CORD Webinar 2 – July 14, 2026

The Cost of Waiting

Socioeconomic impact of rare disease in Canada

IHE/CORD Survey provides evidence of cost.
Moments that Matter personal stories show who really pays when the system is not ready.

Core message

The cost of waiting is paid first by families — in time, income, energy and hope — and later by health systems through crisis care and avoidable burden.

Source: IHE Technical Report, June 2026; CORD Webinar Series framing.

Two personal stories.

A national pattern.

ALICE • ONTARIO

Wilson disease

The cost when diagnosis comes after the treatment window

Missed early detection → emergency transplant → crisis-level system cost and family trauma.

Live Q&A: what should have been in place before crisis care?

KIM • NEW BRUNSWICK

CVID

The lifetime cost when diagnosis comes too late

Delayed diagnosis → disability, lost income security, housing insecurity and unresolved harm.

Live Q&A: what would earlier recognition and support have changed?

Their experiences are personal. The pattern is national. The costs are measurable.

Sources: Moments That Matter responses for Alice/Wilson and Kim/CVID

Alice: When waiting becomes crisis care

A treatable rare disease found only after the family and the system were already in crisis.

Missed Warning Signs

Acute system cost

A young adult who seemed healthy until Wilson disease presented as end-stage liver disease requiring emergency transplant.

Readiness lesson

Early detection, clinical suspicion and screening can prevent crisis care for some rare diseases.

1. What was your daughter's life like before everything changed?
2. What was the moment you knew this could not wait?
3. What do you wish had been in place earlier?
4. What worked once the diagnosis was made and she reached the transplant team?
5. What should policymakers remember about screening, early diagnosis and treatment windows?

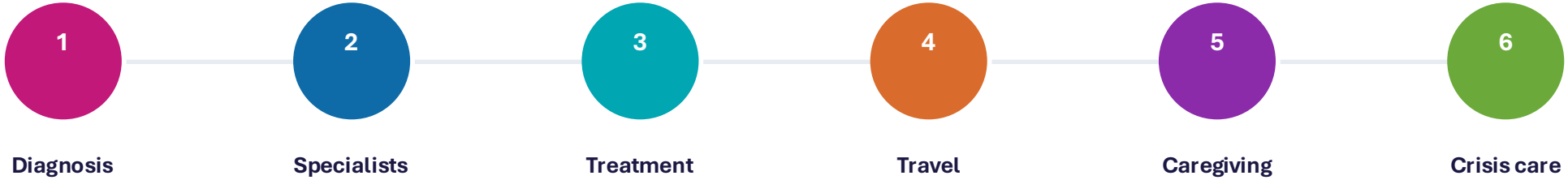
Lesson: "Alice's story shows one of the most expensive forms of unreadiness: when a treatable condition is not identified until the family and the system face crisis care."

Story source: Moments That Matter response, Alice, Wilson disease, Ontario.

From two lives to a national pattern

BRIDGE

Alice and Kim help us feel the cost. The survey helps us see how often and where it appears.



What data show Quantify the burden and show where costs are borne.

What stories show Show who carries the burden and what readiness would have changed.

One headline. One human callout. One readiness lesson.

Source: CORD Webinar Series framing; IHE Technical Report domains; Moments That Matter story set.

What the IHE/CORD survey makes visible

SURVEY

The 2025 survey gives Canada a clearer picture of costs that families often carry quietly.

