagnosed with a disorder via the NBS

a positive rare condition result, the guardians of the infant are contacted to inform them of the result the assurance that qualified professionals will be in contact to help establish an appropriate care plan Difficulties in the delivery of information to the care providers and the guardians can lead to problems follow-up2, distress4, confusion and strong emotional reactions2. Parental knowledge of the condition knowledge gap is another pain point that must be addressed. The internet provides an opportunity for tion about NBS screening with health department web pages', and there are resources such as Baby's aim to collate information. But a Google search of 'nbs cystic fibrosis test result' brings back 142,000 s obviously overwhelming.

is a routine clinical service, there are still improvements that can be made in the typical NBS workflow here is the need, in many cases, to manually collect additional health information about the mother and asses where the interpretation of the results could change depending on the presence or absence of these ers. This collection is a burden to the interpreting clinician who now must wait for what can be a slow tion exchange process, to the guardian who now knows that something isn't right but must continue to fied results are returned, and for the care provider of the mother who must search through that mother's as well as the infant's health history (which may include several previous clinical care providers who tacted). When the results are finalized, they are typically compiled into a static portable document report. Representing clinical data within a PDF locks the data out of the health system and renders it adable. Storing the data as computable data points however, would increase the likelihood of the data nhance the care of the patient in future encounters?

Background

Fast Healthcare Interoperability Resources (FHIR) is a rapidly-growing Health Level Seven International (HL7) standard that represents specific data artifacts found in typical clinical scenarios (Patient, Condition, Encounter, Observation, etc.) as standardized "resources". These resources are linked together via identifiers to provide a standardized and robust representation of clinical events. The resources themselves can be thought of as both the

#### Scoping the work for max impact:

- In Utah in 2022
  - Number screened: 46,754
  - Confirmed diagnosis: 676
  - 544 Hemoglobinopathies
    - 3 disease
    - 541 traits = 80%
- Test fee is \$200 = minimal budget

Psychosocial distress & Uncertainty







## **GARDE-chatbot platform**

					Study/projec	et	Domain	Implementation	Study type	Status	Setting and
Study/project description	Domain	Implementation approach	Study type	Status	Setting and sample size	ditary adapted		approach Epic® EHR and MyChart patient portal.			Sample size University of South Carolina
	tion and test genetics integrated with population registry in population registry in Epic® EHR and Whychart patient portal. Chatbot interactions or create an open-by eversion of the SE trial about genetic testing University Langone Health (N=3,073 patients)	integrated with population registry in		Ui N	Utah Health and New York	DGE trial		Chatbot interactions and patient decisions about genetic testing stored in the EHR.			(MUSC) (N=92 patients)*
cancer (GARDE-Chat used to create an open- source version of the BRIDGE trial chatbot)[24]		tic testing ancer[26]	Cancer genetics	Chatbot hyperlink sent to patients via e-mail upon consent. Patients share hyperlink with family members using their preferred method.	Pilot observational study	С	Weill Cornell Medicine Gynecologic Oncology Clinic (N=100 patients)				
Education and access to free, mailed, at-home COVID-19 test kits (SCALE-UP II trial)[25]	l access to ti-home diseases   Infectious diseases   Eligible patient cohorts obtained from multiple EHR systems via data reports. Chatbot interactions and patient decisions about test kits tracked in study dashboard.   Pragmatic clinical trial   Community Health Centers (N=33,556 patients)	Community Health Centers	lts e cell trait	Disease screening	Chatbot embedded within Qualtrics® survey.	Usability testing	С	Survey with parents of a child <12 months old (N=250)			
		,	ive sults with tations for ser	Cancer genetics	Transcript of simulated chatbot conversations recorded in PDF format and shared with experts for rating.	Expert rating of LLM component	С	Survey with genomics experts (N=8)			
Bundled intervention providing education and access to state tobacco guittine, and services	iding education and services taste tobacco inge, and services essing social needs -UT trial)  prevention obtained from multiple EHR systems via data reports. Chatbot interactions and patient decisions tracked in study dashboard. Chatbot delivered via SMS.  Chatbot delivered via SMS.	obtained from multiple EHR systems via data reports. Chatbot		Co He (N	Community Health Centers (N=1,560	ut cancer	Cancer research	Chatbot available at the cancer center's Website and through QR codes displayed at clinics.	N/A	E	University of Utah Huntsman Cancer Institute
(REI-UT trial)		persistent poverty census tracts)*	reminder of follow- screening ial)	Cancer screening	Participants recorded in REDCap, database by study coordinator. Chatbot hyperlink sent to participants via REDCap, e-mail or	Pragmatic clinical trial	D	University of Utah Huntsman Cancer Institute Mobile Mammogram Unit (N= 2,700)*			
Education and access to telehealth-based shared-decision making for lung cancer screening (LungSMART trial)	Cancer screening	Eligible patient cohorts obtained from multiple EHR systems via data reports. Chatbot interactions and patient decisions about test kits tracked in study	Pragmatic clinical trial	D	Utah Community Health Centers (N=18,994 patients)*	risk r patients ow-up	Cancer genetics  Cancer	Patient portal (MyChart) message sent to each patient by clinical staff after mammogram appointment. Participants recorded in	Pilot clinical study  Community	D	University of Utah Huntsman Cancer Institute University of
Education and access to online evidence-based weight management program (SMARTLife trial)	dashboard.  Disease prevention prevention obtained from multiple EHR systems via data reports. Chatbot interactions and patient decisions tracked in study dashboard. Chatbot delivered via	Pragmatic D clinical trial	D	Utah Community Health Centers (N=5,354	sting offer n rural iplete screening	genetics	REDCap, database by study coordinator. Chatbot hyperlink sent to participants via REDCap, e-mail or SMS.	engagement studios		Utah Huntsman Cancer Institute Mobile Mammogram Unit (N=48)	
		decisions tracked in study dashboard.			patients)*	h (LIFT)	Disease prevention	Chatbot embedded within Qualtrics® survey.	Expert rating of LLM component	D	University of Utah Huntsman Cancer Institute

