Rare Diseases: Opportunities, Challenges, and Future at UNC
October 7th, 2024
Workshop Executive Summary

Rationale:
The "*Rare Diseases: Opportunities, Challenges, and Future at UNC*" workshop, hosted by the UNC Program for Precision Medicine in Health Care (PPMH) and the UNC National Organization for Rare Disorders (NORD) Center of Excellence, focused on brainstorming strategies to advance rare disease diagnosis and care at UNC Health and across the region. Gathering 60 clinicians, researchers, and patient advocates in the rare disease space at UNC, this inaugural workshop allowed participants to come together and share ideas on their experience and perspectives in relation to rare disease care and management. Using these insights, we will begin to move forward with strategic planning to improve the rare disease space at UNC Health and beyond.

Workshop Summary:
Opening Remarks: Stephanie Davis, MD

Keynote Speaker: Sam Young, PhD
Roper Investigator; UNC Gene Therapy Center Director
Dr. Young, recently appointed as the new UNC Gene Therapy Center Director, used the keynote speech to emphasize the importance of modern molecular medicine and targeted therapeutics, building on strengths of UNC's Gene Therapy program to transition into the Center for Molecular Medicine.

Session 1: Referral pathways for patients to appropriate providers or diagnostic clinics (eConsults & triage)
Leader: Melissa Haendel, PhD
Director of Precision Health and Translational Informatics
Dr. Haendel, who is new to UNC, presented on the diagnostic journey of patients with rare disease, primarily, how these pathways vary significantly between institutions and among various rare diseases. She highlighted the importance of coordinated expertise, which requires overcoming current barriers such as access and billing. Dr. Haendel emphasized that in order to create a functioning rare disease network, it is vital to take a systems approach.

Session 1 Group Activity: Envisioning an Informatics Network to Support Rare Disease Patient Care

Common Themes:

- EHR/Epic
    - Better integration of genomic tests
    - Improved clinical decision support (CDS)
    - E-consults available for PCPs
    - Rare disease patient registries
    - "Flagging" potential rare disease patients in the EHR
    - AI-facilitated Informatics of EHR data
- Additional genetic counselors on staff and more genetic counselor awareness
- Additional MD providers with rare disease expertise (Clinical Geneticists, Rare Disease Specialists, Radiologists, etc)
- Better data sharing within UNC (and with other institutions)

Session 2: Identifying patients with suspected rare disorders and facilitating testing (EPIC genomics module)
Leader: Michael Adams, MD
Genomics Module and Precision Medicine Fellow, PPMH
Dr. Adams is highly engaged with the development of decision support in Epic at UNC and took this opportunity to present his expertise with Epic decision support pathways. He outlined and gave specific examples of how UNC is utilizing Epic to test and identify patients with suspected rare disorders, as well as leveraging Epic decision support for downstream rare disease care management.

Session 2 Group Activity: Needs Assessment to Facilitate Testing at UNC

Barriers	Tools to Overcome Barriers

• Cost / Insurance
• Patient concern / reluctance
• Physician knowledge
• EHR / Epic
  ○ Unclear test selection
  ○ Inconsistent test interpretation
• Inaccessibility of genetic counselors
• Community relationships
• Referral pathways
• Differentiation in lab practices (i.e. VUS reinterpretation)
	• Streamlining of insurance processes
	• Patient education
	• Physician education (medical school & beyond)
	• EHR / Epic
	  ○ Decision support
	  ○ Assistance with test interpretation
	• Increased access to specialists / genetic counselors
	• Community outreach

Session 3: Managing patient care for rare diseases (therapies, social support, & patient networks)
Leader: Arti Pandya, MD, MBA
Chief, Division of Pediatric Genetics and Metabolism
Dr. Panya's presentation outlined the rare disease care landscape within and outside of UNC. In order to improve the rare disease space at UNC, it is essential to recognize what already exists and how we can build upon that foundation. Dr. Pandya emphasized that not only are provider perceptions important in rare disease management, but patient and family perspectives are vital to consider in determining the pathways to best care for these patients.

