Building the Blueprint
March 2022 - Workshop

Working Group 3
Ultra-Rare Disorders
Research priorities for Ultra-Rare IBDs divided into three categories

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<th>Focus Area</th>
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<td>Diagnostics, Systems Biology, Mechanistic Science</td>
<td>Diane Nugent, MD</td>
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<td>Clinical, Data Collection, Research Infrastructure</td>
<td>Suchitra Acharya, MD</td>
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<td>Regulatory Processes For Novel Therapeutics &amp; Required Data Collection</td>
<td>Amy Shapiro, MD</td>
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## WG 3: Subgroup participants

### 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?