Primary care

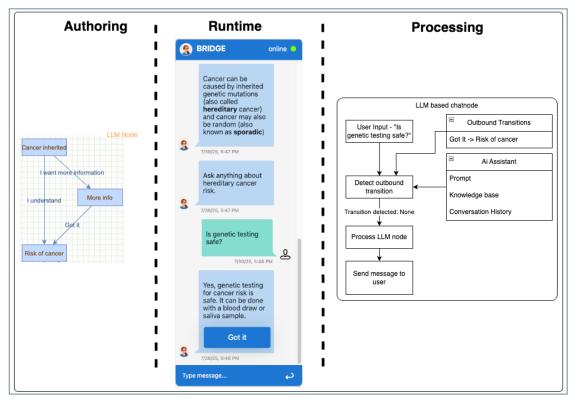
patients at

Pre- and post-test

education and test

GARDE algorithms

observational

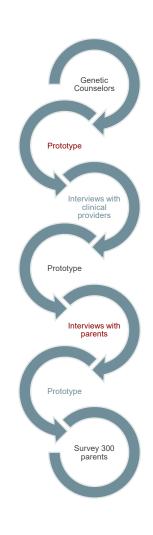


- Scripted
- Al
- Hybrid option

GARDE is an open-source software platform that supports the design and implementation of various interventions designed to promote patient engagement, education, and access to services, including PHM interventions



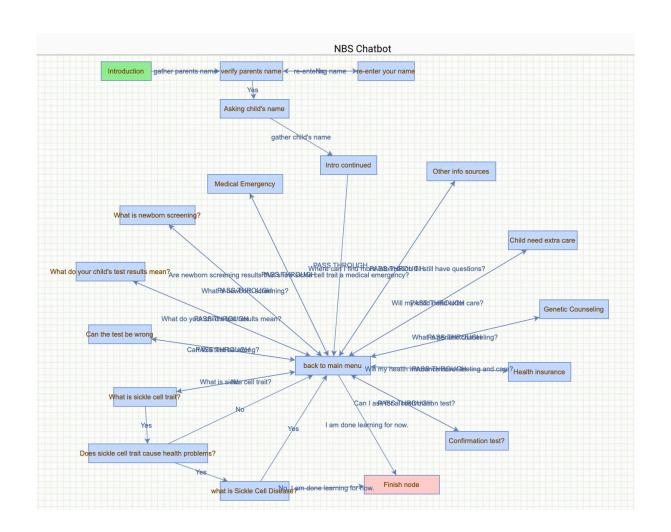
## Iterative Development of Scripted Chatbot

