



NATIONAL  
RESEARCH  
BLUEPRINT

# A world without bleeding disorders begins with research:

Advancing Toward a  
Community-Driven  
National Research  
Blueprint for Inherited  
Bleeding Disorders



NATIONAL HEMOPHILIA FOUNDATION  
*for all bleeding disorders*

Summer 2023

# A world without bleeding disorders begins with research.

The National Research Blueprint (NRB) is a multi-year, comprehensive research effort underway across the bleeding disorders community, and championed by NHF. Together, participants in the NRB effort are rethinking the approach to research in bleeding disorders by creating a comprehensive, collaborative, and community-driven national research strategy.

During Phase 1 of the process, NHF convened the community, including health professionals, community leaders and most importantly, those affected by bleeding disorders – referred to as lived experience experts (or LEEs; more on this later!) to identify gaps and propose research priorities.

We are now steeped in Phase 2 of the NRB process, which involves defining a blueprint to address these questions. Importantly, this blueprint is not only addressing resource and infrastructure needs to accelerate research, but is centered around the concept of LEEs as integral partners of the entire process, as well as the principles of equity, diversity, representation, and inclusion. In this progress update, we are pleased to share with you what we have achieved so far and what lies ahead for the NRB. Take a look.

## WHAT'S INSIDE:

- How far we've come together
- The central role of Lived Experience Experts (LEEs)
- SOS manuscripts published!
- Designing the National Research Blueprint
- What lies ahead
- How you can get involved

## NRB: Fueling an Ambitious Vision

The National Research Blueprint for inherited bleeding disorders will define:



**A patient-centric multidisciplinary research enterprise**



**National research infrastructure expansion**



**Reinvigorated and sustainable workforce**

- Activated and driven by an educated, informed, and engaged BD community of lived experience experts (LEEs) and health care providers (HCPs)
- Grounded in sound principles of equitable and inclusive access to care and research
- Fueled by strong research advocacy, communications, funding and policy progress



# How Far We've Come Together

A community-led effort from the beginning

We started by listening to and learning from the community.

This collective input has informed our journey together.

## Three years of progress driven by community dialogue



*“As someone living with a rare bleeding disorder, it’s exciting to be involved in discussions about research that could benefit my life.”*

– Esmeralda Vazquez, Steering Committee patient representative



# The Central Role of Lived Experience Experts (LEEs):

## The voices of those affected by bleeding disorders are integral to the research process

The NRB effort aims to bring the experiences of people affected by inherited BDs to the forefront of transformational research, understand the most pressing issues challenging them, examine where research can be most impactful and define a nimble, collaborative, patient-centric, equitable research infrastructure to address them. As such, one core aim of the NRB is to be fully aligned with the values and needs of lived experience experts (LEEs) – those who live with or love someone with a bleeding disorder. Since the NRB process was initiated, community members have been integral to conversations, surveys and working groups that have informed this work.

Initially, to recognize the unique and important perspective that they possess, these participants were referred to as “subject matter experts” or SMEs. With input from these

representatives, NHF now refers to personally affected individuals as “lived experience experts” or LEEs. This term better reflects the distinctive expertise LEEs acquire by living with these disorders and the imperative that their perspective remains at the center of every step of the research process.

Especially in rare disorders, LEEs have unique, valuable expertise to contribute to all stages of research (e.g. planning and designing, participating and recruiting participants, communicating its importance and results). LEE participants in the NRB outline the importance of integrating LEE voices within the research process and the opportunities for more impactful results to benefit patient care in an editorial published in **Expert Review of Hematology in March 2023**.

### “Patients” as Partners



If you are interested in joining this group and contributing to the advancement of science in bleeding disorders, please contact NHF at [research@hemophilia.org](mailto:research@hemophilia.org).



Esmeralda Vazquez



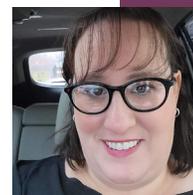
Kyle Davis



Michael Glenzer



Ray Stanhope



Sammie Valadez



# The State of the Science Research Summit (SOS)

The first phase of the NRB, focused on community listening, focus groups, surveys and working groups, culminated in the virtual State of the Science (SOS) Research Summit in 2021. During this Summit, each of the six SOS working groups presented and ranked priority research questions in key areas of unmet need, welcoming comments and additional questions from community representatives to uncover breakthrough challenges and opportunities. This process has formed the basis for the NRB now in development.

