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Illinois Rare Disease Commission

August 21, 2023
12-1 PM

Focus Topic: Access to Genetic Counseling, Care, & Treatments

MINUTES

Name	Present (Y/N)	Role	Affiliation
***Maria "Ria" Pollock**	X	Affected / Caregiver; Advocacy Group	*(CHAIR)* Living with rare disease, Vice chair
Vacant	-	***Appointed***	
Joyce Clay		Affected / Caregiver; Health Professional	Daughter with rare disease
Tim Cunniff	X	Industry	Paragon Biosciences
Stacey Feuer		Affected / Caregiver; Health Professional	Living with rare disease
TaLana Hughes	na	Affected / Caregiver; Advocacy Group	Sickle Cell Disease Association of Illinois (SCDAI)
Katherine Kim	X	Provider	Genetic Counselor Lurie Children's Hospital
Lara Pullen	X	Affected / Caregiver; Advocacy Group; Industry	Chion Foundation
Stacey Pigott		***Pending***	
Vacant	-	***Appointed***	
Vacant	-	***Appointed***	
Vacant	-	*Policymaker*	*TBD*
Linda Holmes		Policymaker	Living with chronic illness
Sonya Harper		Policymaker	
Vacant	-	*Policymaker*	*TBD*

Attendance: Victoria Richter, student with rare disorder, Kate Segal, Sarepta Therapeutics, Samantha Ropski, EDS Chicago, John Conrad, iBio, Dana Clemmans, Xenis Pharmaceuticals, Ann Vogel, iBio, Hank Chiupi, Kurt Anderson, Matthew Botos, Alyssa Valentine, ISGP, Kevin Johnson, Amgen-Horizon Therapeutics, Roy Pura, CSL Behring,

Matthew Botos, iBio, Ken Sprague, BioMarin Pharmaceutical, Lindsey Viscarra, NORD, Jonathan Buckner, Biogen

The meeting was called to order by the Chair, Maria Pollock, at 12:01 (with summer highlights). There was not a quorum. Recording started at 12:03.

Jason Rothstein, Vice Chair resigned for personal reasons.

Late Submissions: None

Approval of Meeting Minutes (06/21/2023): deferred due to lack of quorum.

Old Business: Approval of Meeting Minutes (06/21/2023) and amendment of Bylaws were deferred due to lack of quorum.

New Business: Tim Cunniff agreed to the appointment of interim Vice Chair until elections can be held.

Public Comment: Samantha EDS Chicago, described her experience living with a chronic disease (EDS). She suffered a minor aortic tear which led to her clinical diagnosis. She is having genetic testing now. Her family has other cases of medical mysteries, relatives with similar issues. Samantha has encountered a lot of challenges in getting genetic testing because of her age (over 18), not having documented developmental delays (denied by insurance), as well as provider pushback (resistance to ordering). Samantha spoke to the difficulty getting appropriate treatment and management without a diagnosis. She believes that molecular genetic testing for herself could potentially advance understanding of medicine generally for other with similar health problems, as well as for herself.

Lara acknowledged the feeling of need for genetic diagnosis and also the fact that the impact on the treatment plan is uncertain. She asked Samantha her feelings as a patient. Samantha stated that initially she wondered “what is the point” of molecular testing? Her medical condition is already complicated, would it add one more thing to the list? On the other hand, would it help medical providers take her more seriously as a patient? Katherine spoke about the mysteries still remaining in genetics and unknown connections between genetic changes and medical issues. Medical research is needed to help patients, and their providers, understand their conditions.

Ria shared that she personally had genetic testing in 2015, 2017, and then whole exome sequencing (WES) in 2020. After surgery in 2015 she learned that she does not respond to most pain medicines (a topic for another meeting). Ria is not followed in genetics clinic currently. She contacted a colleague in industry regarding her pharmacogenomics

result, hoping to learn how it related to her pharmaceutical response. At the time that the molecular variant was identified, there was no information on how that variant might impact her. She has learned since that it affects the liver and inactivates many medications.