205
rare disease survey respondents

141
persons living with rare disease

64
caregiver respondents

50+
conditions represented

2025
costs in CAD

Healthcare use
hospital, ED, family physician and specialist visits

Caregiving
daily unpaid support and illustrative annual value

Out-of-pocket costs
care-related and living-with-RD household spending

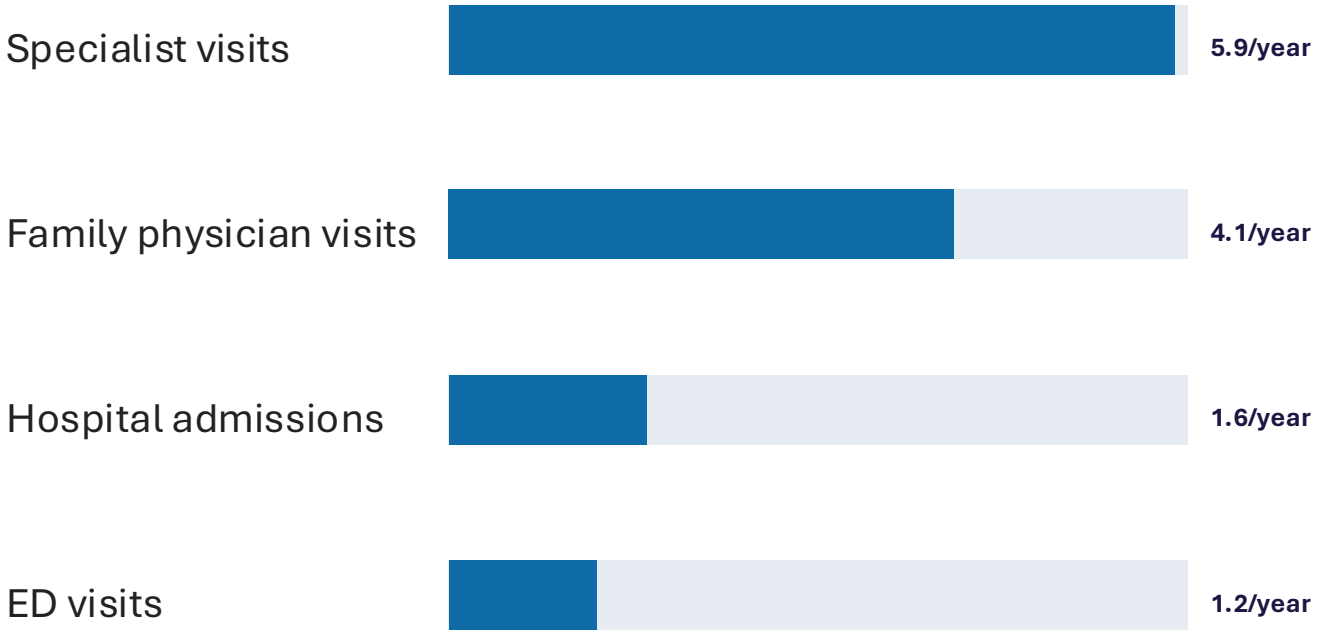
Diagnosis + treatment
diagnostic delay, misdiagnosis and drug access barriers

Travel burden
distance, travel time, transport and accommodation

Source: IHE Technical Report, Executive Summary and Methods, June 2026.

Rare disease care is a marathon of appointments

Behind every "average" is a calendar full of appointments, referrals, tests, emergencies and follow-up.



Numbers = Costs

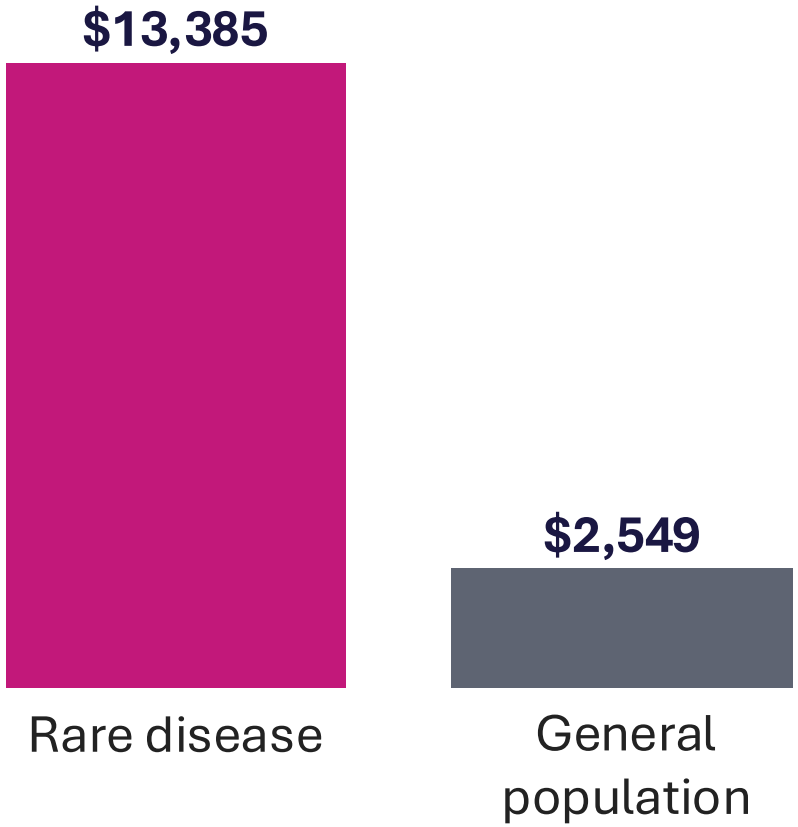
The health system bears rare disease costs through "repeated" encounters, specialist use and crisis care.