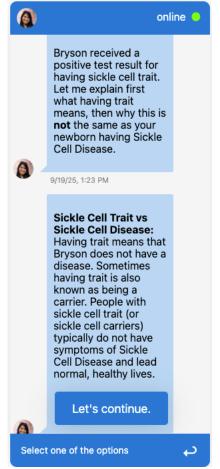


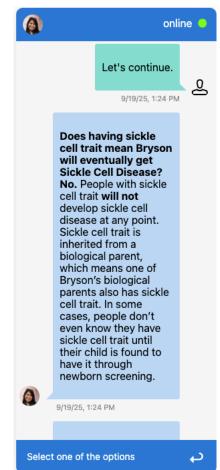
- 1. Developed a prototype with Amy Gaviglio as our content expert.
- 2. Use the prototype to frame the conversations in interviews, using the critical incident technique and think aloud with parents and providers
- 3. Modified the chatbot accordingly
- 4. Embedded in a Qualtrics survey and used validated instruments to measure uncertainty, negative and positive emotions and self efficacy



### Qualitative: Interviews with parents and GCs









**Qualitative: Parents' Reactions** 

"It seemed very compassionate."

• "The additional information is helpful because parents will have unanswered questions. As a worried parent, we would rather have immediate answers rather than waiting for a follow-up with a medical professional."

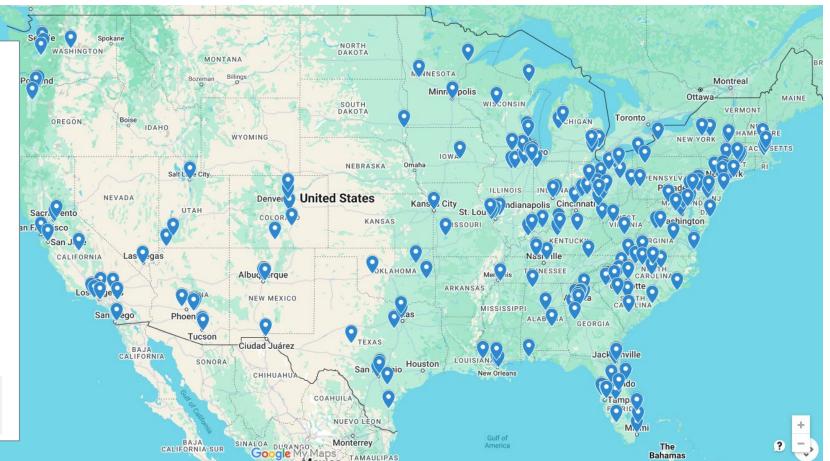
• "It was useful to have different options to choose from on what you wanted to know. I liked that it gave detailed information on the topic and also other topics that may be of interest."



## Quantitative: 300 parents across the US

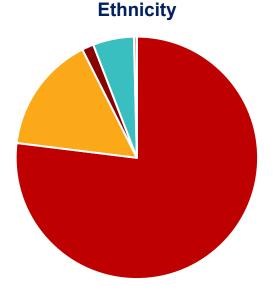
**INSTRUCTIONS:** The following questions ask about how you **would feel** after reading this letter about your child's newborn screening results. Please indicate how much you would expect to have each specific feeling after reading the letter by choosing one answer for each question.

	None at all	A little	Somewhat	A good deal	A great deal
How <b>upset</b> would you feel about the newborn screening result?	0	0	0	0	0
How <b>happy</b> would you feel about the newborn screening result?	0	0	0	0	0
How <b>anxious or nervous</b> would you feel about the newborn screening result?	0	0	0	0	0
How <b>relieved</b> would you feel about the newborn screening result?	0	0	0	0	0



