ILLINOIS
RARE DISEASE COMMISSION

ANNUAL REPORT 2023
submitted by
Maria G. Pollock - Chair

2023	**The Illinois Rare Disease Commission purpose is to increase awareness of the more than 10,000 unique and rare disorders that impact the lives of over 1.5 million Illinois residents and their families.**

The Illinois Rare Disease Commission (IRDC) received no funding for raising awareness, research, and events outside of the Illinois Department of Public Health (IDPH) budget. IRDC members volunteered their time and expertise.

ACKNOWLEDGEMENTS

The IRDC would like to thank the following for supporting the commission's work, and for their commitment to centering people with rare disease and their families' voices in efforts to address health, care, and support gaps; and to build and improve a service network for rare disease patients and their families.

The patients, parents, and community leaders, who participated in the IRDC meetings, for making sure the voices of people most affected by rare disease are at the forefront of advancing systemic changes that will create better services throughout Illinois.

Joan V. Ehrhardt, CGC from IDPH, for working tirelessly to help the IRDC to become efficient and relevant in identifying the issues of rare disease patients across the state.

ILLINOIS RARE DISEASE COMMISSION - WHO WE ARE

The **Illinois Rare Disease Commission** (IRDC) was established in 2017 to increase awareness of rare and orphan diseases that impact the lives of 1 in 8 people. There are more than 10,000 unique and rare disorders that affect an estimated 1.5 million 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, pharmaceutical industry, researchers, and government officials.

The IRDC is composed of 15 members. Eleven members are appointed by the governor, are residents of the state, and are familiar with the concerns, needs, and recommendations of people with rare disease based on their knowledge, experience, or position. A minimum of five such appointees are people who either are themselves affected with a rare disease or who have a family member living with a rare disease.

The final four members are policymakers:

· Two are from the Senate (one each appointed by the Senate president and the minority leader)
· Two are from the House of Representatives (one each appointed by the House speaker and the minority leader)

Illinois Rare Disease Commission Members 2023

Commission bylaws (Appendix A) provide for meeting and conducting business with current active members.

Governor Appointees

· *Susan Axelrod (Rare Disease and Bioscience Industry Representative) - resigned*
· *Maria Bellefeuille, Co-Chair (Rare Disease Representative)- resigned*
· Joyce Clay (Rare Disease Representative, Health Professional)
· Tim Cunniff (Bioscience Industry Representative)
· Stacey Feuer (Rare Disease Representative, Health Professional)
· Talana Hughes (Rare Disease Representative, Rare Disease NPO)
· *Katherine Kim (Health Professional)- resigned*
· *Stacey Pigott – pending approval*
· Maria "Ria" Pollock, Chair (Rare Disease Representative)
· Lara Pullen (Rare Disease Representative)
· *Jason Rothstein (Rare Disease NPO)- resigned*

Policymakers

· Sonya Harper
· Bill Hauter
· Linda Holmes

Dear Readers,

We are the members of the Illinois Rare Disease Commission (IRDC). At this very moment, over 1.5 million people in Illinois are struggling to get care for a rare or unknown disease or illness. It's time that we talk about the need to address the burden families and individuals with a rare illness are asked to shoulder.

During 2023, we found that people who raised issues and provided testimony during public comment in the IRDC meetings said that they had looked for care and were unable to find it. Families are asked to make unspeakable sacrifices: leave their job to give care, sell their house to pay for medical care, travel to other states to see specialists, live destitute. Children and adults suffer preventable deaths. This is heart-breaking!

Illinois provides care and services for people with preventable illnesses but when someone has been dealt an unlucky hand by nature's lottery they are on their own. We believe that ignoring rare disease patients and excluding them from insurance and public services is wrong.

We need to start creating policies that center around relieving individuals and families by providing care giving services, genetic testing and therapies, pain management, and palliative care inpatient, outpatient and at home. We need to protect rare patients from insurance discrimination and false accusations of child abuse and neglect.

Illinois has a lot of catching up to do. We need to invest in services, resources, and pass legislative mandates to protect Illinois patients with rare disease. We hope that you join us in the fight for rare disease patients and their families to end the financial bleeding-out of families and individuals living with rare disease. We believe that we can lead this country in providing compassionate care and have well-staffed public service agencies that help make health services accessible, affordable, and of top quality for rare patients.

Sincerely,

Maria G. Pollock
Rare Patient
Illinois Rare Disease Commission Chair

OVERVIEW AND ACTIVITIES

The Illinois Rare Disease Commission (IRDC) is facilitated by Joan Ehrhardt, CGC from the Illinois Department of Public Health. The commission held 11 virtual public meetings during 2023 under the leadership of Maria "Ria" Pollock, chair, and Jason Rothstein, vice chair. Following Rothstein's resignation from the commission, Tim Cunniff assisted as interim vice chair; he was confirmed as vice chair in September 2023.

To engage members of the lay public, advocates, health care providers, and industry representatives in attendance more effectively, IRDC identified a series of focus topics of relevance and interest to the commission members and the rare disease community:

1. **Caregiving Challenges (July 2023)**
2. **Access to Genetic Counseling, Care, and Treatments (June 2023)**
3. **Access to Pain Management (September 2023)**
4. **Best Practices and Standards of Care (October 2023)**
5. **False Allegations of Abuse and Neglect (May 2023)**

ILLINOIS
RARE
DISEASE
COMMISSION

Summary

About 1 in 10 people or 1.2 million people in Illinois live with one or more rare diseases. Since 2017, the IRDC has advised the General Assembly on the concerns and needs of individuals and families with rare disease. In 2023, the IRDC held 11 public virtual meetings to hear public comment and find out the needs of individuals and families who live with rare disease.

About 8,000 rare diseases have been identified, but many conditions remain complex, unnamed, rare, and extraordinary (C.U.R.E. conditions).

