Illinois Rare Disease Commission

February 22, 2022
12 PM -1 PM - Virtual

MINUTES

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

**Cochair, ** Vice Chair, ***Withdrawn*

Attendees: Joan Ehrhardt (IDPH, Facilitator); Melissa Stalets (IDPH); John Conrad (iBIO)

Welcome and Introductions (began at: 12:02)
Maria Pollock (Ria) began introductions, followed in turn by the commission members in attendance. Maria lives with a rare debilitating connective tissue disorder, diagnosed later in life**;**

Joyce Clay, has a child with Pitt-Hopkins syndrome, a rare condition with recently recognized genetic cause and variable features, frequently including seizures. The diagnosis was made at age 25 yo after multiple tests. They live in central Illinois, so focused on access in that part of the state. She is joining from the hospital today where her child is hospitalized.
Tim Cunniff, represents industry, research and development, predominantly focused on therapeutics and rare disease;
Jason Rothstein, represents community-based advocacy, providing awareness, education and resources about rare genetic conditions impacting the Jewish community. Interested in community service and improving the landscape for all in regard to rare genetic diseases.
Lara Pullen, has a young adult child with Prader-Willi syndrome (PWS), is an immunologist and works on clinical trials make therapeutics available, e.g., repurpose medications to move from off label usage to approved treatments; this is for treatment of narcolepsy in PWS and is expected to enter phase 3 trials soon.
Talana Hughes, has a young adult child with sickle cell disease and represents community-based advocacy;
Stacey Feuer, lives with a rare disease. She was diagnosed with Gaucher disease type 1 as a younger adult and experiences chronic medical issues (joined at ~12:27). *There was a late change in meeting link that caused some confusion.*

Member of Public – John Conrad, coming from industry. John is President CEO for the trade association for the life sciences industry in Illinois and helped write legislation to establish the commission and extend through 2026.

Late submission from public to consider the proposed SB2246 and give input- https://www.ilga.gov/legislation/103/SB/10300SB2246.htm. This legislation is supported by the connective tissue disorder community because it may help protect families from false allegations of abuse based on lack of understanding of these disorders and association with easy bruising. Maria requested members to review and be prepare for further discussion next meeting.

Approval of agenda with addition: Quorum was established. Agenda was approved.

Old Business
Approval of meeting minutes and bylaws:
- **Minutes**: Approved (none opposed, none abstained)
- **Bylaws**: Tabled for lack of 2/3 majority

New Business
Education and Outreach Priorities:
Maria opened discussion by stating that the RDC would like to have more of a web presence, with resources and community connections (e.g., life with rare disease). She asked the members to share their thoughts.

Lara - would like to have a section focused on transition to adult care. She herself wants to know and make it easier for all families/caregivers to find what resources there are for young adults with disabilities and what Illinois offers. Particularly concerning transition of care for those individuals. Young adults are particularly vulnerable.

Ria – agrees and mentions that as a graduate student the experience of young people with rare disease is a focus of her studies;

Joyce – agrees with Lara and adds that it is especially hard for young person who has left school and community supports are far below what support was previously in place for them. The care burden becomes huge for families at that point.

Jason –another dimension is the diagnostic odyssey to an accurate diagnosis. Would like to increase awareness of genetic counseling. Many people lack awareness, including providers. What is the place for prevention information, resources available for public, etc. and reproductive implications? Does this fit with the RDC mandate?

Joyce – focus while her child is hospitalized is with their current challenge: lack of understanding by insurance companies. They have gone through approval requests for medication/treatment for weeks. There is a big need for a liaison for rare disease clients/complex care patients within the insurance company.

Talana –increasing awareness of rare diseases overall in Illinois, not only the Rare Disease Commission and efforts and resources in Illinois. Ria agreed with the point that students feel isolated.

Tim – helping families find clinical trials. FDA has a website, but can people find it or navigate it? Sites listed for available trials may not be local. Families may not know of travel coverage availability. Families can be flown to the study site and have travel costs covered. Not know that there is travel assistance can be a deterrent. Regarding qualifying for trials: expanded access policies such as opportunities for single patient compassionate use and "Right to Try" policy.

Lara – add to a rare disease website information about medical research; studies in Chicago and elsewhere. A good focal point. Perhaps include or focus only on those with travel options. Contact the manufacturer to find out if there are travel options. Visit ClinicalTrials.gov. Include information about compassionate use. Link to the FDA. One pager for other options to access drugs. That could be a great resource for patients and providers.

Jason – agreed and mentioned it would be important to include some sort of primer for patients who have questions: we have a diagnosis what now? They need a handout or tipsheet, or something more interactive.

Stacey Feuer joined at ~12:27 and briefly introduced herself. Ria updated Stacey regarding the discussion.

Ria suggested that the RDC may begin to address these gaps through providing web resources/website development. Ria asked how such a site might be maintained. Joyce suggested members might approach complex care teams at health centers and check on the resources available. Their family has been helped by the Pediatric Adult Complex Care team (PACC) in central Illinois. You may also find this at e.g., Lurie Children's and other medical centers. OSF multidisciplinary team for patients with high medical complexity. That team, their service, helps with continuity of care. They help families know what resources and services are available. That could be a starting point. Lara stated she is a professional medical writer and she does not mind writing content. She stated that the information must be very generic unless there is the ability to keep the information current. Lara does not know where to find information about transitional care. Joan interjected to share her understanding of IDPH agency policy around commission facilitation and resources for web hosting and support.

Ria noted that while we wish to share resources for all, sometimes there are resources (for specific conditions), and sometimes not. Rare diseases without a name, for example. Ria stated that 1 in 4 people has a rare condition. She asked if private foundations might take on the role of sharing and publishing resources. Joyce described the role of the PACC in providing multidisciplinary care as a kind of

bridge between pediatric and adult care. They provide a constant in health care and make a big difference in continuity of care. Lara mentioned that many foundations focusing on rare disease are national. Supporting community level focus is a challenge. Non-profit organizations may not have that focus or capacity to provide that function**.** Lara is unsure what not for profit would fill that role versus state government. Ria agreed and asked if the IRDC might learn from other states.
Lara expressed frustration about lack of dedicated funds or resources or ability implement best practices in order to make a meaningful difference for rare disease patients and their families. Ria agreed and suggested that the RDC might develop a proposal regarding the needs. Stacey agreed and asked about a project lead and funding, to include supporting position (assigned staff support). She acknowledged that everyone on the commission has full-time lives, and it is hard to add more. Is it possible that the state could contribute resources such as money for a person? money for a project? What should RDC ask? A dedicated position? Webmaster? Ria offered to look into that as well, for example, if able to have budget to fund dedicated staff.

Ria wondered how can medical providers be better equipped to recognize, treat, and relate to individuals with rare disease? Some states require specific educational credits/focus for medical providers in training. How does a provider distinguish between the unusual presentation of a common condition and the usual presentation of a rare condition?

Ria suggested commission members bring resources for a website to the next meeting, and that they conclude business at 12:50 to proceed to elections.

Selection of Chair and Vice Chair: Ria asked for volunteers for the position of Chair. Nominated Ria; seconded – voted/approved. Ria for Chair. Requested volunteer for Vice Chair(s). Jason volunteered. Nominated Jason; seconded – voted/approved.

Next meeting –

Joan asked the RDC members to consider meeting bimonthly during 2023 due to competing priorities at IDPH. The RDC determined that monthly meetings would be best.

Meetings will be monthly, virtual, typically on the third Monday of the month, for 1 hr; 12 – 1 pm.

Public Comment- None

The meeting ended at 1:00 pm.