**Key Outcome:** The SOS discussions helped us identify key themes and areas of focus:

GLOBAL THEMES	RESEARCH THEMES	RESEARCH PRIORITIES
 <b>People Centricity</b>  <b>Collaboration</b>  <b>Health Equity, Diversity &amp; Inclusion</b>	<ul style="list-style-type: none"> <li>• Understanding the pathobiology of bleeding</li> <li>• Novel and improved diagnostics</li> <li>• Novel and adapted therapeutics</li> <li>• Impediments to care</li> </ul>	<ul style="list-style-type: none"> <li>• Bone and joint health</li> <li>• Influence of sex and gender on disease</li> <li>• Aging</li> <li>• Immunogenicity</li> <li>• Mental health</li> <li>• Pain</li> <li>• Research infrastructure</li> </ul>



We are very grateful to the hundreds of people who presented, participated, or listened to SOS Research Summit. If you missed it, the session recordings are still available!

[Click here for more.](#)



## Thank you to the SOS working group leaders and partners

### SOS STEERING COMMITTEE

#### Co-Chairs

- Len Valentino
- Michael Recht
- Kevin Mills

#### Committee

- Shannon Carpenter
- Esmeralda Vazquez
- Jill Johnsen
- Barbara Konkle
- Sarah O'Brien
- Steven Pipe
- Donna DiMichele
- Michelle Witkop
- Maria Santaella

### SOS ADVISORY COMMITTEE

- Craig Hooper
- Keith Hoots
- Kathryn McLaughlin
- Peter Marks

### SOS WORKING GROUP CHAIRS

#### Research Priorities for Hemophilia A & B

- Bobby Tran
- Annette Von Drygalski
- Kevin Mills

#### Research Priorities for von Willebrand Disease, Platelet Dysfunction & other mucocutaneous IBDs

- Robert Sidonio
- Veronica Flood

#### Research Priorities for Ultra-Rare IBDs

- Amy Shapiro
- Diane Nugent
- Suchitra Acharya

#### Research Priorities for health of women & girls and persons with the potential for menstruation

- Angela Weyand
- Maureen Baldwin

#### Diversity, Equity & Inclusion Health Services Research & Implementation Science

- Judith Baker
- Tyler Buckner
- Vanessa Byams

#### Facilitating Priority Research in the IBDs Community

- Margaret V. Ragni  
*Resources and Funding*
- Jordan Shavit  
*Workforce*
- Guy Young  
*Infrastructure*



# SOS Manuscripts Published

The first phase of this process resulted in important observations and recommendations that are now informing the development of the NRB, and we have been eager to share all of this insight broadly with our community partners. The initial recommendations of each working group from the SOS were published in March 2023 in a **Supplement** of the journal *Expert Review of Hematology*.



## Publications represent the foundations of a clear and actionable NRB

- **Methodical processes** taken to ensure the recommendations are well grounded in clear unmet needs and under-addressed challenges
- Opportunities thoroughly **assessed for feasibility, risk and impact**
- Conclusions of each working group offer **defined, specific steps to make progress** in each focus area

## Most importantly, they reflect the interests and desires of our community

- Combined conclusions reflect months and years of **community dialogue and debate**
- Grounded in social **justice principles, diversity, equity and inclusion**
- Deep **integration of lived experience experts** (LEEs) through the process, supporting more realistic applicability of research priorities that will address care for excluded populations

These manuscripts define specific themes and research recommendations compiled by each working group that have the greatest potential to make a meaningful, lasting impact for people with inherited bleeding disorders. Importantly, these papers have taken the important ideas and unmet needs raised throughout our conversations and translated them into tangible, feasible, and prescriptive guidance on how to improve research for the future. For example:



### Simplify diagnosis for ultra-rare disorders

Build and connect databases to aggregate natural history, treatment, and outcome data for ultra-rare inherited BDs



### Speed research through infrastructure

Evaluate three coordinating network models to integrate data collection and accelerate insight generation



### Reset the dialogue on acceptable bleeding

Establish and broadly disseminate affordable methods to quantitate bleeding in people who menstruate

The guidance provided in these papers has formed the basis of the National Research Blueprint and the NRB Steering Committee extends sincere thanks to everyone who contributed to these important papers.