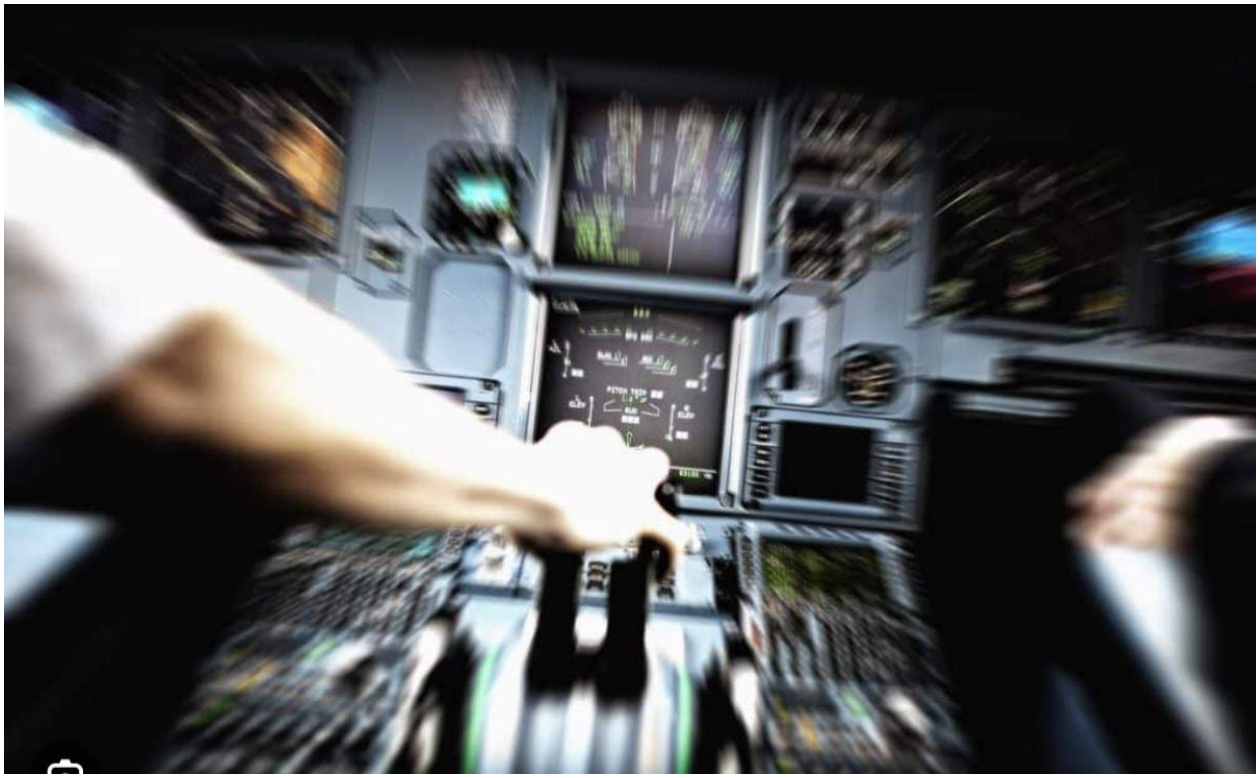
Illinois does not provide sufficient public services to rare families. In Illinois, it is the family's responsibility to meet the care needs of a family member with a rare disease. Family members are asked to give up their job to become the caregiver. Without income and slashed insurance coverage these families are prone to impoverishment, housing insecurity, estrangement, and premature death because of inadequate care and lack of public services.

Some bills that were introduced in the 103rd General Assembly would have removed patients with rare disease from getting diagnostic, curative, and palliative care. Staff and funding are needed to develop concrete solutions to solve the problems of Illinois children and adults with rare disease.

Specialty medications and innovative therapies are often not covered by public and private insurers. Families and individuals are asked to, on average, spend $22,000 more out of pocket than other households (EveryLifeFoundation, 2020). Illinois is cutting off rare disease patients from getting care.

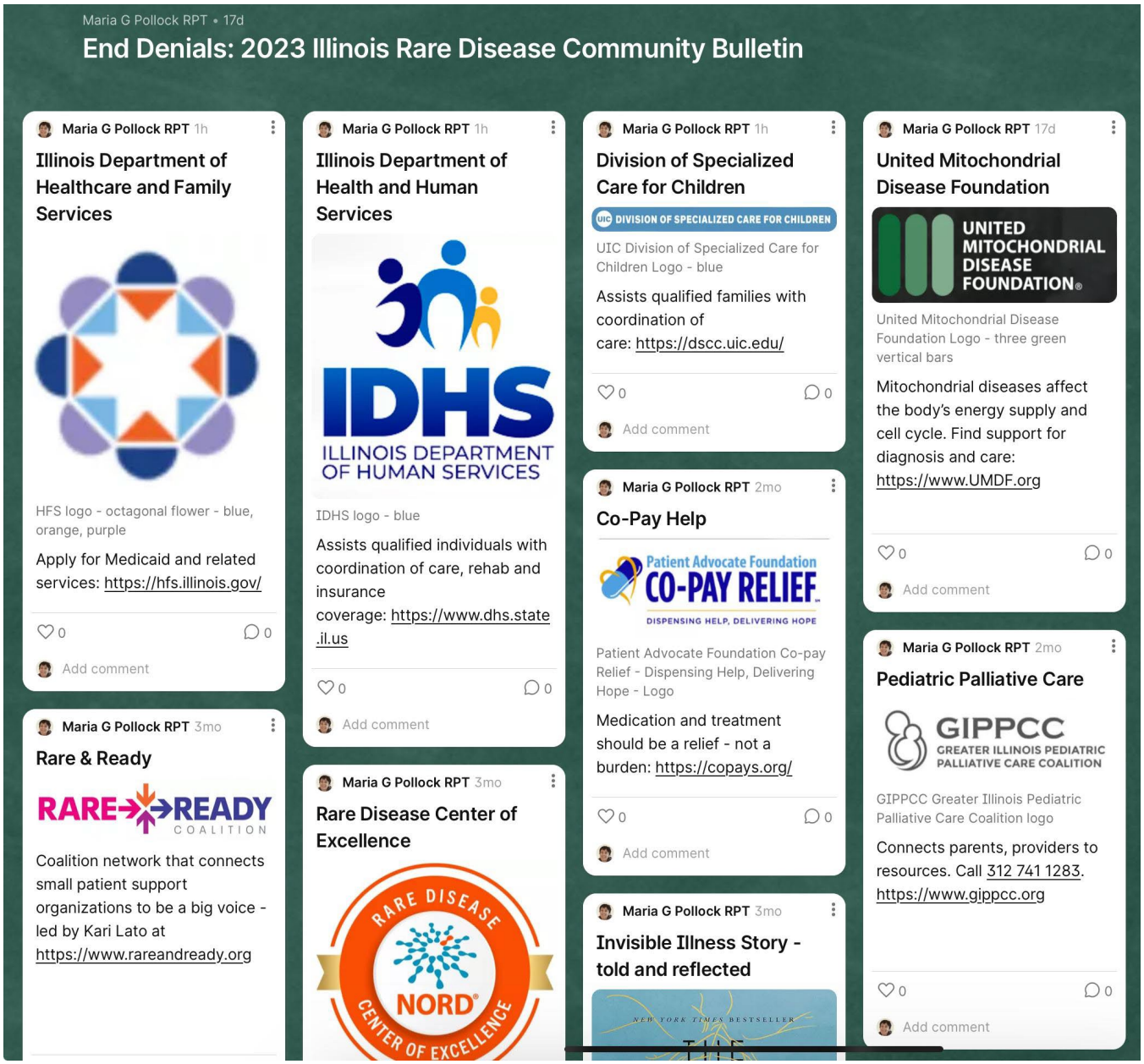
1. Caregiving Challenges for Rare Families

A Fragmented System is Dizzying Illinois Families



“Mothers are the most likely and heavily impacted lifelong care providers. As a result, those women are frequently impoverished and exhausted; left with very little in terms of financial and emotional resources … caregivers, family members, moms, do NOT have the breathing room for self-care,” Lara Pullen, PhD, Minutes, July 2023.