Source: IHE Technical Report, Figure 2 and Table 6.

Crisis care is expensive for everyone

DATA

When systems miss the early moment, the health system may meet the patient later in hospital.



5.3x

higher mean annual hospital admission cost

$440

mean annual ED cost

$497

mean annual specialist visit cost

Source: IHE Technical Report, Tables 7 and 8. Alice story source: Moments That Matter response.

The diagnostic odyssey is not just time — it is cost

Every year without answers can add appointments, expenses, uncertainty and lost options.

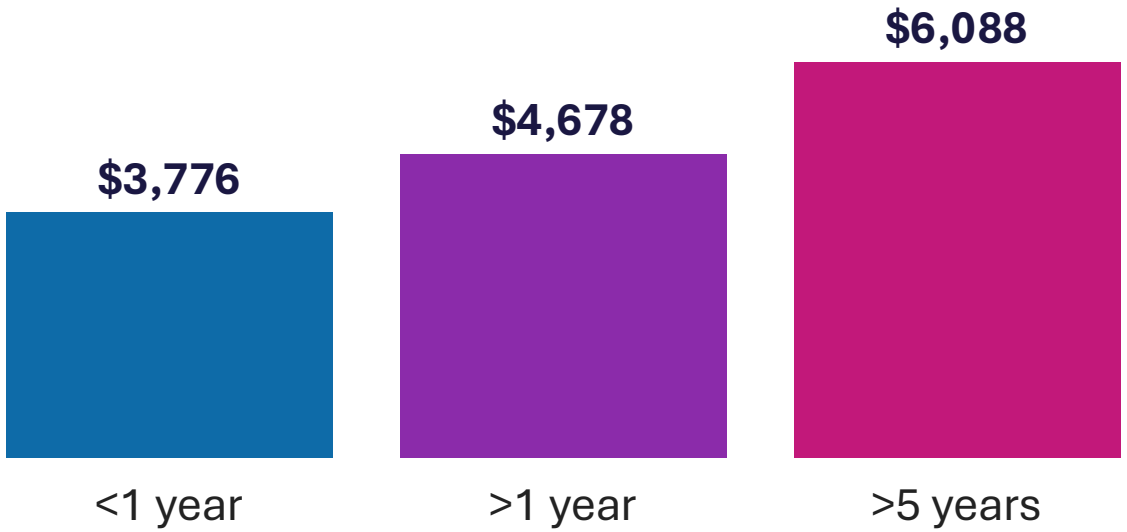
66.8%
waited more than 1 year for diagnosis

30.9%
waited more than 5 years

44.4%
reported at least one misdiagnosis

6.3 + 6.6
mean GP + specialist visits before diagnosis

Mean annual care-related OOP by diagnostic timeline



Late diagnosis can become disability, income loss, housing insecurity and lost independence.

Source: IHE Technical Report, Figure 5 and Tables 15-16.

The "bill" does not stop at the hospital door

Public coverage does not erase the private costs of living with a rare disease.

$4,449

mean annual care-related out-of-pocket costs

Health services • prescriptions • support services • equipment • other care costs

$6,301

mean annual living-with-RD out-of-pocket costs

Home needs • hiring help • medical food • mental health • special education • daily living costs

