Toward a Rare Disease–Ready Canada

Using genomic testing readiness as one concrete example — then moving to an integrated Phase 2 rare disease strategy and readiness scorecard.

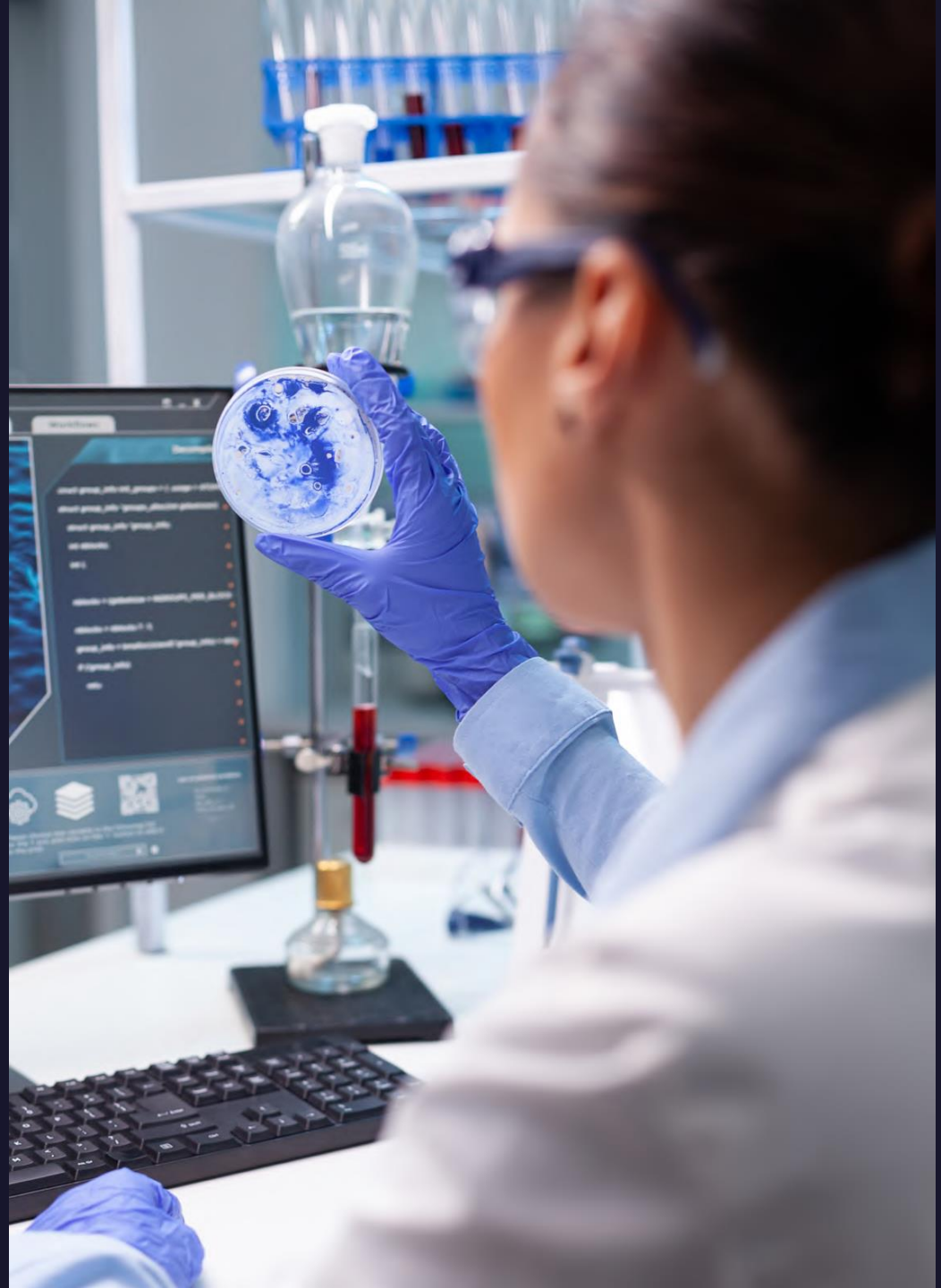
Speakers:

- Don Husereau, Author, State of Readiness Progress Report for Genome-based Testing
- Durhane Wong-Rieger, Canadian Organization for Rare Disorders
- Trish Guimond, Manitoba Rett Syndrome Association

Moderator:

- William Dempster, 3Sixty Public Affairs

Canadian Organization for Rare Disorders



A one-hour arc with one integrated message

Genomic testing is a case study; rare disease system readiness is the webinar objective.