Pullen, who has a doctorate degree, said that as an individual she could not figure out how to find or access services in Illinois on her own. Perhaps she could not locate care giving services in Illinois because they do not exist? Ironically, the Illinois Department of Public Health, Illinois Department of Family Services, Illinois Department of Healthcare and Family Services, and University of Illinois Division of Specialized Care for Children provide resources and care for preventable diseases like heart disease, diabetes, or infectious diseases. But when it comes to patients that have been dealt an unlucky genetic hand by the natural lottery (Marmot, 2015) they are on their own.



Source: EndDenials.org - courtesy of Maria G. Pollock

EndDenials.org provides the first comprehensive website with resources for rare disease patients in Illinois. The Illinois Rare Disease Community Bulletin illustrates how difficult it is to locate and access care for rare disease in Illinois.

> "Additionally, Illinois has issues with funding and staffing in state government agencies. These programs are understaffed, and that makes additional barriers for families to access the services. The additional burden is placed on health care facilities to expand their staff to act as navigators for families. Plus, to go into the community and network. This situation exacerbates inequities in care because facilities receiving care at facilities with fewer resource specialists have less access." Katherine Kim, Minutes, July 2023

Division of Specialized Care for Children: https://dscc.uic.edu/

Illinois Department of Health and Family Services: https://hfs.illinois.gov

Illinois Department of Human Services: https://www.dhs.state.il.us/

Illinois Department of Public Health: https://dph.illinois.gov/

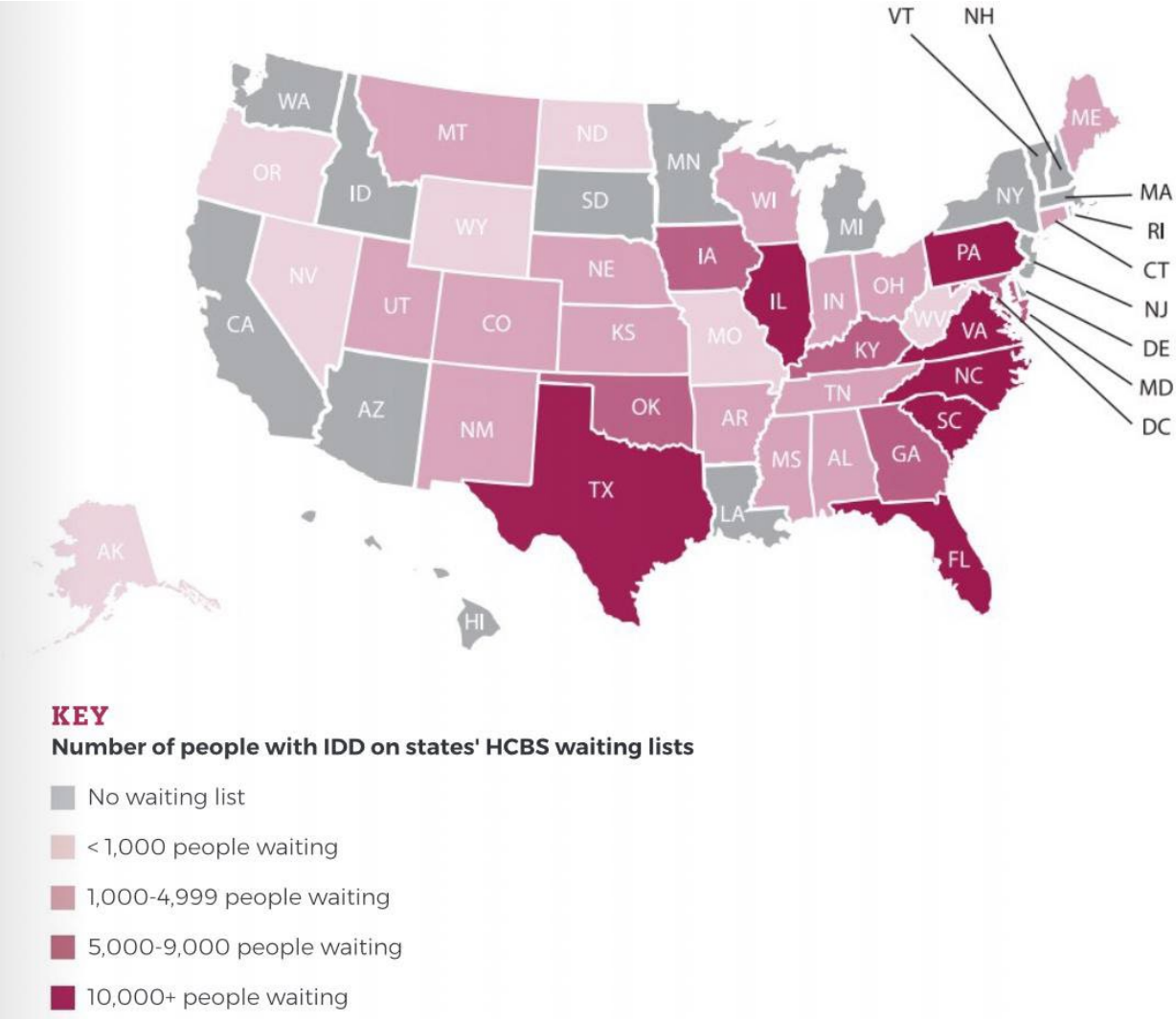
Disorientation and impoverishment of families with rare disease "contributes to the mental health burden," says Stacey Feuer, a psychologist and member of the IRDC.

> "Providing more services is not sufficient. Added resources are needed to address the situation, reduce stress, and lower the burden. There was a wait list of 3,000 for psychiatric and therapeutic services. Now that is down to a few hundred, but wait time is six months. The state is facing an economic and mental health crisis that extends well beyond the rare disease community." (Minutes, July 2023)

Medical providers and genetic counselors like Katherine Kim, MS, CGC, fight this challenge on the front lines.

> "Katherine [Kim, genetic counselor] added that she is not an expert in resources, but she has access at her worksite (Ann & Robert H. Lurie Children's Hospital of Chicago) to social workers and resource specialists. Their facility has needed to increase their resource specialist staff because of the demands on families. Katherine was referred to the Department of Health and Family Services (HFS) and Department of Human Services (DHS) as key providers of assistance to individuals and families with special health needs. DHS does not have a centralized bureau for navigation. Thus, individuals and families are on their own to find out what resources exist and where within the agency they lie." (Minutes, July 2023)

Many family members are asked to leave their job to become their child's caregiver. Highly trained people who could work as professors or doctors have to become caregivers instead. Illinois does not offer sufficient services and the services it has are inadequately staffed.



Illinois ranks in the bottom five states (Arkansas, Texas, Illinois, Mississippi, and Florida) who consistently fall short when providing services for children with disabilities. Why?