## Click on each working group title to read the papers

**Working Group 1:** Building the foundation for a community-generated national research blueprint for inherited bleeding disorders: Research priorities to transform the care of people with hemophilia

- Building on the great progress in treating hemophilia, the focus now shifts toward advancing health equity and improving diagnostics and testing.

“Addressing the many disparities encountered by the community is essential to achieving health equity for all PWH.” – Hemophilia Working Group 1 Authors

**Working Group 2:** Building the foundation for a community-generated national research blueprint for inherited bleeding disorders: Research priorities for mucocutaneous bleeding disorders

- Focused research could improve diagnosis and management of these poorly understood disorders to reduce the substantial impact on quality of life.

In the US, the average delay between symptom onset and diagnosis of von Willebrand disease for females was reported to be 16 years.

**Working Group 3:** Building the foundation for a community-generated national research blueprint for inherited bleeding disorders: Research priorities for ultra-rare inherited bleeding disorders

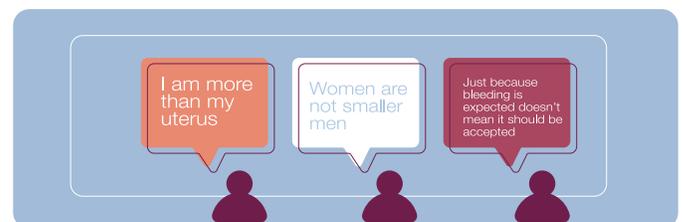
- New research opportunities can improve diagnosis, treatment and testing of new therapies for ultra-rare IBDs leveraging collaborative research networks and infrastructure.

### Defined opportunities:

- National network of specialists
- Centralized laboratory
- Disorder-specific treatment pathways
- New study design for small populations

**Working Group 4:** Building the foundation for a community-generated national research blueprint for inherited bleeding disorders: Research to advance the health of women and girls and people with inherited bleeding disorders with the potential to menstruate

- Poor understanding of the line between normal and abnormal bleeding lead to disparities in proper diagnosis and care. Research can build foundational knowledge, standards for diagnosis and assessment, and patient-centered outcomes goals.



**Working Group 5:** Building the foundation for a community-generated national research blueprint for inherited bleeding disorders: Research priorities in health services; diversity, equity, and inclusion; and implementation science

- Considerable gaps must be addressed to improve healthcare delivery and equitable access to ensure all people with IBDs can receive the care they need.

**Working Group 6:** Building the foundation for a community-generated national research blueprint for inherited bleeding disorders: Facilitating research through infrastructure, workforce, resources and funding

- Accelerating IBD research requires focused investments in infrastructure, workforce development and funding to foster research collaborations and networks.

**Lived Experience Expert Editorial:** Lived Experience Experts: A name created by us for us

- This commentary reflects on the impact of the patient perspective in defining research that will make a meaningful difference.

**International Perspective:** Soliciting international perspectives on an American national research agenda for inherited bleeding disorders

- NHF seeks perspectives on how research priorities for IBDs identified by the U.S. community may apply to other countries to further inform the global research dialogue.

## Research questions to foster health equity:

- Understanding differences in care delivery
- Uncovering barriers to clinical trial involvement
- Knowledge needed to better reach at-risk populations

“Research is critical to advancing the diagnosis and care of people with inherited bleeding disorders. But this requires significant infrastructure, including people and resources.”

– Facilitating Research Working Group 6 Authors

“Particularly in rare disorders, lived experience experts, or LEEs, have invaluable expertise to contribute at all stages of research.” – Esmeralda Vasquez

Improving IBD care worldwide: How do we ensure these priorities are realistic and feasible to help people with IBDs in other countries?