1

0–5 min

Framing the system problem

Rare disease readiness is not just drugs or tests.

2

5–30 min

Genomic testing readiness

Concrete evidence of readiness gaps and progress

3

30–50 min

CORD Phase 2 proposal

From drug access to diagnosis, care, data and outcomes.

4

50–60 min

Readiness scorecard ask

Proposing a practical tool to identify capacity, gaps and pilots.

Takeaway Message

In Phase 1 Canada invested $1.5B in rare disease medicines. In Phase 2 we must build the pathway from diagnosis, treatment, and care to patient and system outcomes.

Start with the pathway patients experience

The system must connect the first concern to outcomes — not just fund one step in the middle.

Recognition fails

Patients remain in broad categories too long.

Testing fails

Targeted therapies remain theoretical.

Monitoring fails

Coverage cannot learn from outcomes.

Coordination fails

Families become the care system.

Readiness question — **Are the conditions in place for diagnosis, treatment, care, data, equity and outcomes to connect?**

Source basis: CORD–CRDN implementation pathway and determinants → delivery → outcomes logic model.

Why now: a time-limited national policy window

Phase 1 created a foundation; Phase 2 must prevent an access cliff and build system readiness.

2024–2027

Phase 1 rare disease drug strategy

Bilateral agreements create a national platform and urgent expectations for access.

Budget 2026

Phase 2 decision point

Sustain medicine funding and build infrastructure around diagnosis, data, care and outcomes.

March 2027

Transition risk

Without a commitment, patients and provinces face uncertainty and uneven planning.

Budget 2026 Ask: **Confirm, operationalize and expand a Phase 2 National Rare Disease Strategy.**

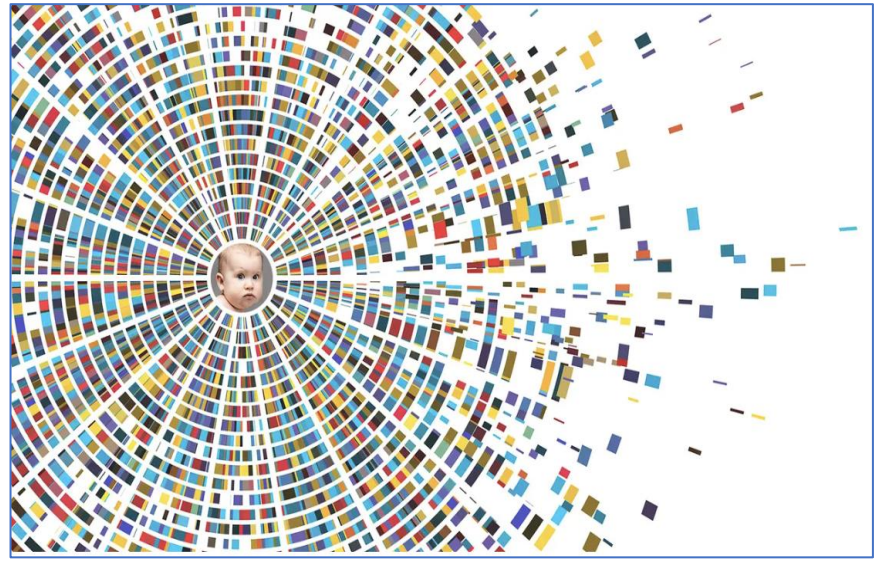
Source basis: CORD 2026 federal pre-budget submission.

Genomic testing readiness is a concrete example

It is a stress test of whether a system can adopt complex innovation — setting up the question about innovation throughout the rare disease system.

The 2026 genomic readiness report asks whether enabling conditions are present to consider and adopt genomic/genetic tests.

1. **Scope matters** — Focused on hereditary testing and testing in cancer; infectious disease surveillance is outside scope.
2. **Method matters** — A readiness framework was applied province by province to assess enabling conditions.
3. **Lesson matters** — Readiness to adopt a test is not the same as readiness to deliver, monitor, and improve care.



Our proposition: genomic testing readiness is the worked example; Rare Disorders readiness is the broader implementation challenge.

Source basis: 2026 State of Readiness Progress Report.

Why the genomic readiness paper matters

It gives us a concrete example of system readiness — but is not a proxy for the whole rare disease system.