> "There is no centralized location to find resources and services. Katherine [Kim] shared that she relies on her site's social work team and Google to find resources. Resources available are also dependent on the individual and family's health care coverage [and the] specific insurer. There is limited access to the Division of Specialized Care for Children for 'extra' coverage. To her knowledge, there is no one site to find the information." Minutes, June 2023

Is there a better model for Illinois to follow?

The best Medicaid service systems for individuals with intellectual and developmental disabilities are found in Arizona, Vermont, New Hampshire, Michigan, and Hawaii, according to the annual Case for Inclusion report released by United Cerebral Palsy (2023). “In Arizona, there is an intentional community developed for individuals with disabilities. Homeowner association fees go toward disability care services. This setting is like a retirement community with a disability focus.” (Minutes, July 2023)

Minnesota has established its own executive branch state agency to address the multi-faceted concerns of people with rare disease (https://mnraredisease.org/who-we-are). Full-time professional staff can research efficient solutions and conduct legal research that get rare families and individuals the support and care they need. They maintain a comprehensive website. The Minnesota Rare Disease Advisory Council has made strides to pass legislation that protects rare disease patients from excess cost. Minnesota Statute 62Q451 guarantees in-network rate for out-of-state specialists for rare disease patients:

> Subd. 2. Unrestricted access. (a) No health plan company may restrict the choice of an enrollee as to where the enrollee receives services from a licensed health care provider related to the diagnosis, monitoring, and treatment of a rare disease or condition, including but not limited to additional restrictions through any prior authorization, preauthorization, prior approval, precertification process, increased fees, or other methods.

In Illinois, families are still left to fend for themselves. Amy’s parents tag-teamed to cover their daughter’s care needs. While Amy was in the hospital when … “these people started calling me in the middle of the night, because they were seeing Amy having a seizure on TikTok.” Testimony Terri V., 2023)

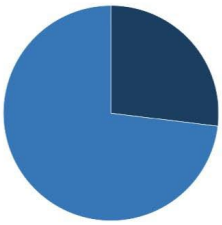
Terri’s daughter has multiple rare conditions and was admitted. But she was not allowed to stay with her overnight. Amy was alone. She talked to friends on TikTok. She felt a seizure coming and pushed the call button. Nobody came for more than 25 minutes.

> “As a patient with rare disease, you must have an advocate caregiver for health care in the hospital, as an outpatient, and in home care. Emergency providers particularly were identified as being doubtful and dismissive of care needs communicated on admission to the health care facility.” (Minutes, July 2023)

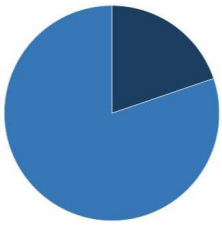
MEDICAID IN ILLINOIS

June 2023

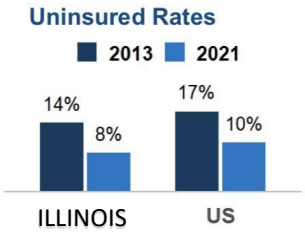
3,817,674 enrolled in ILLINOIS Medicaid



27% of population is low-income (<200% FPL*)

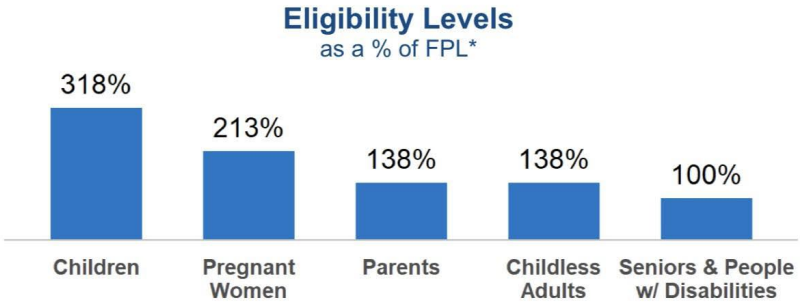


20% of population is covered by Medicaid/CHIP



Expansion Status:
Adopted

Adults in Expansion Group:
1 million



***100% of Federal Poverty Level (FPL):** $24,860 for a family of three; $14,580 for an individual

Medicaid Covers:

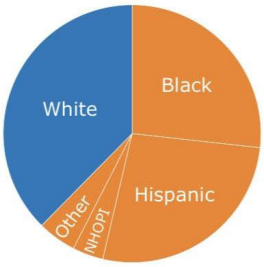

1 in 7 adults ages 19-64


3 in 8 children


2 in 3 nursing home residents

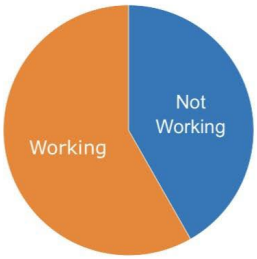

1 in 6 Medicare beneficiaries


1 in 3 people with disabilities



62% of non-elderly Medicaid enrollees are people of color

58% of adults on Medicaid are working



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KFF

Recommendation #1:

Establish and publicize a hotline (e.g., Arizona) and a comprehensive website (e.g., Minnesota) about public services. The hotline should provide access to care, support, and community care giving services like in Arizona. Providers need to know where and how to find help for patients and families.

"Providers need better access and understanding of DHS, the pitfalls to navigating, how to improve access." (Minutes, July 2023)

"Divisions within DHS need to have better collaboration and coordination … Perhaps state agencies such as the Department of Healthcare and Family Services and/or the Department of Human Services develop such a resource [for rare disease families]." Minutes, July 2023

Recommendation #2:

Fund and staff a state agency for the concerns of rare disease patients and their families over the next three years. This agency should administer home and community-based services for the 1.25 million Illinoisans with rare disease.

Accomplishment of the Myers Family

Metachromatic Leukodystrophy (MLD) added to Illinois newborn screening panel.

Public Act 103-0368

Amends the Newborn Metabolic Screening Act. Requires the Department of Public Health to provide all newborns with screening tests for the presence of metachromatic leukodystrophy. Requires the testing to begin within 6 months following the occurrence of specified milestones. Allows the Department to require payment of an additional fee for the provision of metachromatic leukodystrophy screening tests. Contains other provisions.

In the photo … we were talking to the governor. We explained we had to go to numerous specialists and testing in Illinois, all who could not help us. Finally, we went to an emergency room in Madison, Wisconsin to get [her] diagnosis.

…

We are currently working on meeting with a Wisconsin senator to sponsor a bill to try to add MLD to the newborn screening for Wisconsin! Our daughter was born in Wisconsin, which is why newborn screening should be the same in every state!

(Email communication, K. Myers, September 5, 2023)

2. Access to Genetic Counseling and Care

Nearly Four Million Illinoisans Without Access to Genetic Diagnosis, Counseling, and Care

Ann & Robert H. Lurie Children's Hospital of Chicago estimates 72 % of diseases are genetic (2023). More than 3.8 million Illinoisans - nearly 1 of 3 people - receive health care through Medicaid. Yet, Illinois does not recognize genetic counselors as health care providers.