Session 3 Group Activity: Panel Discussion (Panelists: *Claudia Testa, MD, PhD; Kim Stephens, DBA; Carlee Friar, BS; Cindy Powell, MD; Logan Blinman, MS)*

Common Themes: Problems within rare disease patient management

- Shuffle of patients to various providers and clinics across country – huge burden on patient / family
  - Nurse / Care navigators need to be part of the landscape (e.g. utilizing Social Work and care coordination similar to other chronic disease clinics at UNC)
- Lack of provider knowledge – clinical geneticists and genetic counselors can play a role in supporting primary care and specialist providers
- Accessibility of Information – patient confusion and feelings of "aloneness"
  - Utilizing family advisory councils
  - Utilizing patient advocacy groups
  - Utilizing Epic for patient education
  - Keeping information current
- Socioeconomic issues – getting a diagnosis and treating a rare disease is very expensive in terms of both time and money
- Transition from childhood to adult care for rare disease patients – big area needing improvement

Session 4: Building rare disease research infrastructure (patient registries, biobanks, clinical trials, & machine learning)
Leader: Liz Jalazo, MD
Assistant Professor, Pediatrics; Director of Clinical Integration, Angelman Syndrome Foundation (ASF)
Dr. Jalazo outlined various rare disease communities and research infrastructures throughout the US, showcasing how these rare disease networks can be leveraged by UNC to both set goals and identify gaps in rare disease research infrastructure. In addition, by identifying the strengths and weaknesses of the current state of rare disease research infrastructure here at UNC, we can plan how this group can move forward in this space. It is important to consider that there may be overlapping infrastructure needs for rare disease clinical trials and delivery of approved therapies.

Session 4 Group Activity: SWOT Analysis for UNC / UNC Health Research Infrastructure

Strengths	Weaknesses
• Access to UNC Health patient population • PPMH / ISD – UNC Epic implementation • Disease-specific research at UNC • Specialist expertise at UNC • Collaborative culture – research and clinical at same place with connections to one another • UNC Health brand recognition • Multidisciplinary care teams • Vision / Drive / Innovation	• Fragmented registries • No biobanks / repositories • Time constraints • Genetics workforce limitations • Epic has room for improvement • Dissemination of information from research / clinical sides • Regulatory barriers (i.e. data access) • Expenses / Cost (i.e. insurance barriers)
Opportunities	**Threats**
• Access to clinical trials units • UNC partnerships within RTP / industry • Access to e-consults • AI / Other technology • PPMH / ISD – Epic implementation • New disease discovery • Research from bench to bedside • Philanthropy • Care / Nurse coordinators	• Private money / institutions • Further developed institutions / hospitals • Cost • Grant funding limitations • Distractions in HCS away from rare disease • Care inequity

Closing Remarks: Cristy Page, MD

<u>Next Steps</u>:
In order to accomplish the collective workshop goals and move forward, we propose 4 principal Rare Disease Working Groups at UNC:

1. **Identifying Patients with Rare Disease Working Group**
   Concentrating on the identification of patients with rare disease. This group will work in the EHR to improve the Epic landscape for rare disease diagnosis, including electronic phenotyping, computational decision support ("Zebra Button" and "Zebra Flag"), e-consults, and enhanced triage process for patients with rare disease concerns.
2. **Rare Disease Clinical Care Task Force**
   Primarily clinicians, working on improving the harmonization of the clinical care teams diagnosing and treating rare disease. Principally, identifying existing multidisciplinary care teams and key areas where additional multidisciplinary clinics are needed.
3. **Research for Rare Disease Working Group**
   Bench and computational scientists are vital in the diagnosis and treatment of rare disease. This group's focus will be to increase connections across UNC's campus to drive research in rare diseases. Strategies include developing a rare disease patient registry, potential biobanking to generate materials for research, computational approaches, collaboration on development of rare disease treatment therapies, and leveraging existing laboratory capabilities and animal modeling for rare disease research.
4. **Rare Disease Education & Outreach Working Group**
   Focusing on rare disease awareness among providers, patients, and the community. This includes activities such as planning Rare Disease Day events and improving patient and provider education. This group will utilize the ongoing education and outreach efforts of the PPMH Education Initiative.