

**Rare Disease Council Quarterly Meeting  
Tuesday, September 20, 2022, 10:00 AM – 2:00 PM**

**Meeting Location & Address:**

Virginia Hospital & Healthcare Association  
Washington and Jefferson Meeting Rooms  
4200 Innslake Dr, Glen Allen, VA 23060

If you would like to make a public comment or view the meeting remotely, please register here:  
[https://vdh.zoom.us/webinar/register/WN\\_sTAIUmrQvCWboQqUIZl6w](https://vdh.zoom.us/webinar/register/WN_sTAIUmrQvCWboQqUIZl6w)

**Meeting Minutes**

Council Members Present (*ex officio members italicized*): Dr. Lisa Kaplowitz (Chair), Shannon McNeil, Ijeoma Azubuko, Megan O'Boyle, Dr. Peter Kasson, Susan Klees, Dr. Richard Nicholas, Dr. Tiffany Kimbrough, *Dr. John Morgan, Jennifer Macdonald*

Council Members Absent: Jana Monaco (Vice-Chair), Sarah Paciulli, Dr. Samantha Vergano, Dr. Stephen Green, Dr. Stephen Rich, Holly Kearl, Rebecca Goldbach, James Fisher, *Dr. Samantha Hollins*

Virginia Department of Health (VDH) Staff Present: Christen Crews, Katherine Crawford

--

**Call Meeting to Order, Roll Call and Introductions, Review Agenda**

Dr. Lisa Kaplowitz, Chair, called the meeting to order, and VDH staff conducted roll call. The Chair then reviewed the agenda and introduced the first presenter.

**Presentation: Joann Bodurtha, MD, MPH, FACMG, Project Co-Director, New York Mid-Atlantic Caribbean (NYMAC) Regional Genetics Network**

Dr. Bodurtha provided Council Members with an overview of NYMAC, one of seven HRSA funded Regional Genetics Networks focused on improving access to quality genetics services for individuals with genetic conditions and their families. Supported by its partnerships, NYMAC serves as a hub for rare disease work spanning from New York through Virginia and West Virginia, as well as Puerto Rico and the U.S. Virgin Islands. One of NYMAC's goals is to improve the genetics delivery system: for example, through better integrating genetics services with health services, or finding a way to provide care in areas without medical geneticists. A second goal is to determine how telemedicine might make appointments easier for patients and families. A third goal is to educate and support primary care providers and families through identifying ways to make complex information more accessible. NYMAC established teams in each of its 10 jurisdictions to work closely with local family-to-family groups to identify barriers to genetic services, and to guide and improve integration with referrals, genetic testing, awareness at the primary care level, etc. For example, the Virginia team established a Genetics Navigator training for professionals working with families in Virginia, while other jurisdictions initiated projects relevant to their specific needs. NYMAC provides a variety of resources through its website, annual conference, and other support materials.

After the presentation, Council members discussed questions with Dr. Bodurtha. Topics included the importance of including both parent and professional narratives when working to identify and understand access-to-care, referral, screening, and data issues. Other topics included a need to address the low numbers of genetic counselors, to improve public trust in genetics services, to provide ongoing support for individuals after diagnosis, and to improve accessibility to care coordination resources to help identify and address barriers to care.

**Presentation: Tiffany Kimbrough, MD, FAAP, Rare Disease Council Member  
Medical Director Mother Infant Unit & Associate Professor, General Pediatrics  
Children's Hospital of Richmond at VCU**

Next, the Council received a presentation by Dr. Tiffany Kimbrough, a member of the Rare Disease Council and a pediatrician who provides care to children with medical complexity. Dr. Kimbrough shared gaps in care and challenges families face, including a care map that one set of parents put together to help others better understand the complexity of their day-to-day life. Children can need care from numerous specialists to manage diagnoses, and many require home nursing services, which is an issue due to staff supply and insurance coverage. Home modifications, modes of transport, and medical equipment require updates as children grow, and many families have additional financial needs despite insurance.

Dr. Kimbrough reviewed findings where parents of children with medical complexity reported their needs were unmet in a variety of areas, including: patient and family mental health; difficulties communicating; challenges with insurance coverage or obtaining durable medical equipment; dental care; and a lack of respite care or support. Caregivers also noted gaps in home care, case management, financial stressors, and caregiver health. Dr. Kimbrough shared the importance of providers using person-first language, recognizing that each child and family is unique, and factors associated with an increased risk of child welfare involvement.

Dr. Kimbrough next addressed special considerations for pediatric versus adult complex care, noting that pediatric conditions are often rare and heterogeneous, without a standard of care to follow, and that access to specialized care is an issue for rural areas. Additionally, children cannot manage their own care, which increases the need for home care, though low comparative pay rates for home nurses furthers the gap in staffing resources. Primary care providers also experience challenges with providing adequate care due to factors such as: managing multiple medications, meeting patient and family needs within time allotments, rare or unfamiliar diagnoses, coordinating multiple specialty providers, etc. Additional issues related to complex care can include medical errors, and a lack of communication about ongoing care needs among providers, the school system, specialists, and hospitals.