Illinois also does not have enough geneticists that can diagnose patients, order genetic testing, and subsequent genetic counseling. In 2020, the U.S. had a total of 1,240 geneticists nationwide.

> "There are [currently] 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 join the workforce each year. The number of residency training programs has decreased. Many [geneticists] in practice are within a few years of retirement," reports Katherine Kim, MS, CGC. (Minutes, August 2023)

It is more likely that a disease is genetic than the result of a communicable disease or an accident. A geneticist in the U.S. roughly serves 250,000 people (GAO, 2020), or 25,000 patients, estimates Ria Pollock.

By cutting off a third of the population from genetic care we stretch the already slim contingent of geneticists and genetic counselors. Are we providing genetic care according to need? Gatekeeping in this manner violates the principle of fairness and medical necessity.
How can people get timely care if genetic testing and counseling can only be ordered by the few geneticists that are in practice? The wait times for an appointment with a geneticist are frequently measured in years.

> "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 U.S. It is the only genetic counseling training program in Illinois," says Katherine Kim. (Minutes, August 2023)

“If there's time, we'd like to discuss the commission's thoughts on increasing Illinois protections against genetic discrimination as well as potential support from the commission as we work with Medicaid to add genetic counselors as recognized providers.” (Valentine, email communication, June 20, 2023)

Illinois will not be the first choice of any genetic counselor because they have to wait more than four months to receive a license. Even with a license, genetic counselors cannot serve nearly a third of the population, because the Illinois Medicaid program cannot pay them directly.

Besides the workforce shortages of geneticists and genetic counselors, IDPH has acknowledged the benefits of genetic counseling only for special populations for genes associated with rare cancers (Lynch disease and the BRCA2 mutation). The last published Illinois Genetics State Plan is from 2007.

More Red Tape to Genetic Testing

> “Samantha Ropski, MA, a rare disease patient from EDS Chicago, 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).” (Minutes, August 2023)

When we have cleared the red tape to genetic testing and counseling - only then it is fair to enter the moral discussion whether to order testing or not. Otherwise, we will continue to discuss health care equity.

> “Samantha R. wondered ‘what is the point’ of molecular testing? […] Her family has other cases of medical mysteries and relatives with similar issues. Would it help medical providers take her more seriously as a patient? She believes that molecular genetic testing for herself could potentially advance understanding of medicine generally for others with similar health problems, as well as for herself.” (Minutes, August 2023)

Katherine Kim 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 Pollock, rare disease patient, shared that after a surgery in 2015 she learned that she does not respond to most pain medicines. She personally had genetic testing. She has learned since that [the genetic mutations] affect the liver and inactivate many medications." (Minutes, August 2023)

Prior to genetic testing, Pollock was accused of being a drug seeker. She is no longer forced to try inefficient medications and she now has better access to pain management. She has a private health care plan that covered the tests because she had a documented medical need.

> "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." (Kim, Minutes, August 2023)

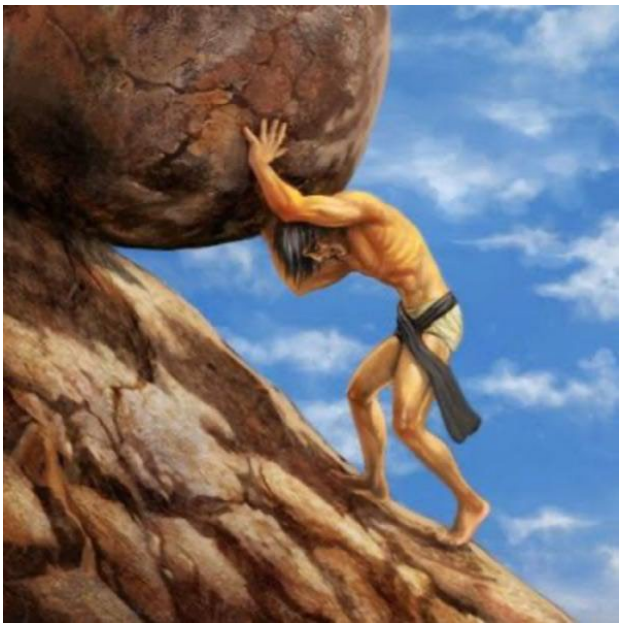
Similarly, the cost of not providing molecular testing is also high. Over the course of three months Pollock was given cocktails of all possible pain medications. She was unable to work and it took years of detoxification. She lives on Social Security Disability Insurance benefits.

> "Alyssa [Valentine], a genetic counselor working at Stroger and a 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." (Minutes, August 2023)

Genetic counseling has a specific CPT code (96040) and is reimbursable by Medicaid/Medicare. But genetic counselors are not recognized as providers by the IMPACT portal. This makes it difficult to deliver services to Illinois patients on Medicaid plans.

Recommendation #3:
Streamline licensing of genetic counselors and other genetic care providers.
Ensure all allied health professions are included in the Impact portal for reimbursement of services provided through public insurance (Indiana and a few other states recognize genetic counselors as providers for people on Medicaid).

3. Access to Pain Management for Rare Disease Patients



Illinois Physicians Leave Rare Disease Patients in Pain

> "Nobody believed us. We thought it might be genetic. Three months after the surgery, a resident suggested genetic testing for altered drug metabolism. I had … several nerve blocks, which lasted only a few hours. This made them think that my body's metabolism was different. The lab test confirmed that my body could not activate most pain medications - not even morphine or fentanyl." (testimony Maria P., September 2023)

It is estimated that about 5% to 12% of the U.S. population have a genetic variation that impacts the efficacy of pain medications. According to Wikipedia, there are 6% to10% of Caucasians who are genetically resistant to pain drugs. African Americans have an even higher incidence of being poor metabolizers of pain drugs. Gaslighting them - disbelieving these pain patients - is just one of the responses that ignores their need for more effective pain care.

Lack of Availability and Experience

> "In the sickle cell community access to ketamine can be a challenge, particularly for children. Some institutions, some providers do not have experience administering it. Similarly for lidocaine, reports TaLana Hughes." (Minutes, September 2023)

> "Nic does get ketamine infusions when they are available. The problem is less insurance and more that hospitals have had trouble getting ketamine this year. Supply has been the barrier, along with the discomfort of some physicians and anesthesia teams with using ketamine on children (not at Comer, at Edward). He has never had a lidocaine infusion." (testimony Amy, September 2023)

If ketamine and lidocaine injections are not known to providers, then provider education needs to be a focus.

