Models of Rare Disease Networks

NORD RD Centers of Excellence

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Preamble and Acknowledgements

- Marybeth McAfee, MA, GC the Associate Director, NORD RD CoE Program kindly shared information

- I do not officially represent NORD
- I was a previous director for a NORD CoE at Nationwide Children's Hospital in Columbus Ohio

- My additions are noted with asterisks or UCalgary slides

- Disclosures:
- Funding for clinical trials from Abeona, Biomarin, Homology, Ultragenyx
- I serve on DMC for Sanofi

Challenges of Rare Disorders

- Limited abilities to recognize rare disorders at entry to healthcare
- Lack of care pathway leads to:
  - Delays (diagnosis, treatment) and inequity
  - High burden of care coordination and financial strain on families
- Applying clinical recommendations requires organizational structure
- Centres of Excellence (CoE)
  - Some disease specific CoE (CF) have good model for CoE structure
  - Many RD are very rare, not enough expert people, lack of good evidence for CoE
- Network of expertise to optimize/distribute and ensure equity of care
  - Complex care not just specialists - Chronic care, allied health care, etc.
  - Infrastructure – hospitals, institutes

Treatment Guidance Uncertainties

- Most rare diseases do not have detailed natural history studies
- Rare disease therapeutics will continue to be challenging