Why assess readiness?

Complex innovations need enabling conditions — not just clinical evidence.

Genome-based testing depends on labs, specialists, IT, HTA, financing, training and care navigation.

The report creates an objective platform to identify gaps and catalyze action.

Boundary for this webinar

Scope: hereditary testing and cancer testing.

Not in scope: infectious-disease surveillance or local care-delivery performance.

An example that shows what a readiness scorecard can reveal.

Bridge to Phase 2: from readiness to adopt a test → readiness to deliver an integrated rare disease pathway

Source basis: State of Readiness Progress Report, pp. 3, 9–11.

How the genomic readiness scoring was done

A mixed-methods framework was applied province by province to assess enabling conditions.

1 Gather evidence

Narrative literature / grey literature review + semi-structured interviews with laboratory leaders and policy stakeholders.

2 Define conditions

Same readiness conditions as the 2023 report, organized into three system domains and tied to the Quadruple Aim.

3 Apply + validate

Conditions applied to provinces; draft grades presented to steering committee, regional informants and publicly for feedback.

Readiness parameters grouped into three domains

Infrastructure

- Communities / networks
- Resource planning
- Linked information systems

Operations

- Entry / exit for innovation
- Evaluation function
- Service models
- Awareness + navigation

Health care environment

- Innovation integration
- Financing approach
- Education + training
- Regulation / quality

Important nuance: this scores system readiness to consider and adopt genomic tests; it does not score the full rare disease care system.

Source basis: State of Readiness Progress Report, pp. 15, 24–27.

From the 2023 report card to the 2026 update

The baseline assessment helped trigger action; the update shows progress but no province is fully ready.

2023 baseline

Five provinces assessed

AB, BC, ON, QC, NS — representing >85% of Canada's population

Alberta	B+
Quebec	B-
British Columbia	C
Nova Scotia	C-
Ontario	D

Key gaps: linked data, fair/timely review, navigation/education, financing and innovation pathways.

What changed

- BC + AB genomics innovation funding
- Alberta transparent evaluation framework
- Ontario province-wide education standards
- Quebec gene-test guidance + analytic standards
- Nova Scotia single intake / stronger evaluation
- CDA-AMC biomarker assessment framework drafted

Message: readiness assessment can catalyze system action.

2026 update

A more complete national picture

Original five re-assessed + remaining provinces; PEI and territories treated as referral jurisdictions.

Alberta / Newfoundland & Labrador	B+
Ontario / Nova Scotia	B
Quebec / Manitoba / New Brunswick	B-
British Columbia / Saskatchewan	C

All 2023 provinces improved; biggest gains were Ontario and Nova Scotia.

Persistent gaps: HTA, financing/test development, information linkage and education.

Source basis: State of Readiness Progress Report, pp. 4, 10–11, 29.

What the genomic readiness report found

Progress is real — but Canada remains only partially ready.

C → B+

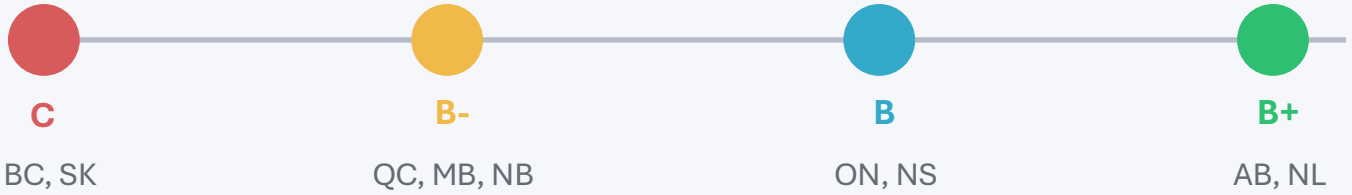
Range of provincial readiness grades for considering and adopting new testing

2

Most notable improvements since 2023: Ontario and Nova Scotia

0

Provinces rated fully ready; persistent gaps remain



Persistent gap areas

- transparent evaluation
- flexible financing
- linked data systems
- education + navigation

Source basis: 2026 State of Readiness Progress Report, executive summary and provincial grade summary.

The transferable lesson: readiness is a system property

The genomic testing gaps are useful because they point to capabilities every rare disease pathway needs.

1

Plan

Can the system anticipate need and allocate capacity?

2

Evaluate

Are decisions transparent, timely and consistent?

