ILLINOIS DEPARTMENT OF PUBLIC HEALTH
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Illinois Rare Disease Commission

December 18, 2023
12-1 PM

Minutes

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

Attendees: Joan Ehrhardt, IDPH Facilitator, Brielle Dozier (Artia Solutions), Steve Patterson (Acadia Pharma National Account Manager), Hank Chiuppi (rare disease patient), Samantha Ropski (EDS Chicago leader, graduate student and rare disease patient), Sylvia Washington (United Mitochondrial Disease Foundation Ambassador, and Founder/CEO One Drop Rule Rare Disease Foundation)

Welcome and Introductions – *Ria P./all at 12:01*

Late Submissions - None

Adoption of Agenda & Approval of Meeting Minutes

a. Adoption of Agenda
b. Approval of Meeting Minutes (11/20/2023) – approved with corrections.

Old Business

a. Approval of Meeting Minutes (09/18/2023, 10/17/2023) – approved.

New Business

a. IRDC 2023 Report – Maria provided a draft to all committee members by email prior to the meeting this morning.

Public Comment

Discussion – *Ria P. /all*
2023 Commission members' report and member challenges and hopes for 2023:
Maria discussed her hopes and frustrations. TaLana discussed rare disease advisory activities nationally and within Illinois. She also mentioned recent Food and Drug Administration (FDA) approval of treatment for people with sickle cell disease. Joyce agreed with challenges of medication approval, coverage, and so forth. Her daughter has been hospitalized for approximately seven months out of the year in 2023. Lara mentioned the effort to move their son to a community in Arizona. The adjustment has been challenging. What does independence look like for the medically fragile? How can one balance the desire for independence with the risks for gaps in care? Lara now has more time to work professionally. She has been able to collaborate with many stakeholders on a paper about the many levels of challenges and potential opportunities to address them. Second paper is planned on communications and disconnects between clients and medical professionals. Tim mentioned excitement about moving from small chemical therapies to molecular – genetic based therapies, including some potentially curative strategies. Epidermolysis bullosa was called out as specific conditions with new therapy to build/replace collagen. Treatments for sickle cell disease and other blood disorders are first to be approved; nephropathy treatments – on the verge of large number of new approvals coming. Maria also referenced improvements in molecular methods to decrease the potential for cancers as a risk of treatments.

Sylvia mentioned that she learned about her own mitochondrial disease which was confirmed by specialists out of state. She states also being homozygous for Tay Sachs disease. Sylvia mentions that she will not be treated for her mitochondrial condition - CPT2 deficiency – until spring 2024. Sylvia is now a mito-champion. Her story is going to be documented. Maria asked Sylvia to contact her so that the information could be shared generally. Enddenials.org. [developed by Maria with the help of IRDC members and public] Sylvia commented about the discrepancy/difference between her diagnostic odyssey vs. those currently diagnosed following newborn screening. There are also challenges accessing appropriate treatment as an adult, with private or public (CMS) payors. Lara mentioned the need to continue focus on access to treatment and needed supports in tandem with improved diagnostics. In clinical use of whole genome sequencing (WGS), whole exome sequencing (WES), genetic panels, diagnostic filters are applied (e.g. race-based) which does

not recognize multiracial ancestry. Sylvia says her genetic CPT2 variants are prevalent in European, aka white, ancestry.

IRDC in-person meeting in Springfield for Rare Disease Day 2024 (Thursday, February 29, 2024 – In person early March, TBD)

Announcements
Next meeting: TBD (Monday 15 January 2023 is Martin Luther King Jr State Holiday)
There was discussion of Illinois Rare Disease Day activities. Individual groups may want to conduct activities separately also on February 28th or 29th, as National Rare Disease Day is typically the last day of February. Illinois Rare Disease Day activities in Springfield will be on Monday 6-7 March 2024 co-sponsored by iBio. Legislators return to Springfield Tuesday March 5.

-iBio will sponsor a Wednesday morning breakfast for legislators in the Stratton Building.
-Opening Ceremony Monday 6 March 2024: Kristen Bauer is to develop fliers for distribution. Conference room at the State House is planned for award ceremony in the evening. Awards for Rare Disease were mentioned.
-Need an announcement for rare disease day.
-Wednesday Feb 7th Maria will start to distribute fliers in Springfield. TaLana offered to help distribute fliers on that day.
-In person meeting of the commission. (Stratton Building – no charge for space, no food served during meeting). IRDC In-person meeting Tuesday 7 March 2024: 10:30-12:30 or 10 am to noon.

Concerning the 2024 Meeting Calendar, IRDC members approved monthly meetings, on Wednesday when Monday is a state holiday.

There was some brainstorming around discussion topics for 2024. Maria mentioned developing a survey in 2024, e.g., RDC members and all thanked Maria for her efforts this year and the end result. Lara suggested identifying a legislative goal for next year, even if small. For example, invite a legislator to present to IRDC and discuss something doable in the realm of policymaking. This could provide a concrete ask for the in-person visit to Springfield. Maria said access and coverage to compounded medicine, medical food and … some research in that direction. Stub bill (placeholder) is in development. For January, what are asks and what are posters that need to be there for legislators to see. Can one of the sponsors come to the next meeting? How can IRDC move the bill forward? Maria stated that the policymaker needs to demonstrate that there is a public need (not personal pet project). For example, do they need survey results or testimonials? There will be a more specific call out if more information is available about specific needs. Maria will ask Representative Harper to come to the January meeting to present, and perhaps Bill cosponsors.

Adjourn: 1 pm