Accusations of Drug Seeking and Doctor Shopping

> "Becky Fleming, a U.S. Pain Foundation support group facilitator, added her experience with challenges accessing medication. If the identified pain provider or pain center is uncomfortable offering certain treatments, the individual patient is viewed as 'provider shopping and medication seeking' when they need to change providers to access additional or different therapies. Form changes have made it more challenging also. Some providers are not comfortable submitting information needed to authorize treatment, e.g., medical marijuana. There are many hoops, including quarterly preauthorization." (Minutes, September 2023)

Insurance Coverage, Prior Authorizations, Individualized Drug Metabolism

> "Many patients seeking care at [Lurie's] voice similar concerns about the challenges of obtaining insurance coverage and prior authorizations… People experience pain differently and respond to medications individually. Their center has begun to focus on individual profiles that include some pharmacogenomic information as well as educating emergency room providers about the condition(s). They are beginning to use more than the pain scale to gauge, monitor, and manage pain." (Meredith, Minutes, September 2023)

Ketamine is not an opioid. Ketamine is a narcotic and has a long history as a pediatric anesthetic. It is on the World Health Organization's list of essential medications. So how can it not be routine to stock it in Illinois?

Drug Shortages?

> "Generic medications are not seen as money makers. Due to low overhead, manufacturers here in the U.S. do not 'oversupply' the market. Because of quality concerns regarding medications manufactured overseas, there is some return of manufacturing to the U.S. There is a chronic shortage of IV drugs. (See also https://www.accessdata.fda.gov/scripts/drugshortages/default.cfm). Margins are higher for non-generic prescription drugs." (Tim Cunniff, Minutes, September 2023)

If there are no market incentives to stock ketamine and lidocaine, the market fails rare pain patients. Ketamine and lidocaine are not expensive drugs, they have a small or no

profit margin. Is a small profit more important than treating someone who is in severe pain?

Costs of Losing Access to Pain Medication and Maintenance Drugs

The IRDC discussed how losing access to maintenance and pain medications affects patients, families, providers, and state agencies:

Patient	Family	Providers	State Agencies
worse symptoms, heightened anxiety	increased caregiving needs	more patients with increased symptoms	coverage of potentially avoidable/under-supported disabilities
progression from acute to chronic stage	loss of productivity [need to reduce work to provide caregiving]	productivity loss	coverage of medical care for acute and increased chronic symptoms
reduced function		administrative overload	[overload]

> "I was unable to regain my prior baseline of health and function despite care and therapy." (Patient testimony, Samantha R., Minutes, September 2023)

> "This limits the access to providers because of the increased amount of time taken to manage rare disease and all chronic conditions." (Lara P., Minutes, September 2023)

Meredith suggested that primary targets for provider education would be, for example, emergency medicine, pain management providers, family medicine, internal medicine, and anesthesiologists. This would help increase familiarity and remove stigma.

> "It should not be a privilege to have adequate pain management." (Maria P., Minutes September 2023)

Hughes said that any opportunity for education for the prescriber, staff who perform infusions, and others could be helpful. Patients can be seen and portrayed as medication seeking. Removing stigma and increasing empathy among all staff may help people needing care.

Recommendation #4:

Require three CMEs (continuing medical education) for physicians and pharmacists about genetically altered drug metabolism every three years.

Recommendation #5:

Require three CMEs for physicians (emergency physicians, pain medicine, in infusion therapy protocols of ketamine/lidocaine) every three years.

Recommendation #6:

Require three in-person CMEs for physicians and nurses in rare disease awareness, including anti-bias and empathy training every six years.

Recommendation #7:

Require Illinois hospital pharmacies, emergency departments, pain clinics, home-care services, and urgent care centers to stock a mandatory minimum supply of ketamine, lidocaine, and infusion supplies.

4. Best Practices and Standards of Care in the Context of Rare Disease?

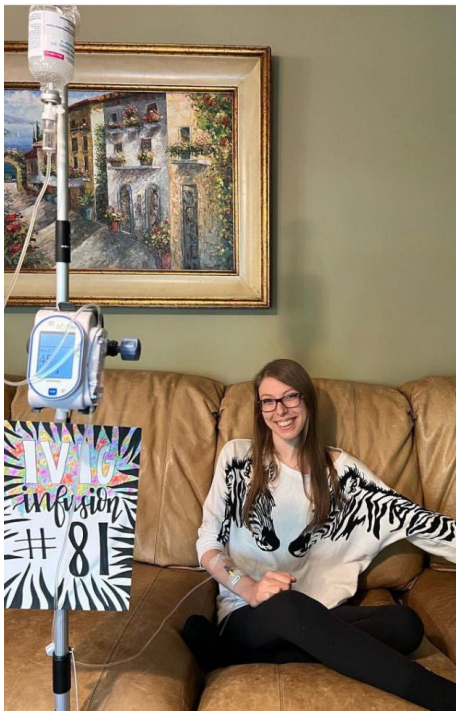
Pathways to Palliative Care for Rare Disease Patients and Their Providers

"… I have a 14-year-old very complex EDS (Ehlers-Danlos-syndrome) kid in Mt. Morris Illinois whose doctor FINALLY has decided he wants to find her palliative care."

[…She is a] Single mom so I am assuming Medicaid. As we all know, finding palliative care that understands EDS and takes under 18 is a big challenge. Maria... do you have any ideas???" (Email conversation with Donna Sullivan, October 5, 2023)

"It sounds difficult to 'qualify' patients for palliative care. There are many uncertainties about the levels of care and how to access care. What is substandard care? What is meant by palliative home care [and who can receive it]." (Minutes, October 2023)

Patients and providers frequently confuse hospice care with palliative care. Palliative care is comfort care. Palliative care can be offered to anyone at any time. People must know how to ask for it. Stacey F., IRDC member, added that education is needed. A campaign perhaps targeted at both health professionals and the lay public. Providers need to know to offer it.



Lara suggested that families be informed to request a palliative care provider to be part of the multidisciplinary health care team. A directory of providers may help facilitate access.

"Palliative care is not available to me because of my condition; chronic pain, is not recognized or treated by the system, asserted Karin P. (Minutes October 2023)

The extent of palliative care and where it can be provided may depend on insurers and care providers. Samantha R., rare disease patient, receives her infusion care at home (photo). Palliative care is often hospital based. There are outpatient palliative medicine providers. However, home-based care requires insurance approval. Community-based/outpatient care might be a better in between option versus inpatient or in hospital care. Does Illinois provide adequate palliative care?

Traveling to get to care is another barrier, e.g., for individuals too sick to travel. Many patients, like Karin P. would benefit from home visits. She is vision-impaired and lives with chronic pain. Maria P. has become less homebound. She is able to get outpatient care with a portable IV pump and a portable oxygen concentrator (photo).

"What would be the solution if we wanted to spread and establish outpatient palliative care across communities statewide?" asked Lara P., IRDC member. (Minutes, October 2023)

Some states surveyed individuals in the rare disease community about unmet needs. That strategy could provide more information to inform models and activities to improve access.

Recommendation #8:

Publish, distribute, and maintain a directory of Illinois palliative home-care providers.