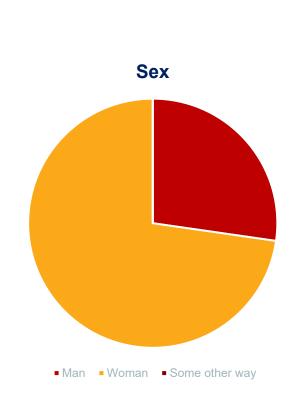


## Demographics of participants

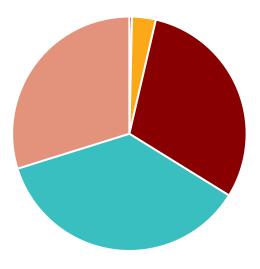




- American Indian or Alaska Native
   Asian or Asian American
- Native Hawaiian or Pacific Islander
   Some other ancestry or origin



#### **Education Level**



- Elementary School
- Junior high or some high school
- High school diploma or Graduate Equivalency Diploma (GED)
- Some college or Associate Degree
- College degree or higher



## Significant differences in positive & negative emotions, and uncertainty

FACToR validated Survey	Before chatbot	After chatbot	T statistic	p-value	Cohen's D
Negative subscale (0-12)			6.1	p<.0001	0.3881
Means (SD)	7 (3.2)	5.8 (3.2)			
Range	0-12	0-12			
Positive subscale (0-16)			3.6	p<.0001	0.2301
Mean (SD)	8.6 (4.2)	7.8 (4)			
Range	0-12	0-12			
Uncertain subscale (0-12)			4.9	p<.0001	0.3091
Mean (SD)	6.5 (3)	5.5 (3.4)			
Range	0-12	0-16			

- •Negative emotion = significant decrease
- Uncertainty = significant decrease
- •Positive emotion = significant increase
- •Was the information helpful
- = significant increase
- •Usability Score 73.5 (above 68 is good, above 80 excellent)
- •Self-efficacy did not change.



### Themes derived from the parent narrative about using NBSchat.

ning that just because someone has a trait doesn't mean they have the disease.
ught that it was useful how the bot gave an in-depth explanation of the results and what mean and explaining the difference between sickle cell trait and disease
reassurance that the trait doesn't mean Sickle Cell Anemia.
fast clear responses. The response wasn't in medical terms, so I could understand it y. I also liked that the chat answered my questions and honestly would have made me better if this [w]as a real situation
kly find out that everything is okay without waiting until the morning, when the doctors' ing day begins.
worried parent, we would rather have immediate answers rather than waiting for a followith a medical professional.
the pre-developed questions to ask further information.
nd it useful that it gave me pre-question to ask further, because it could be hard at this ent to even know what to Google and seek out more information.



# Combing quantitative and qualitative results

- Consistent message across both investigations
- Parents like having a menu to guide learning
- They like the idea of having immediate answers to health concerns
- Filling the knowledge gap reduced negative feelings and was described as reassuring
- The usability was good.

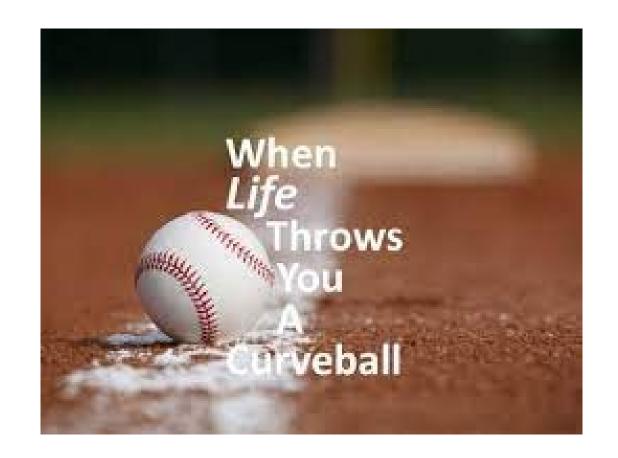


### Time to Pivot!

In May 2025, Utah passed House Bill 363—Maternal and Infant Amendments.

#### This means the law now requires:

- The Department of Health must publish a privacy consent form for parents.
- This form must include:
  - Why biological samples (like residual blood dried blood spots) and genetic data are retained.
  - How long the state keeps those samples/data (retention policy).
  - A clear option for parents to consent or decline retention





## **Why This Matters**

Previously, many parents were not aware that their child's blood spots could be stored and used beyond the initial newborn screening test.

This change **formalizes transparency**: parents must now be told the purpose and retention policy, and must actively consent.

The law creates a **shift in responsibility**:

Parents need understandable information.