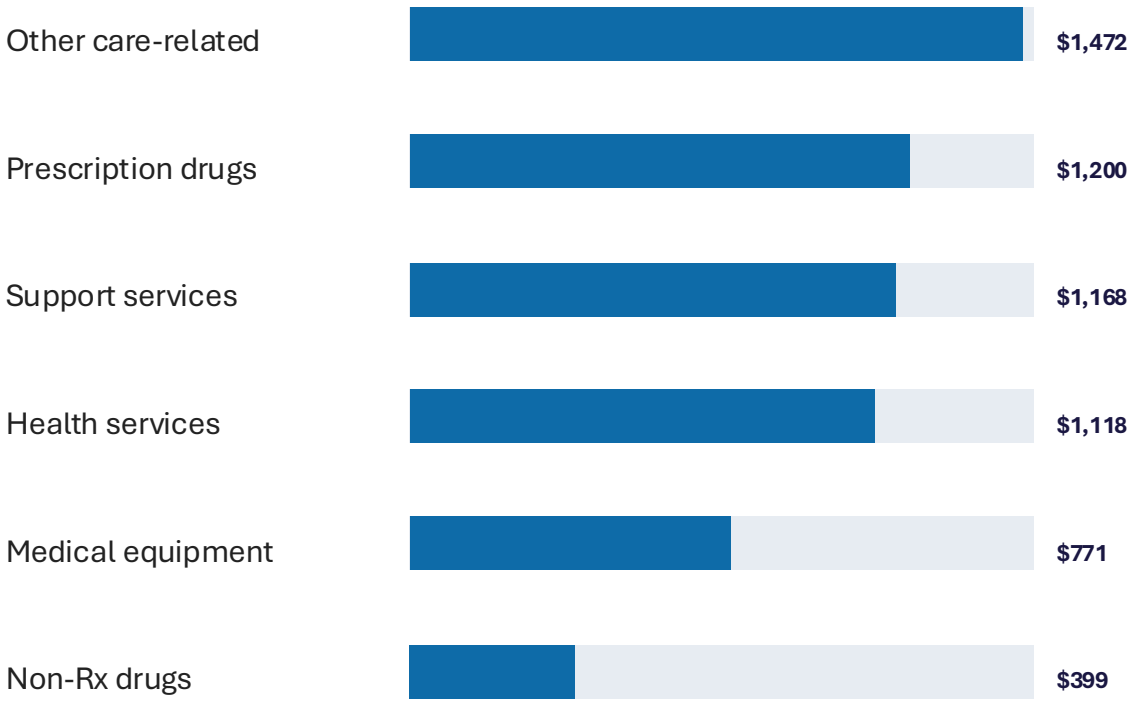
Policy implication: rare disease value assessment should count household and caregiver burden, not only drug or hospital costs.

Source: IHE Technical Report, Executive Summary and Tables 10, 11, D11 and D12.

When treatment is food, policy matters

DATA + VIGNETTE

Sandra’s PKU story shows why rare disease costs do not always look like a prescription claim.



SANDRA • ALBERTA • Phenylketonuria (PKU)

When treatment is food: financial supports matter

Sandra is a parent/caregiver whose child was diagnosed through newborn screening. Even with early diagnosis, the cost of low-protein food created major financial strain. After two Disability Tax Credit denials, she prepared her own tax court case with CanPKU support. A favourable ruling brought 10 years of back pay and helped other PKU families access DTC.

$1,036

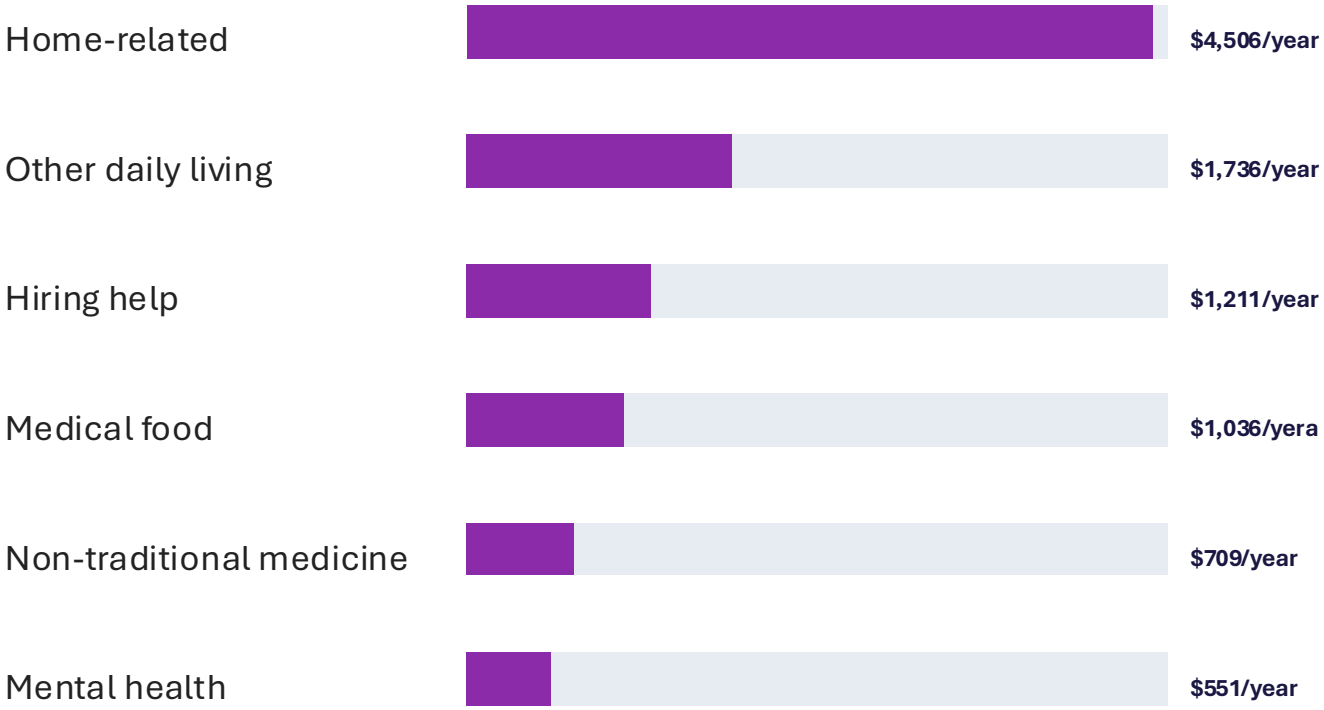
mean annual medical food cost (IHE/CORD survey)