Recommendation #9:

Fund and distribute a statewide survey to all patients and providers to identify barriers and gaps in access to care for rare disease patients.

Recommendation #10:

Mandate insurance coverage for palliative care in hospitals, outpatient settings, and homecare for patients with complex, unnamed, rare, and extraordinary conditions.

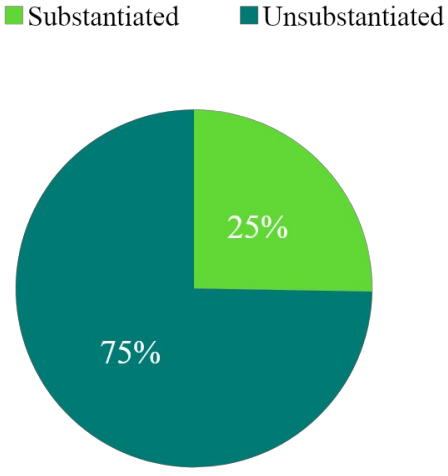
5. False Allegations of Child Abuse and Neglect

Not Every Bruise is Abuse

In April 2023, Ann & Robert H. Lurie Children's Hospital of Chicago emergency physician Dr. Mary Clyde Pierce and Senior Research Scientist Kim Kaczor published an application that helps to determine whether child injuries were caused by abuse or neglect. The LCAST app (Lurie Children's Child Injury Plausibility Assessment Support Tool) is based on an National Institutes of Health study (https://jamanetwork.com/journals/jamanetworkopen/fullarticle/2778559), which was co-authored in 2021 by Clyde Pierce, Kaczor, Douglas and others. The application claims 95% sensitivity and 87% specificity (https://www.luriechildrens.org/en/news-stories/ lurie-childrens- hospital-launches-app-to-help-screen-bruises-in-young-children- for-potential-abuse/).

The Ehlers-Danlos syndromes (EDS) community in Chicago and the Family Justice Resource Center (FJRC) were alarmed by the application. Maria Pollock, an EDS patient, tested the application with the bruises of her own children. If the emergency physician failed to recognize EDS, the application would identify her as an abuser rather than a mother with connective tissue disorder.

In Illinois, more than 141,241 children were investigated as possible victims of child abuse or neglect in 2021 (Annie E. Casey Foundation, 2023). It turns out that more than 105,528 of these children were not maltreated.



Source: Annie E. Casey Foundation, 2023

Taking Care of Maya (Netflix, 2023), the Florida case of Maya Kowalski, illustrates how the trauma of false allegations of child abuse causes irreparable damage to families. Maya's mother committed suicide. A settlement of $261 million cannot bring this mother back.

The American Medical Association's code of ethics requires its members to "study and prevent error and harm." Child abuse pediatricians need to be able to rule out rare disease before causing error and harm to families.

The protection of children is paramount, but if 75 families suffer trauma from false allegations to identify 25 cases of abuse it is time to pause. Why is the number of unfounded accusations so high? The current profiling of families at increased risk for child abuse and neglect (NCANDS, 2021) is not evidence-based. Similar to drug abuse, the suspicion of child abuse and neglect (CAN) is based on stereotypes and stigma. How do we rule out abuse?

The Royal Children's Hospital in Melbourne, Australia provides a quiz for its providers about "Skin Conditions that Mimic Child Abuse" (https://www.rch.org.au/uploadedFiles/Main/Content/vfpms/ 8_Ciara_RCH temp skin conditions_ni.pdf). Would every Illinois child abuse specialist pass this quiz?

One in 500 people (Demmler, 2019) is living with a connective tissue disorder. We cannot afford to overlook this incidence because false accusations harm families and their children. The Illinois Department of Children and Family Services has contracts with child abuse specialists. It lists over 34 child abuse specialists, but Illinois offers only one residency for genetics and currently has one provider for connective tissue disorders.

The first Illinois Center for Rare Disease was established at Northwestern University's Ann & Robert H Lurie Children's Hospital in Chicago in May 2023.

We need to raise awareness of diseases that cause excessive bruising. By implementing mandatory screening for conditions that mimic child abuse as part of CAN screening, the numbers of false accusations of child neglect and abuse will be improved. Many families will be spared the trauma of a false accusation.

Recommendation #11:

Illinois Department of Children and Family Services should amend the training for mandatory reporters of Child Abuse and Neglect (https://mr.dcfstraining.org/UserAuth/Login!loginPage.action):

Excessive bruising can be caused by health conditions that mimic child abuse, like mastocytosis, Ehlers-Danlos-Syndromes, bleeding disorders and other rare conditions (and include a picture of EDS labeled: This is not child abuse).

Recommendation #12:

Require all contracted child abuse specialists to pass an annual screening that they are able to recognize and to identify health conditions that mimic child abuse.

Because of its high incidence of 1 in 500 people, connective tissue disorders need to be ruled out first, before accusations of child abuse and neglect (CAN) may proceed. Child abuse specialists must follow the recommended diagnostic regimen (nosology) for connective tissue disorders. To rule out EDS, the physician must take a family history and conduct a Beighton score evaluation (hypermobility test) of the birth parents.

6. Online Resource

The IRDC chair procured a website that provides resources for people with rare disease in Illinois. The website lists advocacy organizations, state departments, and events that are relevant for patients with rare disease, their families, and their health care providers. The website also announces research opportunities and pilots. The site is currently maintained by M. G. Pollock.

Recommendation #13:

Until IDPH can publish a web resource for rare disease patients and providers in Illinois, we ask IDPH to post a prominent link to the Illinois Rare Disease Resources Bulletin Board at https://www.enddenials.org.

7. Open Meetings Act

The Illinois Open Meetings Act requires boards and commissions to meet in person to do business. The majority of the IRDC are patients with disabilities and family members with caregiving duty or health care providers. It is not reasonable to ask these volunteers to travel for IRDC meetings - many of whom are exempt from attending in person.

Recommendation #14:

Continue to allow the IRDC to conduct their meetings and do business in public virtual meetings.

8. Reduce Paperwork

Several members of the commission had to file for reappointment within less than three months because they were finishing the unexpired terms of previous members. Newly appointed members should start serving a full term and finish unexpired terms. This alleviates the administrative burden and the burden on the volunteers who serve on the IRDC.

Recommendation #15:

The IRDC recommends amending the Illinois Rare Disease Commission Act as follows:

410 ILCS 445, Section 10 (f)

(f) Vacancies in commission membership shall be filled in the same manner as initial appointments. ~~Appointments to fill vacancies occurring before the expiration of a term shall be for the remainder of the unexpired term.~~

9. Realistic Mission

The mission of the IRDC is to make recommendations to the General Assembly about:

(1) Prescription drugs and innovative therapies
(2) Legislation to improve care and treatment
(3) Expanding and improving newborn screening

Recommendation #16:

The IRDC's mission should be amended to include:

(1) Public services.
(2) ~~(1)~~ Prescription drugs and innovative therapies
(3) ~~(2)~~ Legislation to improve care and treatment
(4) ~~(3)~~ Expanding and improving newborn screening and genomic care.