Healthcare providers are expected to explain it at a sensitive time (right after birth), but often lack detailed knowledge about storage, secondary use, and research impacts.





## **Gap in Practice**

Most clinicians know how newborn screening works for detecting disorders BUT

Few are trained to explain what happens to leftover blood spots:

- Storage duration.
- Future research use.
- Privacy protections.
- Impact on children's health, public health, and medical discoveries.

Without clear explanations, parents cannot give true informed consent

### **Our Solution:**

Pivot and Use NBSChat (SCT Chatbot) for Newborn Screening Education

Currently writing R01 proposal to the NIH to utilize NBSChat to educate parents during the 3<sup>rd</sup> trimester.

Created to **bridge the knowledge gap** between policy and practice.

Parents can access clear, plain-language answers about:

Residual dried blood spot storage.

Why retention matters for quality assurance and future research

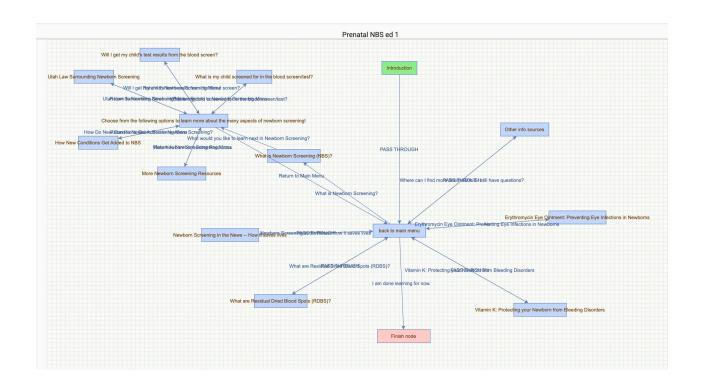
Their rights to consent or decline.

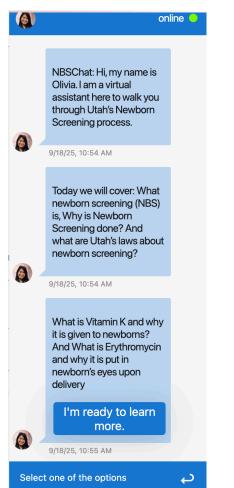
Ensures consent is informed, not just signed.

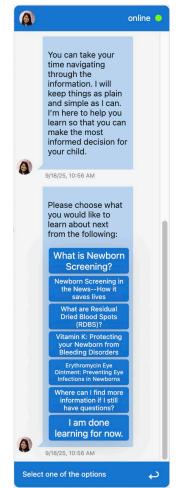
Supports healthcare providers by giving families a reliable, 24/7 educational tool.



## **NBSChat Expanded**









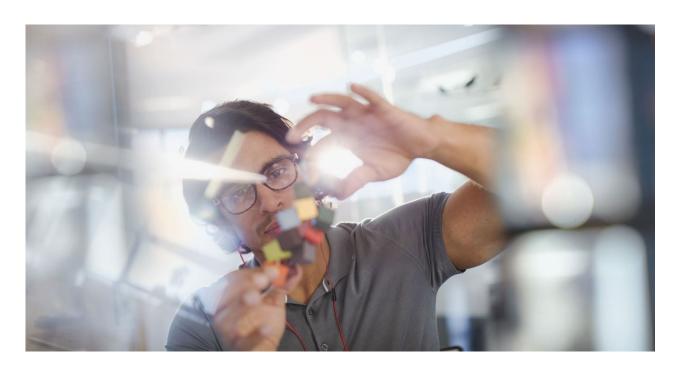
## Where do we go from here?

We have NOT forgotten the important work we began with our **Sickle Cell Trait Chatbot.** 

 Our team has met with other states and continue working towards implementation

Our new Newborn Screening and Residual Blood Spot Education Chatbot builds the same foundation:

 Using informatics and technology to empower families with knowledge, support providers, and strengthen the future of newborn screening.







# This is JUST the BEGINNING!

Whether it's sickle cell trait, residual blood spots, or future conditions, we are committed to ensuring parents have the tools they need to make informed choices and that public health continues to advance through innovation.

We want to collaborate on NBS issues



## **Q&A Session**

## **Share your feedback**

Please let us know what other topics you would like us to consider for future webinars!



## Thank you!!!



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