Source: IHE Technical Report, Tables 10-11. Story source: Moments That Matter response, Sandra / Alberta / Phenylketonuria (PKU).

Daily life has a price tag too

DATA

Rare disease costs show up in kitchens, classrooms, bedrooms, vehicles and monthly bills.



$150,700

maximum reported living-with-RD OOP

Maximum values show variability — not typical costs. Means and SDs show large heterogeneity in cost impact.

Source: IHE Technical Report, Table 11.

Distance turns care into a lifelong journey

DATA + VIGNETTE

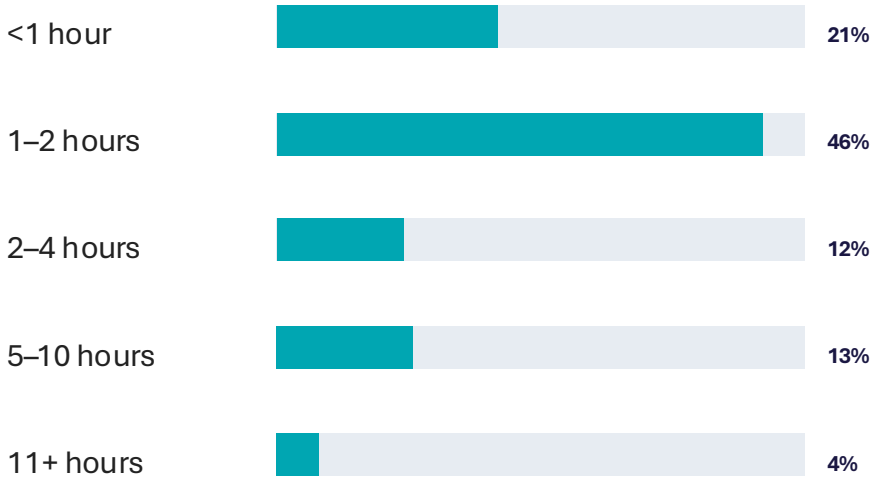
For some families, getting care means leaving home, paying privately and becoming system navigator.

58%
reported transportation costs

31%
reported accommodation costs

35%
travelled more than 2 hours one-way to specialist care

54%
of those with accommodation costs reported >$1,000



MONICA • NOVA SCOTIA • DeSanto-Shinawi Syndrome (DESSH)

Diagnosis did not mean systems were ready.

Monica is the parent of a child with DeSanto-Shinawi Syndrome (DESSH), an ultra-rare congenital/neurodevelopmental condition. After early developmental concerns and a 5–10 year diagnostic journey, the family paid out of pocket for comprehensive genetic testing and evaluations in Mexico, where DESSH was diagnosed. There they also accessed integrated care — genetics, pediatric neurology, therapies, therapeutic riding and specialized school support — showing how travel, private costs and care coordination gaps converge.