## Soliciting Perspectives to Inform the NRB

We believe that fully integrating the voices of those affected by these disorders is paramount to developing research priorities that will deliver the greatest possible impact for our community and transform care for future generations. Importantly, societal norms, access to care, and research infrastructure varies around the world, so open dialogue is critical to accelerating global progress.

To better understand international perspectives on BD research priorities and the role of LEEs in the research process, NHF invites you to contribute to two surveys. The results of these surveys will inform the NRB and will be integrated into future NRB-related publications.



International perspectives on BD research priorities



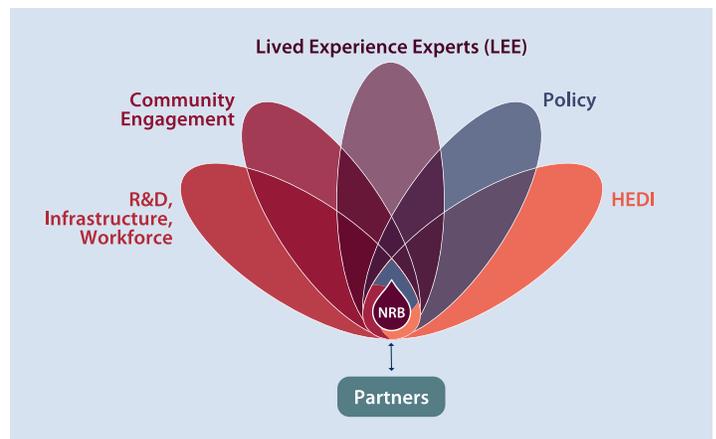
LEE contributions to the BD research process

# Phase 2: Designing the National Research Blueprint

Since the SOS Summit, the NRB leaders have been hard at work building an NRB that encapsulates the priorities dictated by the community and provides a clear roadmap on how to activate these priorities while fostering a productive, inclusive research culture in our community. The NRB is purposefully designed to address three critical needs:

- 1. Multidisciplinary research enterprise** that is centered in a network of both specialty and community-based care; driven by patients/families/caregivers as the lived experience experts (LEEs); embedded in the principles of social justice and sustainability; and that steadily advances the standard of care for people with IBDs through impactful basic, translational, clinical, health outcomes, and implementation research as well as advocacy and education
- 2. National research infrastructure** expansion that supports the envisioned research enterprise through facilitation collaboration and inclusion
- 3. Reinvigorated and sustainable workforce** across comprehensive care and scientific disciplines that incorporates the LEE perspective to advance health through the seamless integration of care and research as well as active community engagement to foster a research culture

The SOS focused on defining the most critical research questions. The NRB is now defining how these questions will be addressed in a collaborative, efficient and effective manner in a process that involves LEEs in all stages of the research enterprise and that leans heavily into social justice principles.



## Meet the NRB Steering Committee



Len Valentino  
Co-Chair



Michael Recht  
Co-Chair



Keri Norris



Michelle Witkop



Donna DiMichele



Esmeralda Vazquez



Maria E. Santaella

## NRB Working Group Chairs

<b>Research &amp; Development; Infrastructure; Workforce</b>	<b>Research &amp; Development:</b> Jill Johnsen, MD	<b>Infrastructure:</b> Moses Miles & Margaret Ragni, MD, MPH	<b>Workforce:</b> Lynn Malec, MD & Amy Shapiro, MD
<b>Community Engagement</b>	Nathan Mermillod	Shannon Carpenter, MD	Jeremy Griffin
<b>Lived Experience</b>	Erin Cirelli, Esq.	Sammie Valadez	Esmeralda Vazquez, OTD, OTR/L, ATP
<b>Policy</b>	Michelle Conte	Rich Pezzillo	Nathan Schaefer, MSW
<b>Health, Equity, Diversity and Inclusion</b>	Keri Noris, PhD, JM, MPH, MCHES Samantha Carlson, LMSW		



# Building the Blueprint

In 2022, we hosted two immersive workshops with representatives from across the inherited BD community to discuss how to translate broad research themes into a clear, defined roadmap toward research progress. During these workshops, participants assessed research questions identified by the community and prioritized them based on defined set of feasibility criteria to help crystallize our opportunities to make the greatest impact on patient care.

