Illinois Rare Disease Commission

April 17, 2023
12-1 PM

Minutes

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

*Chair, Vice Chair

Attendees: Joan Ehrhardt (IDPH, facilitator), Christine Hannon, Mary Broderick (Representative of the Family Justice Center). Keri Lachapelle (SIUE Nursing Grad); Anissa Reed (Associate Director of State Policy with the National Organization for Rare Disorders (NORD)); Elizabeth Hellebusch (SIUE Nursing student); Michelle Weidner, MPA (Executive Director, Family Justice Resource Center); Ann Welsh (Local Public Health Nurse, Kankakee Co HD); Jesse Austin (SIUE nursing student). Patti Krueger: Lori Heitman;

Sue Silbernagel (Wegener's granulomatosis); Jesse Austin (SIU DNP Program student); Samantha Ropski (Administrative assistant, EDS Chicago Support and Awareness Group, graduate student and rare disease patient)

Meeting started: 12:02

Welcome and Introductions: IRDC members introduced themselves. Members of the public in attendance were identified. The policymaker appointment previously held by Jason Barickman is now vacant. Joan will notify IDPH Governmental Affairs.

Late Submissions: addition to legislative review.

Approval of Agenda & Meeting Minutes (03/20/2023): The agenda was approved with none opposed and none abstaining. The minutes were approved with as submitted with one opposed and none abstaining.

Old Business: There was not a two thirds majority of the commission present, so the revised bylaws could not be approved. The IRDC 2022 Annual Report was approved with none opposed and none abstaining.

IRDC Semiannual In-person Meetings

Jason met with Ria separately to discuss in-person IRDC meeting planning. Ria is meeting with other RDC members individually as well. If IDPH wishes to host the in person meeting(s), that would be fine. Also, Jason has secured ability for the Jewish United Fund, Sarnoff Center (JUF) to host an IRDC meeting in their downtown Chicago offices. Stacey asked if the meeting would be outside of business hours? She would find it very challenging to participate in person during business hours. The idea is for a two to three- hour working meeting, ideally. IRDC has not had that kind of in person meeting previously and could not during the pandemic. There are two proposed in person meeting timeframes and locations, one in the next couple months in Chicago and one next spring in Springfield (around Rare Disease Day):

Chicago (May/June 2023, October 2024): Board members noted that this is right around graduation time. Jason has reserved space in their offices. Looking at already scheduled. JUF can offer assistance with parking and lunch. Joan will check on capacity to facilitate a longer meeting, plus working groups with hybrid options for IRDC members and the public. Joan is to check and connect with Ria about meeting in compliance with OMA requirements. Ria told the commission that we will survey attendance. Since meeting with fewer than five commission members may not be workable.

Legislation –

SBOO67 Newborn screening (NBS) expansion: metachromatic leukodystrophy (MLD): Katherine discussed where this bill is in the process of development. It has recently passed out of committee. She discussed MLD, describing the condition symptoms, age of onset, etc. Neurodevelopmental, progressive deterioration of abilities, cognition, behavior/mental health over time. Currently, the only treatment is HSCT. Outcome most effective when occurs prior to onset of symptoms. Currently multiple treatments in development including enzyme replacement and gene therapies. Bill would introduce adding MLD to state NBS so all states in IL would be screened from the same amount of blood currently collected. Additional costs would be billed to patient insurances for testing. Any children screened positive would be eligible for DSCC coverage and assistance with diagnostic testing. Screen positive – referral to consultant – further diagnostic testing to confirm. As with others, if insurance issues covering the evaluation, DSCC will assist in covering the costs of the diagnostic work up. Hoping will be approved. It will be mandated; however it will be a lengthy process. Kim discussed the various requirements of providing / implementing a new screen on the NBS panel.