Source: IHE Technical Report, Table 12, Figure 3 and Table D13. Story source: Moments That Matter response, Monica / Nova Scotia / DESSH.

Where you live can shape what you pay

DATA

Access is not equal when the right specialist, therapy or support is far away or unavailable.

Factor	Group	Care-related OOP average	Living-with-RD OOP average
Distance to specialist	≤50 km	**$3,104**	**$7,424**
Distance to specialist	>50 km	**$5,960**	**$7,182**
Travel time	<1 hour	**$2,643**	**$1,466**
Travel time	≥1 hour	**$5,098**	**$9,789**
Drug therapy access	No	**$7,055**	**$4,589**
Drug therapy access	Yes	**$3,711**	**$6,639**

1.9x care-related OOP when >50 km from specialist vs ≤50 km • 6.7x living-with-RD OOP when travel ≥1 hour vs <1 hour

Source: IHE Technical Report, Tables 13 and 20. Comparisons are descriptive, not causal.

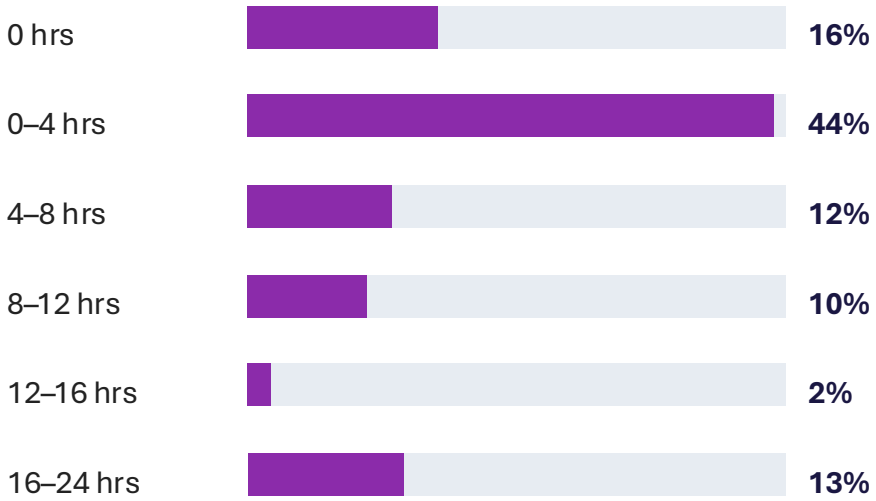
Families are the hidden workforce

Dawn's story also shows when services are missing — families absorb cost of care.

37.2%
primary caregivers provide more than 4 hours/day

13.2%
provide 16–24 hours/day

$187K
upper illustrative annualized value for high-intensity caregiving



DAWN • NEW BRUNSWICK • Dravet syndrome

When support does not grow with need

"No family should have to become the healthcare system."

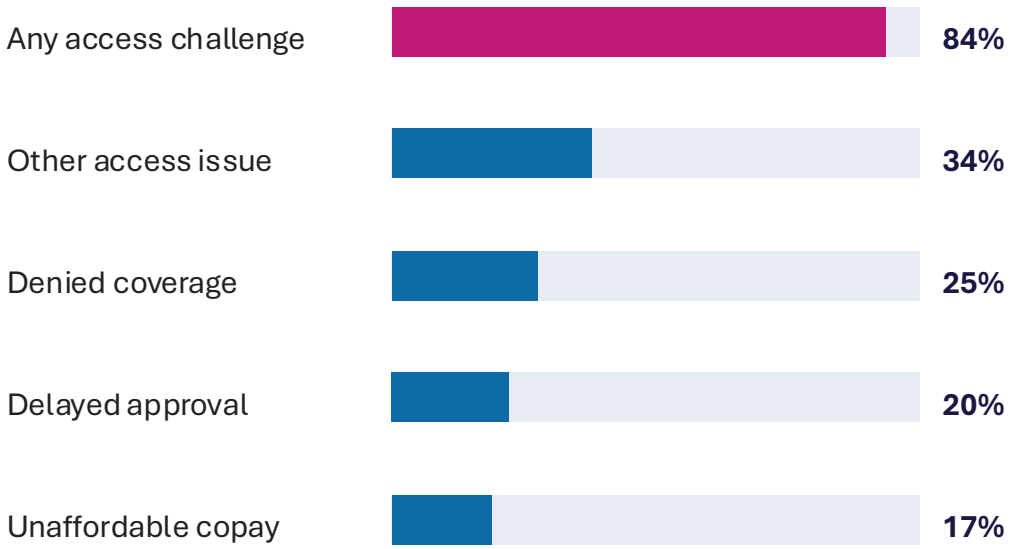
Dawn is the parent/caregiver of a son with Dravet syndrome. As his neurological decline and medical complexity increased, direct therapies and coordinated in-home supports did not keep pace. She became nurse, therapist, seizure monitor, advocate, coordinator and daily caregiver, stepping away from work. The family remains largely stuck without coordinated in-home services and specialized respite that grow with need.