After the presentation, Council members discussed questions with Dr. Kimbrough. Topics included the loss of education and Medicaid services as children transition to adults, and a lack of knowledge and support for adult patients by new primary care providers as compared to the pediatricians already familiar with their care. Council members discussed accessibility and availability of, and knowledge about, Medicaid waivers in Virginia, and expressed interest in a Department of Medical Assistance Services representative speaking to the Council about waivers in a future quarterly meeting. Council members also discussed challenges associated with: home health care, how access to resources and services might affect a caregiver's ability to meet a child's needs, and if an economic analysis might help to identify and address potential areas for improvement.

### **Public Comment Period**

No individuals registered online to provide a comment for the public comment period. Ms. Heidi Dix, who works for the Virginia Association for Health Plans, provided an in-person comment to the Council after Dr. Kimbrough's presentation. She indicated she previously worked at DBHDS, and recommended the Council bring in someone in who is an expert on waivers. She shared about efforts to reduce waitlist times, and noted that DBHDS and Medicaid may have data from a partnership that could potentially help with an economic analysis.

### **Council Updates**

Council members next shared updates about information gathered since the last quarterly meeting.

Council members spoke about the virtual National Organization for Rare Disorders (NORD) Rare Disease Advisory Council Members Meeting in June, sharing how some state councils used websites and social media to raise awareness about rare diseases and their respective councils. Additionally, a few states shared survey efforts and results, including the Chloe Barnes Rare Disease Advisory Council in Minnesota, which presented to Virginia's Rare Disease Council in December. Representatives from the Minnesota council shared that a paper had been published about their patient/family survey, and that a provider survey was also conducted.

Council members next discussed findings from a meeting with the Genetics Team from Virginia Commonwealth University in July, which the Chair noted were very similar to themes the Council had discussed in today's meeting. Topics included: access to care, with wait times as long as six to ten months; a lack of providers, including very few medical geneticists, with care focused primarily around hospitals with academic centers such as VCU, UVA, and CHKD, making access difficult for those in remote regions; telemedicine; challenges with managing special diets; transition out of school; managed care, etc.

Next, Council members reviewed notes from a Virginia Bio Rare Disease Briefing in September focusing on "The Transformative Potential of Gene Therapies." Council members discussed key points from the briefing, including new treatment approaches for rare diseases, with a focus on gene therapy, which could be a theoretical one-time treatment with a very high price tag upfront versus years of healthcare. Various payment models were presented in the briefing, as well as considerations related to treatment access, driving future innovation, FDA approvals, and cost comparisons with traditional treatments.

### **Council Business**

After a break for lunch, the Council reconvened to conduct Council business. The Council did not meet the requirements for an in-person quorum, so the Council was not able to vote or make formal recommendations, but rather discussed each agenda item.

Council members discussed the Council's draft bylaws, including the need to clarify the length of appointment terms after the initial staggering of terms. The Council also discussed the requirement to adopt an electronic meetings policy before Council members may participate in meetings from a remote location or hold all-virtual meetings, as per changes to the Code of Virginia effective September 1, 2022. The Council discussed challenges and barriers to meeting an in-person quorum in order to adopt an electronic meetings policy.

The Chair provided an opportunity for Council members to comment on past meeting minutes, the draft mission statement, the draft vision statement, and proposed workgroups. Council members expressed interest in prioritizing strategic planning ahead of other workgroup items, and noted challenges with moving forward with work items when the Council has not yet been able to meet a quorum. The Council then discussed telehealth and the Interstate Medical Licensure Compact as it could relate to patients from Virginia receiving care in other states, or patients from other states receiving care in Virginia.

Council members then moved to planning for the next quarterly meeting, including proposing agenda items. In one of the two next quarterly meetings, the Council requested that a representative from the Department of Medical Assistance Services speak to the Council about waivers and address specific questions. The Chair also requested that another Council Member present in the next meeting, as Dr. Kimbrough did today.

### **Discussion about Legislative Processes and Public Meetings**

#### **Joseph Hilbert, Deputy Commissioner for Governmental and Regulatory Affairs, Virginia Department of Health**

After completing the Council business portion of the agenda, Mr. Joseph Hilbert led a discussion with the Council about legislative processes and public meetings, as requested by Council members in the last quarterly meeting. Mr. Hilbert provided Council members with an overview of key dates and timeframes in the legislative session and processes for proposing changes to legislation. Mr. Hilbert noted that the Council's annual report could serve as a resource where the Council could make recommendations for changes to the Code of Virginia. These recommendations could potentially then be developed into a legislative proposal through an agency initiating the process to introduce a bill, or through a delegate submitting a bill.

Mr. Hilbert also discussed requirements for public meetings, including changes that took effect on September 1, 2022, and addressed Council member questions. The Council confirmed the need to meet the requirements of an in-person quorum in its next quarterly meeting in order to adopt an electronic meetings policy to allow Council members to participate in meetings from a remote location.

### **Council Member Closing Discussion, Adjourn**

After Council members completed their discussion, the Chair adjourned the meeting.