IRDC Flyer -

Lara met with Ria separately to discuss updating the IRDC flyer. Lara has reviewed it, and commented that it is very superficial. Lara would like to understand the point of the IRDC flyer. Is that something that is still done? As a writer her question is who is the target audience? What is the purpose? Ria shared the flyer on screen. Lara says that to have the information available on the website is critical. Stacey commented that it could be useful to have a couple versions with a blurb for specific audience. For example, a sentence or two targeting physicians; potential patients and families; a little differentiation. Lara agrees. And what is the message – is it a resource? And for whom? If asked by others in the rare disease community, Lara would suggest NORD as a resource before IRDC. A flyer could notify the community that meetings are open to members of the public. Stacey agrees that IRDC does not want to raise expectations that they cannot meet. Is it a callout to both, to increase knowledge of what is happening for Illinois residents with rare disease [by attending IRDC meetings]? Jason asked to what end? What is the purpose of attendance for patients and/or providers? Others interested?

Lara mentioned that it could be possible to have an open house or coffee hour or some in person chat venue around rare disease day. A call for action could get lost if disseminated after a flyer. Ria sees the flyer as a helpful handout for any who are not online much. Ann Welsh requested to comment and was acknowledged by Ria. Ann said that she did not know IRDC existed until recently. She agrees that it is good to know that it exists. And she would like to be able to share that knowledge with others. Good for Ann (LHDs) to know that IRDC exists and know of possible resources for assistance. Jason said that he sees value of input from committee, for the public to attend and comment on relevant issues. Would members be promoting meetings for input? If so, then it may be good to be more systematic. For example, to invite the public to comment about a topic of the month, such as insurance coverage or whatever major topic. Communication would then be more targeted and the public could attend as relevant and make comment intentional vs. random. Lara said she agrees if the purpose is to hear from the public. In that case, the agenda needs to be structured accordingly, with public comment identified at roll call. Annissa Reed, NORD was acknowledged by Ria and asked if more of a one-pager be beneficial?

Public Comment

Michelle Weidner shared her story and links to Bill: SB0376 and the fact sheet: www.famjustice.org/protecting-innocent-families-act. [Note: scroll down to see the fact sheet]. They are developing protections against families being wrongly accused of child abuse. Michelle described requirements of DCSF related to this policy.

She went on to say that bruises, unexplained fractures, [listed others] trigger child abuse evaluation. Providers under contract to DCFS and …are presenting themselves as treating providers. There is lack of transparency around their role. There is a lack of consideration of rare conditions that can cause symptoms that overlap with those triggers. Stacey acknowledged that these are important issues. Legislator – how does this bill differentiate from existing protections? There is a practitioner code of ethics, child abuse pediatricians should be identifying themselves, their role, contracts, affiliations. There is a right to a second opinion. However, DCFS is not required to accept or consider the second opinions. Safety plans, etc. are being put in place (to protect the child) that are causing harm to children and families. Ria thanked the speaker.

Mary Broderick described her family's experience with false allegation of child abuse 20 years ago. Her son has a condition ~1/2200 not considered rare (hydrocephalus). She echoed concerns about the practices of providers around these investigations. Some are promoting an app to prevent child abuse, support early action. Mary has concerns about the way the app is being promoted, use and follow-up-action taken/recommended (fairly extensive). Mary shared a link to a news story regarding a concerning approach: https://www.luriechildrens.org/en/news-stories/lurie-childrens-hospital-launches-app-to-help-screen-bruises-in-young-children-for-potential-abuse/

I am Lori Heitman and I am a rare disease patient. My affiliation is with the Chicago Ehlers Danlos support group. I joined today's meeting to find out what I can do to advocate for Ehlers Danlos patients.

Michelle Weidner to everyone: Our bill was originally SB 2246 but the number is now SB 376. Senator Linda Holmes was a sponsor of SB 2246 and we anticipate that she will transfer her sponsorship to SB 376 this week. We'd like to get the support of the Rare Disease Commission for this bill. Ria thanked her for comments.

Announcements

Next meeting: Monday May 15, 2023 12-1 pm

Adjourn: 12:58.

ILLINOIS
RARE
DISEASE
COMMISSION

The **Illinois Rare Disease Commission** was established to increase awareness of rare and orphan diseases that impact the lives of 1 in 10 people. There are more than 7,000 unique and rare disorders that affect many Illinois residents and their families. The commission is made up of representatives from health care professions; people affected with rare disorders, their parents, or caregivers; and government officials.

Pursuant to 410 ILCS 445, the commission makes recommendations to the General Assembly in the form of an annual report. Commission activities are extended through 2026, pursuant to Public Act 102-0671 (Section 75).