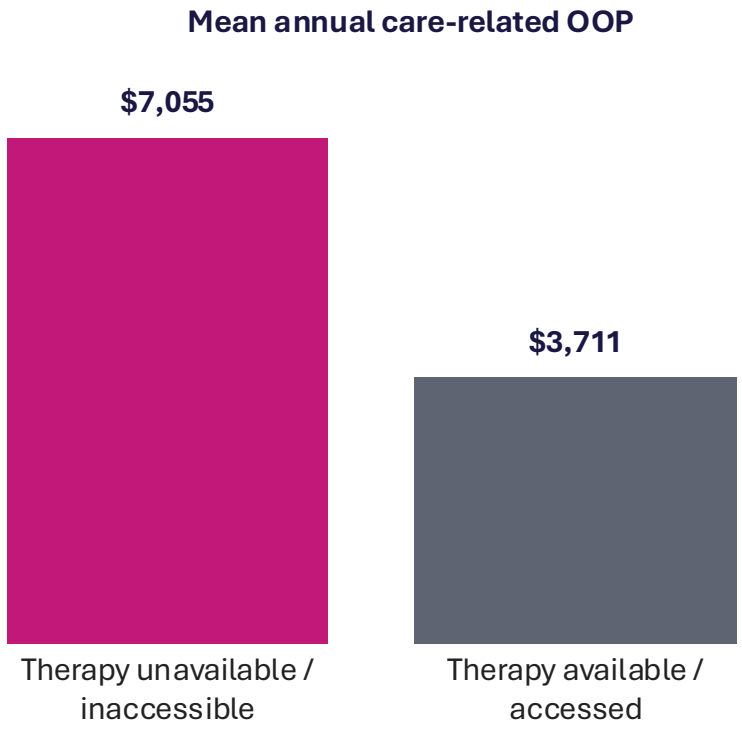
Source: IHE Technical Report, Figure 4, Table 14 and Table D24; Moments That Matter response, Dawn / New Brunswick / Dravet syndrome. Name used per speaker preference.

Treatment delays shift costs back to families

DATA

When the right therapy is delayed, denied or unaffordable, families carry both the risk and the cost.





Access barriers are cost drivers: delays, denials and affordability problems shift cost and risk to families.

Source: IHE Technical Report, Figure 6, Table 19 and Table 20.

Kim: the lifetime cost of a late diagnosis

What happens when symptoms are present for years, but no one connects the dots.

Decade of delay

Lifetime household cost

Symptoms from early life but diagnosis after more than 10 years; delayed answers affected health, work, income, housing and independence.

Readiness lesson

Earlier recognition and navigation are socioeconomic interventions — not only clinical improvements.

1. What was life like before anyone connected the dots?
2. Looking back, what signs do you wish someone had taken more seriously?
3. Did the diagnosis feel like an answer — or an answer that came too late?
4. What do you feel comfortable sharing about work, income, housing or independence impacts?
5. What do you want clinicians and decision-makers to do differently when a patient does not fit the usual pattern?

Lesson: "Kim's story shows that diagnostic delay can become income lost, disability support lost, housing security lost and independence lost."

Story source: Moments That Matter response, Kim / CVID, New Brunswick.

When the answer is outside Canada

Nathan's case asks hard readiness question: what happens when needed expertise exists, but not here?

NATHAN • BRITISH COLUMBIA • Grade V cerebral AVM

When the needed expertise is outside Canada

Timely access matters because delay increases risk while treatment remains feasible.

Nathan is a child with a complex, life-threatening cerebral arteriovenous malformation. Conventional treatment options had been exhausted or deemed inappropriate. An international expert identified staged transvenous embolization as the only realistic curative option, requiring expertise, procedural approach and technology available in very few centres worldwide. His BC Children's team supported referral, and delay increases risk of catastrophic hemorrhage.