  - Endpoints difficult and not always practice friendly
  - Evidence will likely always be very incomplete
- Systematic evidence-based reviews often not fruitful
  - Frequently not enough evidence for practice guideline
- Reliable source of clinical practice information still needed
- ***Patient and family experiences need to be incorporated***

Vision of the NORD RD CoE Program

All persons living with a rare disease, regardless of disease, socioeconomic level, or demographics, have access to timely diagnosis, quality, compassionate clinical care, research opportunities, and supportive resources.

- Policy
- Professional Education
- Research
- Care

Shorten Time to Diagnosis

Improve Quality & Access Care

Accelerate Research to Develop New Treatments

Increase the Number of Multi-Site Clinical Trials

Train More Rare Disease Specialists

NORD®

Applications Process – Initial Round 2021

Invitations

- Invitations were sent to the 40 plus Institutions
- Directed to head of clinical genetics programs
- Had ACGME certified clinical training programs

Technical Assessment Call

- Program information – more than a sticker
- Application information
- Explanation of rubrics
- Q&A

Analyzing the Applications

Multiple choice questions:

- Qualitative (what is available for services & specialists) and quantitative (no. of geneticists)
- Transplants, concierge/navigator services, esoteric labs

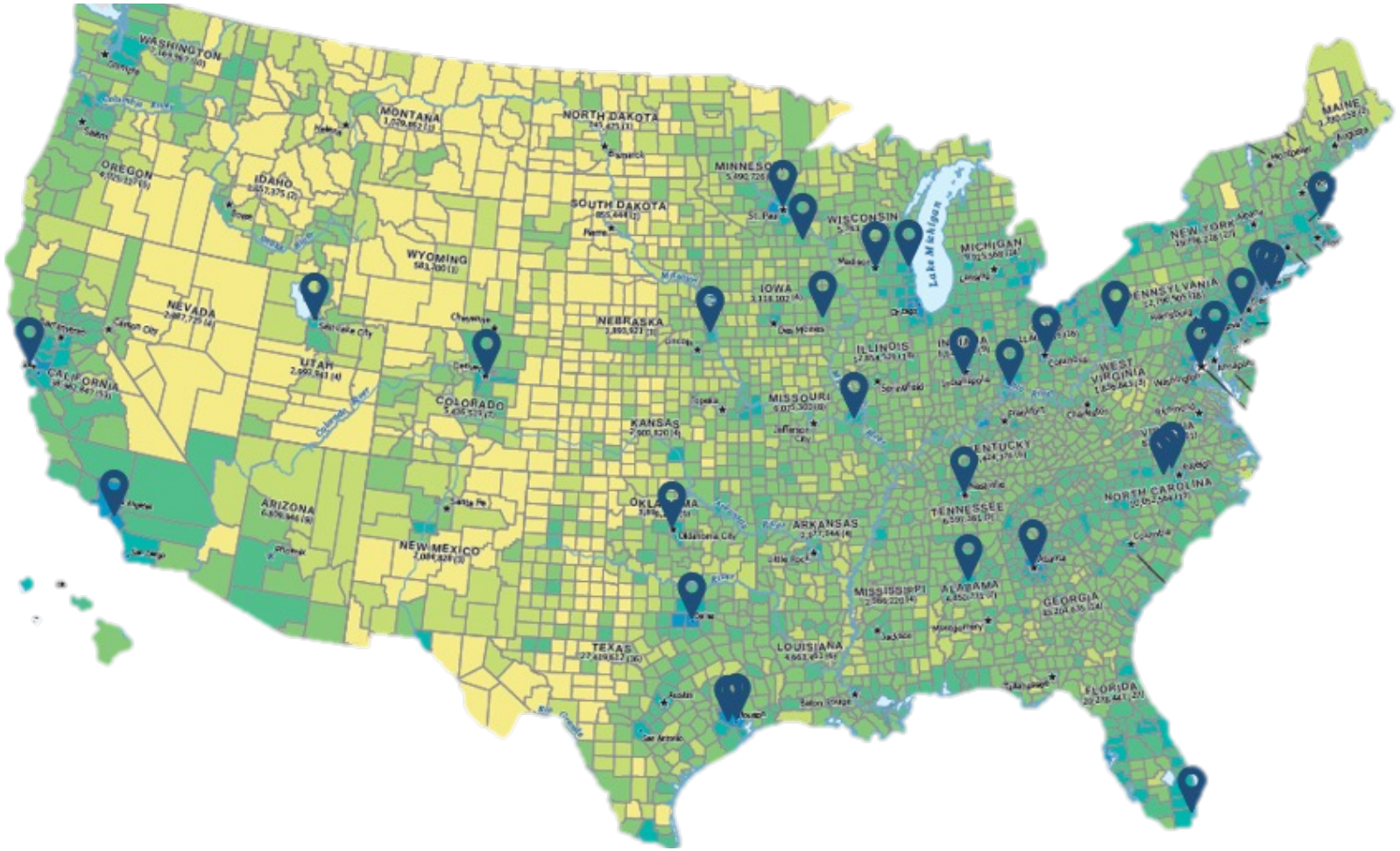
Essay questions:

- Clinics, training programs, patient resources, transition

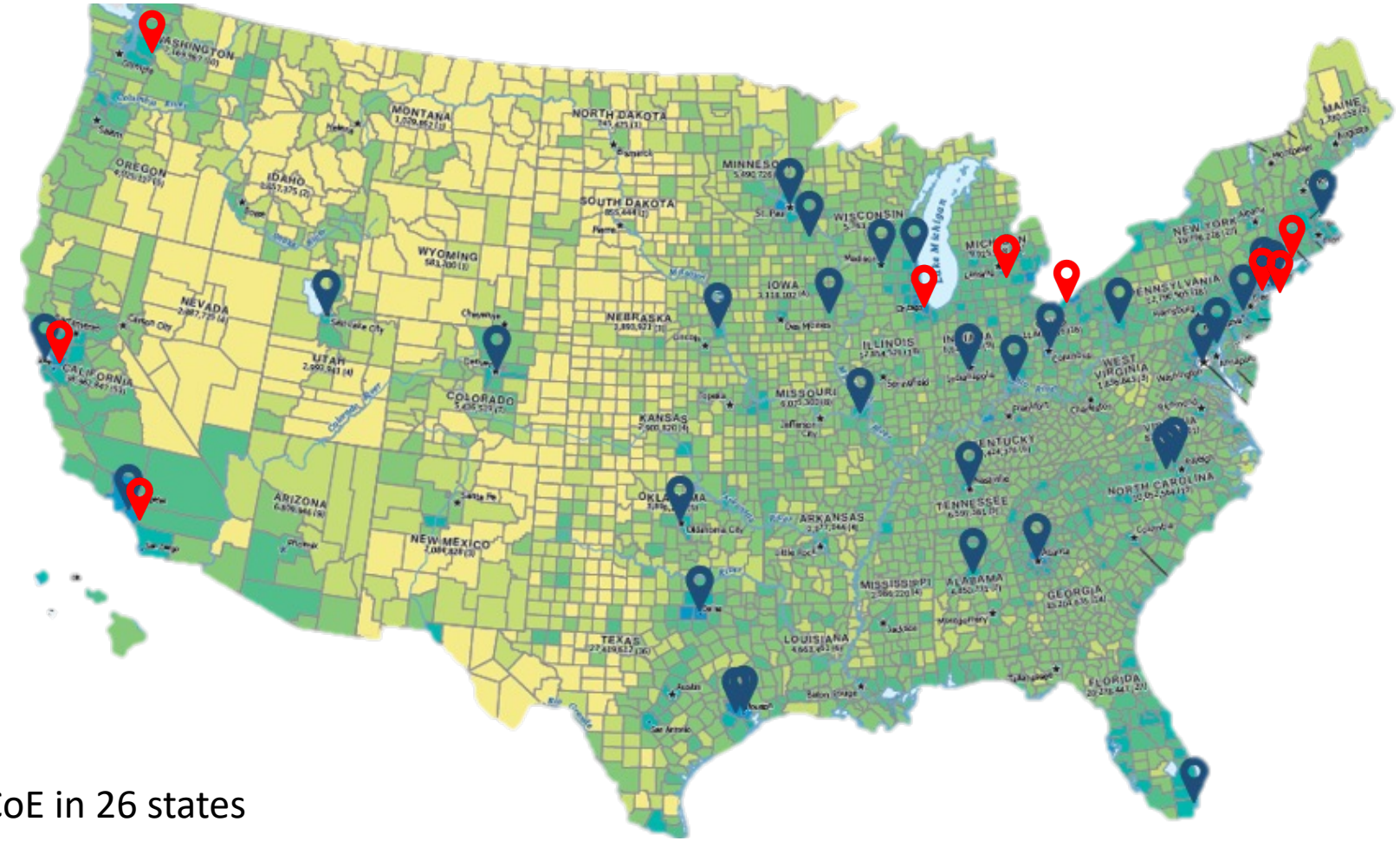
NORD Rare Disease Centers of Excellence

On Nov 4th 2021, NORD announced its designation of 31 medical institutions across the United States with exceptional programs for patients with rare diseases as NORD Rare Disease Centers of Excellence (NORD RD CoE's).

Geographic Distribution vs. Population Density



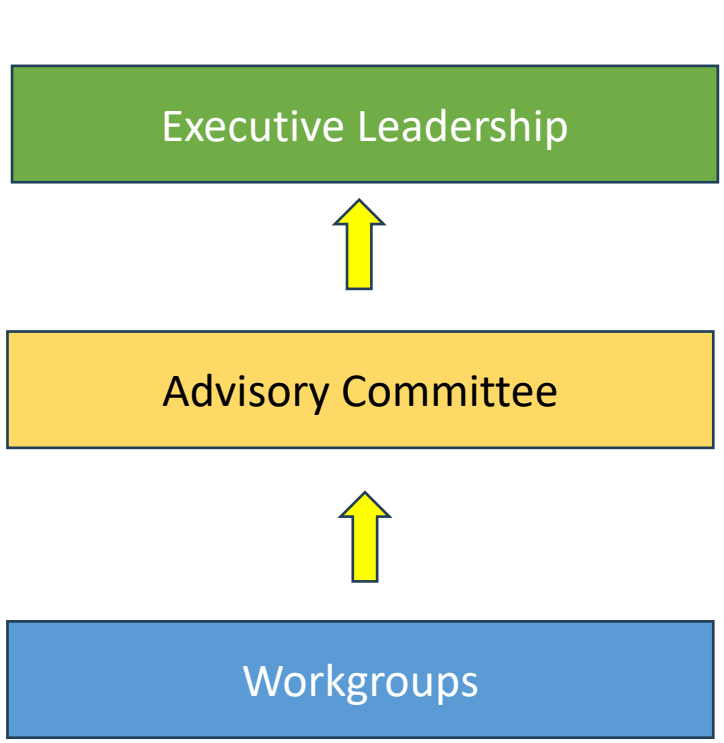
Second Round – Adressing Geographic and Center Gaps



NORD CoE in 26 states

KLM Modified

Structure of CoE



Chief Scientific & Medical Officer Ed Neilan

RD CoE Co-Director Marybeth McAfee

Center Directors

New Technology

Telemed

Diagnostics & Access

Data Collection & Sharing (Ultrarare)