- **Feasibility**, which assesses the difficulty in answering the proposed question, including required expertise, infrastructure, and resources.
- **Impact**, estimating the change we can foster through the priority. Will the answer impact standards of care or access to care? Will it change the therapeutic paradigm, and could it be applicable to other areas?
- **Risk**, which considers the challenges of the research question, such as the risk/benefit ratio for novel strategies and any ethical considerations.



## What will this mean for our community?

Ultimately, this process will allow us to...

- **Rethink** how we prioritize and fund research that matters for those living with inherited BDs
- **Reorient and democratize the research culture** and structure to streamline research efforts, specifically addressing the urgent interests of LEEs and considering social justice principles
- Better apply our collective learnings to **make progress faster** for those who need it most.

**Faster progress** in truly unmet needs

**Novel insights** from integrated data

Clarity from **LEE voices**

Mechanisms to **build quickly** on learnings

**Acceleration** from discovery to delivery

Efficient **resource** use to do more faster

And **more** that we will learn as we go...

# Steady progress toward our blueprint

While we've made great progress, there is still much to do, and we are taking a methodical approach to build a robust, actionable blueprint that takes into account the most critical aspects that the community has identified to date.

Our focus over the last year has been the development and integration of recommendations from each working group toward a robust research enterprise.

- **Individual WG recommendations:** Collecting all of the input so far from community listening, the SOS summit, manuscripts and workshops, our NRB working groups have defined and articulated a core set of recommendations related to their respective focus areas.
- **Working Group integration:** This year the working groups have begun to integrate their recommendations into one consolidated effort. The blueprint framework led by the Infrastructure, Workforce, R&D working groups will outline steps to a functional research infrastructure and initial priorities. This will be enriched and developed through interactions with the HEDI, LEE, Community Engagement, and Policy working groups. Through regular interactions and dialogue facilitated by NHF, one integrated set of final recommendations is expected to be completed later this year.

## What lies ahead

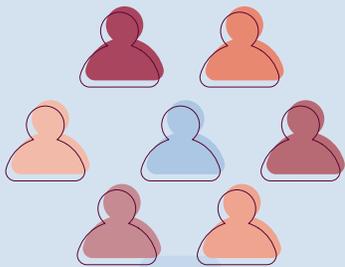
The NRB process to date has been an incredible example of collaboration and persistence to understand the most pressing issues for our community and identify solutions that can make a real difference. Never before have we had so much engagement from every corner of our community, with clear understanding and agreement on the incredible opportunity ahead of us to transform care. This is a critical moment for generational change. What can you expect in the coming months?

- **BDC Update:** Join us at BDC in August for an update on our progress and dialogue on the key themes the NRB will cover; most notably, the role of LEEs in the research process, principles for social justice, and how to build the infrastructure and funding to activate a fully integrated research culture.
- **NRB Summit, early 2024:** We are planning for a Summit in early 2024 to present the NRB approach and collect input. The planned hybrid format will allow us to engage a broad, diverse representation of our community through moderated virtual panels and Q&As coordinated by NHF chapters. Check back for more on that event soon.
- **Looking ahead to 2024:** Once the Blueprint is defined, our work is not yet complete. In this next chapter, we'll define the necessary steps to guide implementation and adoption of this research enterprise.



# How you can get involved

We can only make a real, lasting impact if we all do this together. This NRB is truly a collaborative effort involving the entire community. Learn more about what you can do to join:



- Consider participating as a Lived Experience Expert (LEE)! Contact **NHF** for more information.
- Follow our progress online by visiting the **National Research Blueprint** section of our website
- Join **Community Voices in Research (CVR)** via QR code
- Sign up to get communications from us! **Subscribe for Email Updates** (select all that apply but especially research)
- Join us for a special **Bleeding Disorders Conference (BDC)** session – stay tuned for more details!



We look forward to sharing more updates soon; thank you for joining us on this journey! We encourage you to contact [research@hemophilia.org](mailto:research@hemophilia.org) with any questions or comments.