Conclusion

The findings in this report show that there is a dire need for more investment in services, resources, and legislative mandates to provide support and to care for people with rare disease.

Some of the causes of health inequity can be addressed by additional training of health care professionals. Other deficits will need to be addressed by staffing agencies and establishing additional services for people with rare diseases and disabilities.

The experiences of people with rare disease suggest that legislative mandates are needed to end the discrimination of people with rare disease in Illinois.

The IRDC is calling on the public health state agencies and the Illinois General Assembly to take prompt action to craft policies and budgets that recognize rare disease as a public cause. We need funding to create resources and a helpline to access these services for rare disease patients and their health care providers.

Families deserve support before they are destitute or homeless. Nearly half of the population in Illinois receives public health insurance through Medicaid and Medicare. Rare disease is a complex challenge that requires the support of many state agencies. By directing rare disease patients to appropriate care, we end their odyssey for diagnosis and treatment.

This will position the state as a leader and example for the nation on advancing care for people with rare diseases.

APPENDIX

Appendix A - Charge of the Illinois Rare Disease Commission Act of 2017

LEGISLATIVE MANDATE

The Illinois Rare Disease Commission was established by the Rare Disease Commission Act (410 ILCS 445) to advise the state on issues pertaining to the care and treatment of individuals with rare diseases. Pursuant to 410 ILCS 445, the commission makes recommendations to the General Assembly in the form of an annual report. Commission activities extend through 2026, pursuant to Public Act 102-0671 (Section 75).

Section 410 ILCS 445/15 - [Section scheduled to be repealed 1/1/2027] Study; recommendations
The Commission shall make recommendations to the General Assembly, in the form of an annual report through 2026, regarding:

(1) the use of prescription drugs and innovative therapies for children and adults with rare diseases, and specific subpopulations of children or adults with rare diseases, as appropriate, together with recommendations on the ways in which this information should be used in specific State programs that (A) provide assistance or health care coverage to individuals with rare diseases or broader populations that include individuals with rare diseases, or (B) have responsibilities associated with promoting the quality of care for individuals with rare diseases or broader populations that include individuals with rare diseases;
(2) legislation that could improve the care and treatment of adults or children with rare diseases;
(3) in coordination with the Genetic and Metabolic Diseases Advisory Committee, the screening of newborn children for the presence of genetic disorders; and
(4) any other issues the Commission considers appropriate.
The Commission shall submit its annual report to the General Assembly no later than December 31 of each year.

Appendix B - Required Trainings

Commission members were asked to complete the following trainings:

Appointed members of a public board, commission, or task force (collectively, board) are generally required to participate in six trainings.

1. The Open Meetings Act (OMA)
2. Ethics Training for State Employees and Appointees
3. Harassment and Discrimination Prevention Training

4. Security Awareness Training
5. HIPAA and Privacy Training (Only Quality Care Board and Mortality Review Board members are required to attend this training, but all appointees are encouraged to attend)
6. Diversity, Equity, and Inclusion Training

Appendix C - IRDC Recommendations to the 103rd GA

Recommendation # 1: IDPH, DHS, HHS - interagency effort
Immediately: Fund, establish, and publicize a hotline (e.g., Arizona) and a comprehensive website (e.g., Minnesota) about public services. The hotline should provide access to care, support, and community care giving services like in Arizona. Providers need to know where and how to find help for patients and families.

Recommendation #2:
Long-term: Fund and staff a state agency for the concerns of rare disease patients and their families over the next three years. This agency should administer home- and community-based services for the 1.25 million Illinoisans with rare disease - not just children with disabilities.

Recommendation #3: Illinois Department of Financial and Professional Regulations (IDFPR)
Streamline licensing of genetic counselors and other genetic care providers. Ensure all allied health professions are included in the Impact portal for reimbursement of services provided through public insurance (similar to Indiana).

Recommendation #4: (IDFPR)
Require three CMEs for physicians and pharmacists about genetically altered drug metabolism every three years.

Recommendation #5: (IDFPR)
Require three CMEs for physicians (emergency physicians, pain medicine, in infusion therapy protocols of ketamine/lidocaine) every three years.

Recommendation #6: (IDFPR)
Require three in-person CMEs for physicians and nurses in rare disease awareness, including anti-bias and empathy training every six years.

Recommendation #7: IDPH
Require Illinois hospital pharmacies, emergency departments, pain clinics, home-care services, and urgent care centers to stock a mandatory minimum supply of ketamine, lidocaine, and infusion supplies.

Recommendation #8: IDPH
Publish, distribute, and maintain a directory of palliative home-care providers in Illinois.

Recommendation #9:
Fund and distribute a statewide survey to all patients and providers to identify barriers and gaps in access to care for rare disease patients.

Recommendation #10:
Mandate insurance coverage for palliative care in hospitals, outpatient settings, and home-care for patients with complex, unnamed, rare, and extraordinary conditions.

Recommendation #11: DCFS
Amend the training for mandatory reporters of Child Abuse and Neglect (https://mr.dcfstraining.org/UserAuth/Login!loginPage.action):

> Excessive bruising can be caused by health conditions that mimic child abuse, like mastocytosis, Ehlers-Danlos-syndromes, bleeding disorders, and other rare conditions (and include a picture of EDS labeled: This is not child abuse).

Recommendation #12: IDFPR, IDPH, DCFS
Require all contracted child abuse specialists to pass an annual screening that they are able to recognize and to identify health conditions that mimic child abuse.

Recommendation #13: IDPH
Until IDPH can publish a web resource for rare disease patients and providers in Illinois, we ask IDPH to post a prominent link to the Illinois Rare Disease Resources Bulletin Board at https://www.enddenials.org.

Recommendation #14:
Continue to allow the IRDC to conduct their meetings and do business in public virtual meetings.

Recommendation #15:
Amend the Illinois Rare Disease Commission Act as follows:

> 410 ILCS 445, Section 10 (f)
>
> (f) Vacancies in commission membership shall be filled in the same manner as initial appointments. ~~Appointments to fill vacancies occurring before the expiration of a term shall be for the remainder of the unexpired term.~~

Recommendation #16:
The IRDC's mission should be amended to include:

(1) Public services.
(2) ~~(1)~~ Prescription drugs and innovative therapies
(3) ~~(2)~~ Legislation to improve care and treatment
(4) ~~(3)~~ Expand and improve newborn screening and genomic care.

To view upcoming meetings and agendas, visit:
https://dph.illinois.gov/resource-center/advisory-boards/illinois-rare-diseases-commission.html

For more information, contact the IDPH facilitator, Joan V. Ehrhardt at joan.ehrhardt@illinois.gov, or the IRDC Chair Maria G. Pollock at mariapollock67@gmail.com.

Meetings abide by the Illinois Open Meetings Act.