Treatment Guidelines

DEI

Multisite Research

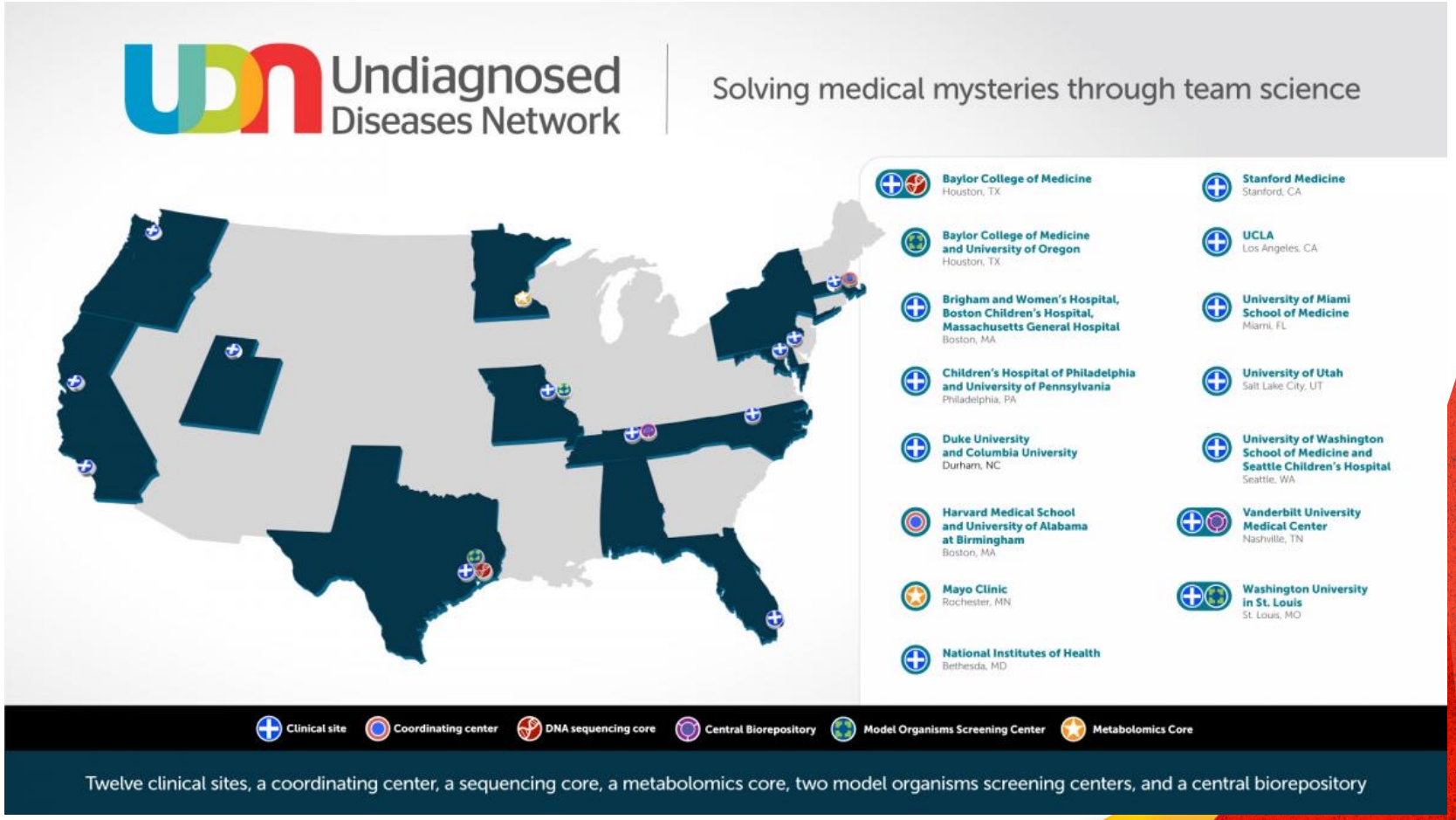
Patient Education

Training

Public Policy & Advocacy

Undiagnosed Diseases Network

- NIH Undiagnosed Disease Program started 2008
- Common Fund NIH program 2012; max program time of 10 years
- Rolled over as a competitive grant into Diagnostic Centers of Excellence



Rare Diseases Clinical Research Network

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- NIH initiative through National Center for Advancing Translational Sciences (NCATS) in 2003, latest round 2019
- Provides support for clinical studies (up to phase IIa), collaboration, enrollment, data sharing in consortia (multi site and patient centric)
- Disease or disease cluster
  - Urea cycle disorders
  - Primary immunodeficiencies
  - Incudes adult and non-genetic
  - ALS, perinatal infection

My View of US Models

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- NORD brought genetics centers together
  - Started framework for sharing and discussion
  - Treatment uniformity and expertise, patient advocacy
  - Unfunded (mostly)
- Center of Excellence is like stamp of approval
  - Patients and families look for places to go
- US healthcare is a business
  - Want to attract patients to your center as Patients = Dollars
  - Insurance coverage and patient's money crosses state borders
- Multiple networks
  - Can create competition to spur things along
  - Fragmentation by disease or focus (research vs diagnosis vs treatment)

Pros for Centres of Excellence

- Having a few places that have high expertise is shown to improve care
- Build expertise in rare disease in general – not every center needs to know all about everything – general concepts on approach to diagnosis, identifying treatment or expertise to tap
- Families often highly motivated to go to best place for treatment
- Places for Education and training

Cons of Centres of Excellence

- Remote and rural locations often left behind
- Solution: Travel vouchers/reimbursements, support for family in community to allow travel
- Solution: Bring some expertise to community
  - Project ECHO (Extension for Community Healthcare Outcomes) from U New Mexico established in Ontario
  - Collaborative hub and spoke model to provide expert care in community by video-conferencing technology to train, advise, and support primary care provider
- Integration into provincial/national systems

Concluding Thoughts on Canadian Rare Disorder Care

- Patient/family centered multidisciplinary and chronic care
- Centre of Excellence model may not be as optimal
  - Creation of network(s) in response to need for equity of care across country and account for provincial/federal framework of health care
- How do we fund this in Canada?
  - Not just treatment but diagnosis, specific specialists, comprehensive care teams, psychosocial supports

Thank You

- Questions or Comments?
- For discussion at end of session: What do you think would work to provide best care