Readiness implications

- Transparent criteria for exceptional and urgent access
- Rapid referral to national or international expertise
- Shared provincial/federal responsibility when equivalent care is unavailable
- Data capture and follow-up so exceptional access builds learning
- A pathway that does not depend on crisis advocacy

Source: CORD letter of support for Nathan's request for out-of-country treatment, British Columbia.

From burden to readiness: what Phase 2 Rare Disease Drug Strategy can change

CLOSE

From evidence to action: when hidden costs are visible, system can be developed to reduce them.

Burden shown by survey	Readiness response
Diagnostic delay	earlier recognition, referral triggers, testing access
High hospital and ED use	earlier intervention, monitoring and care coordination
Out-of-pocket costs	coverage for supports, equipment, medical food, navigation
Travel burden	regional/virtual specialty pathways and travel support
Unpaid caregiving	home care, trained respite, family-centred supports
Treatment access barriers	transparent eligibility, faster access, managed access

“Readiness is how we turn waiting into action, and burden into better lives."

Source: IHE Technical Report; CORD Readiness Webinar Series planning materials.

Moments That Matter

Rare Disease Journeys

Help identify the challenges, breakthroughs and practical changes that can improve rare disease infrastructure.

What was the moment that made — or could have made — a difference?

Share one key challenge, breakthrough, achievement, or small change that others should understand.

1 Recognize

First signs, screening, or symptoms that were missed or understood

2 Diagnose

Testing, referral, specialist access, or finally getting answers

3 Access care

Treatment, clinical care, navigation, monitoring, or coordination

4 Support

Community resources, peer support, family burden, or daily life

Your story helps show **where the system is working, where it falls short,** and where practical changes can improve outcomes.

Share your moment

Patients • caregivers • families • advocates

https://www.surveymonkey.com/r/MKLZGB9

[survey link]

Stories may be short. One moment can reveal where systems need to work better.

Canadian Organization for Rare Disorders

CALL FOR PARTICIPANTS

CORD Rare Disease Readiness Advisory Scan

Help shape rare disease readiness in Canada.

CORD is seeking input from people with rare disease knowledge or experience to identify practical indicators of **readiness** across Canada. Your insights will inform future measurement, policy, and system improvement.

WHO SHOULD PARTICIPATE?

- Healthcare professionals
- Researchers & data experts
- Policy & drug access leaders
- Patient organizations
- Patients & caregivers
- Community & support service providers

WHAT YOU'LL DO

- Complete a **15–20 minute** online survey
- Express your interest in participating
- Rate potential readiness indicators
- Share examples of promising practices

THE SURVEY COVERS 6 READINESS AREAS

- Diagnosis & Screening
- Medicines & Treatment Access
- Care Delivery
- Data & System Learning
- Daily Living & Access Supports
- Community Services, Partnerships & Equity

COMPLETE THE ONLINE SURVEY BY JULY 26, 2026

15–20 MINUTES Your input matters!

SCAN THE QR CODE OR VISIT: https://www.surveymonkey.com/r/CA_Rare_Readiness_Survey

DEADLINE: July 26, 2026

This is not a provincial ranking or audit.

Thank you for helping build a stronger future for people living with rare diseases.

What should Canada make visible next?

ENGAGE

Which costs that are still hidden — and which readiness changes would matter most?

1. Which cost is most invisible today?
2. Where does delay create the greatest burden?
3. Which readiness change would reduce burden fastest?
4. What should Phase 2 measure publicly?
5. Which costs should be included in value assessment?

Next: Webinar 3 turns these costs into readiness domains — diagnosis, specialist access, treatment, integrated care, supports, data and equity.

Register for Webinar 3:
https://3sixtypublicaffairs.zoom.us/webinar/register/WN_8HW4_B9DT5iMLJciiQTipg

Webinar 3 — **July 28 | 12 EDT**

Building the Baseline: How Does Canada Measure Up in Terms of Rare Disease Readiness?

60-minute interactive indicator lab: policy and delivery indicators of rare disease readiness across Canadian provinces/territories

RD Strategy + readiness:

Turns Phase 2 from a funding concept into measurable implementation: governance, diagnosis, access, care coordination, RWE, equity, and reporting.

Source: CORD Webinar 2 pulse survey design; CORD readiness assessment framing.

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