IDPH
ILLINOIS DEPARTMENT OF PUBLIC HEALTH

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Illinois Rare Disease Commission

May 15, 2023
12-1 PM

Minutes

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

*Chair, Vice Chair

Attendees: Joan Ehrhardt, IDPH facilitator; Aaron and Krystle Myers (MLD Foundation); Mary Broderick (FJRC); Samantha Ropski (EDS Chicago); Karin Pritikin

Welcome and Introductions: IRDC Members and public attending introduced themselves. Krystle and Aaron Myers joined to speak about their family’s experiences with metachromatic leukodystrophy

(MLD), challenges around receiving a correct diagnosis, living with MLD, accommodations in education, and other needs.

Meeting started: 12:00 pm

Administrative Updates: member terms have expired. Interested members must submit an application and updated resume. Joan will send an email to the IRDC members regarding renewal applications.

Late Submissions for Discussion: none

Review of Discussion Agenda & Meeting Minutes (04/17/2023): no action

IRDC Semiannual In-person Meetings

Ria asked Jason to describe the JUF spaces. If there is not a requirement for meetings to be at IDPH facilities, then JUF is happy to host. They have a conference center in their building. They have video conferencing. They cannot bring in outside food. They have Kosher kitchens so outside refreshments are not permitted. There was discussion of seeking physical locations downtown Chicago in IDPH facility (e.g., in the loop, W Washington, plus Bellwood, plus Springfield). The IRDC determined to postpone further discussion regarding requirements needed for in person meetings.

Legislation Updates:

SBOO67 Newborn screening (NBS) expansion: metachromatic leukodystrophy (MLD

Katherine provided update that this passed the Senate last week, so now has passed both houses and is on the Governor's desk. They are anticipating signature, with coordinated media/PR event. Katherine thanked the Myers for their work. Katherine described the process of newborn screening validation and a 6-month pilot of select hospitals to prepare for statewide implementation. Ria asked the Myers to share their experience, briefly (2-3 minutes). Their daughter's disorder began with coordination/balance issues, plus cognitive loss of skills. Due to professional medical experience, family recognized neurological symptoms. They went on to describe the diagnostic odyssey. Mayo Clinic is participating in clinical trials for gene therapy. However, their daughter did not qualify because of the delay in diagnosis. They described additional challenges accessing needed support services. Their daughter is receiving some medical care out of state. The IRDC acknowledged the Myers family's struggles and thanked them for their advocacy.

Ria pointed out the similar needs of many individuals and families. She discussed the need to get beyond the "buddy network" of finding the right provider or services or other resources. Ria expressed a call to members to look at medications, treatments, and therapies; legislation for providers, families, and individuals to live better with rare disease; and opportunities to raise awareness of rare disease. Allin order to create better support for individuals and families living with rare disease in Illinois. IRDC is creating community through communication today.

SB 0376-SFA2: Disclosing investigation to alleged child abusers.

Mary Broderick provided updates regarding the recent amendment. The goal of the amendment is that doctors meeting with a family must discuss their affiliation including any with DCSF, and families have a right to a second opinion. Mary described her family’s experience with wrongful allegations of child abuse and emphasized the need for transparency in the process of investigations.

Ria agreed transparency is essential, pointing out that sometimes there is lack of transparency regarding research and inclusion of patient/client information in research without their knowledge.

Public Comment: Karin spoke about her experience. She discussed her initial medical event, treatment, recovery, and subsequent development of a rare endocrine / rheumatologic disease. She has had great difficulty finding medical care in Illinois. Ria expressed her sympathy for Karin’s experience. TaLana echoed the sentiments. Ria discussed potential strategies and avenues for change from Medicaid waiver to new therapeutics. Ria mentioned increased capacity for genetic/genomic testing.

Announcements

Next meeting: Wednesday June 21, 2023 12-1 pm (members present agree that is on their calendar and they are available) facilitation, administrative one hour meeting, in person, with a virtual option.

Adjourn: 1:00 pm

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