Katherine presented and led discussion around the topic area for today's meeting: access to genetic counseling, care and treatment. She shared a powerpoint presentation with background information and data. Katherine shared that patient wait times are increasing. In some instances, these are 18 months to 2 years. One issue is the work force; another is lack of funding at facilities to support staff. Another barrier is highly variable insurance coverage of genetic services. The cost of molecular genetic tests can be very high. Medicare (public) and private insurers limit coverage; the need for provider and patient to show "medical necessity" is a barrier; prior authorization is another hurdle (health care plan/PCP). Plans may limit where health care is received. Some health plan networks may not include genetic providers. Prenatal services seem more easily accessed in regard to approvals needed. Additionally, Katherine mentioned:

- Healthcare disparities due to cultural and socioeconomic factors;
- Increased complexity of care limit (impacts resources available); and
- Telehealth regulations. There was relaxation of regulations to extend coverage across state lines during the Covid pandemic. This has now returned to the prior state, which is fractured; regulations limit care across state boundaries.

Katherine found not that many sites offering genetic services, given the population base of the State of Illinois; most are clustered in Chicago. She went on to discuss the genetics workforce. Specific to Illinois, clinical genetics workforce development is limited. For genetic counselor training the Northwestern University Program graduates 20 students per year, as one of the largest training programs in the US. It is the only GC training program in Illinois. There are no active medical fellowships in medical genetics in Illinois. One will be starting at Lurie Children's in 2024. The Illinois Department of Financial and Professional Regulations (IDFPR) has months' long delays to obtain a license to practice. Nationally there is a need for more workforce development as well. About 300 genetic counselors enter the workforce annually. In 2019, there were fewer than 5,000 nationally. There are many fewer clinical geneticists. About 1,200 total nationally and fewer than 20 joining the workforce each year. The number of residency training programs has decreased. Many in practice are within a few years of retirement.

Ria added at a rough estimate, that works out to 25,000 patients per geneticist. And added, how does this impact view of what is "medically necessary"?

Alyssa, a genetic counselor working at Stroger and member of the Illinois Society of Genetic Professionals (ISGP) agreed with Katherine's summary and the need for a triage system that may help those "at the back of the line". One need she sees is for Illinois

Medicaid to recognize her and other genetic counselors as medical providers for the purpose of billing and reimbursement of services. This is a major focus of ISGP at this time. ISGP is promoting that issue in order to increase access to services. Hospitals and health systems are less motivated to hire, to support health professionals who are not able to bill for the services provided.

Lara asked if genetic counselors can bill in other states? Alyssa responded that CMS federally does not recognize them as providers. State by state – e.g., in Indiana and a few others, genetic counselors have Medicaid recognition for billing. Lara asked if data around billing policy correlated with number of providers. Alyssa responded that this may vary by state. Genetic counselor training tends to be supported in metropolitan areas near large university-based medical centers. ISGP hasn't looked at this issue specifically.

Katherine mentioned that there is a specific CPT code used to bill for genetic counseling services. That CPT code is reimbursable by Medicaid/Medicare. BUT genetic counselors are not recognized as providers to bill under that code. They want to streamline the process for providers to deliver services to patients on Medicaid plans by allowing enrollment, inclusion in the Impact portal. Currently, Impact does not recognize them. Ria clarified this means they cannot be assigned an MPI number. Katherine summarized that genetic counselors encounter barriers at the state level. Anything IRDC can do could help. For example, promote quicker receipt of licenses; making sure all allied health professions are included in the Impact portal for reimbursement of services provided from public insurance. These would be the recommendations she would suggest.

Ria mentioned that some of these are issues at the national level as well. Illinois could possibly create a state-based waiver. Ria has looked into this issue, and Illinois would be a pioneer in crafting this sort of waiver. IF the profession could be added to IMPACT without requiring a formal waiver, that may be more efficient. Also, Ria emphasized the importance of collecting and analyzing data in order to understand the effect of genetic variants.

Announcements

a. Next meeting: Monday 18 September 2023 from noon to 1 pm via WebEx
b. Focus Topic: Access to Pain Management for Rare Disease Patients

Adjourn: the meeting ended at 1 pm.