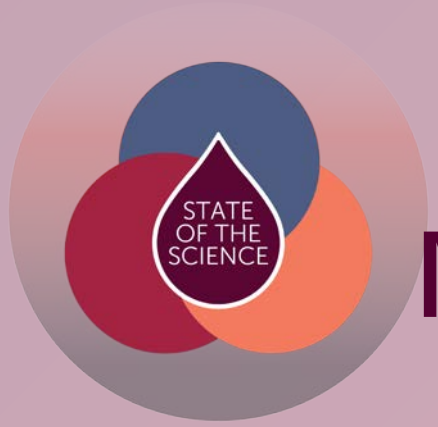
NATIONAL HEMOPHILIA FOUNDATION
for all bleeding disorders



Building the Blueprint March 2022 - Workshop

Working Group 3
Ultra-Rare Disorders

WG3 Organization

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- Research priorities for Ultra-Rare IBDs divided into three categories

Focus Area	Chair
Diagnostics, Systems Biology, Mechanistic Science	Diane Nugent, MD
Clinical, Data Collection, Research Infrastructure	Suchitra Acharya, MD
Regulatory Processes For Novel Therapeutics & Required Data Collection	Amy Shapiro, MD

WG 3: Subgroup participants

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Subgroup A:
Diagnostics, Systems Biology, Mechanistic Science

Diane Nugent, MD [Lead]

Catherine Hayward, MD, PhD

David Ginsburg MD, PhD

Alisa Wolberg, PhD

Roberta Palla, PhD

Rajiv Pruthi, MD

Kate Nammacher [NHF]

Subgroup B:
Clinical, Data Collection, Research Infrastructure

Suchitra Acharya, MD [Lead]

Michael Tarantino, MD

Michael Recht, MD, PhD, MBA [ATHN]

Camille Bedrosian, MD [UltraGenyx]

Roberta Palla, PhD

Marzia Menegatti, PhD

Maggie Miller, MS

Kim Baumann, PT, MPT

Kerry Hansen, RN, BS

Subgroup C:
Regulatory Processes for Novel Therapeutics & Required Data Collection

Amy Shapiro, MD [Lead]

Peter Marks, MD, PhD [FDA]

Deya Corzo, MD [Sigilon]

Michael Recht, MD, PhD, MBA [ATHN]

Kai Brown, MS, MBA [NHF Board]

Rebecca Bialas, MD [PLGD Fdn]

Benny Sorensen, MD, PhD [Codiak Biosciences]

Skye Peltier, PA-C [Community Member]

Amar Haidar [Community Member]

A. Diagnostics, Systems Biology, Mechanistic Science

- Does adequate diagnostic testing exist for each disorder?
- Do physicians have adequate access to diagnostics?
- Can diagnostic testing capabilities, availability be centralized?
- Does there exist a national mechanism to identify the genomics or associate & delineate phenotypes within each disorder?
- Can we identify disease modifiers, genes associated with rare disorders through national genomic banking?

B. Clinical, Data Collection, Research Infrastructure

- Clinical
  - Do providers have adequate knowledge of phenotype, access to required diagnostics?
  - Do defined severity categories exist to predict outcomes, guide treatment?
  - Do treatments exist to address clinical manifestations & prevent sequelae? Global vs. specific treatments?
  - Can affected individuals access best care?
  - What are QoL impacts & are they collected?
- Data Collection
  - What is centralized data collection capability to define natural history, treatments, outcomes?
  - Can it fulfill post-approval regulatory requirements?
- Research Infrastructure
  - What are impediments preventing enrollment, follow-up for research in ultra-rare disorders?
  - Does a national infrastructure exist for centralized testing, sample banking?
  - Does national infrastructure exist for data collection to support research?
  - Is collected data accessible for care & further research?

C. Regulatory Processes For Novel Therapeutics & Required Data Collection

- Are there adequate FDA pathways for very small populations to allow more rapid, less costly access to new therapies?
- What is the need for post-approval data collection if therapies approved on minimal patients/data sets?
- How can we incentivize product development for ultra-rare disorders?
- How can approval for off-label use of licensed therapies be obtained without associated prohibitive cost?