5

Learn

Are data linked to outcomes and reassessment?

3

Finance

Can funding cover development, delivery and monitoring?

4

Navigate

Do patients and clinicians know where to go?


Current Oncology

Article

Readiness to Consider and Adopt Genetic and Genomic Tests in Canada—An Update to the State of Readiness Report

Simple Summary

Abstract

https://www.mdpi.com/1718-7729/33/6/334

State of Readiness Progress Report PDF

Source basis: State of Readiness report recommendations and CORD–CRDN implementation framework.

Ema’s story: readiness gaps are pathway gaps

A Rett syndrome diagnosis came only after years of appointments, partial explanations, and parent-led advocacy.

What the family experienced

~12 months	Loss of words/sounds, sleep problems, fevers and growing parent concern
Early years	Global developmental delay and autism explained some symptoms, but not all
Seizures	Parent recognized episodes but struggled to be believed until a seizure was witnessed
~3 years	Genetics began; early result raised questions, but did not explain the full picture
Before 5	Parent prompted Rett testing; MECP2/Rett diagnosis finally confirmed

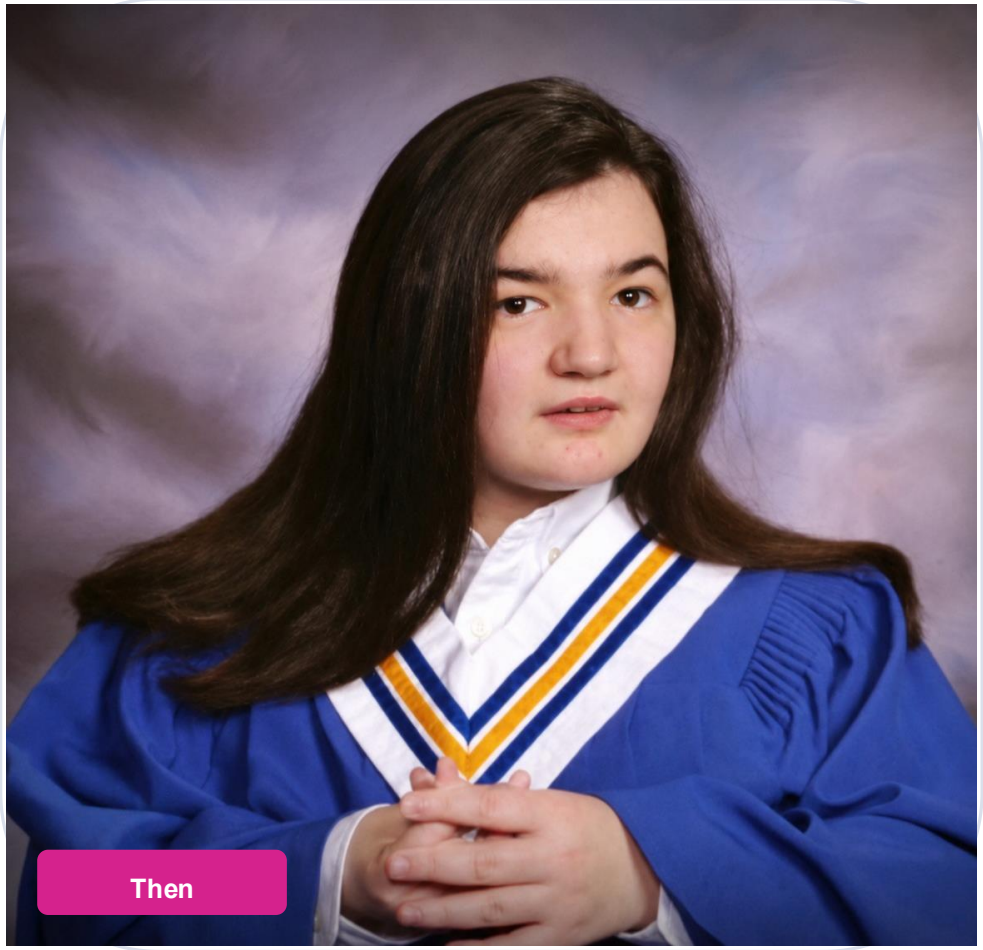
Readiness lesson

Recognize	A rare disease pathway must help clinicians escalate when the usual diagnosis does not fit.
Test + interpret	Genetic/genomic testing must be timely, explainable, and connected to expert interpretation.
Refer + coordinate	A confirmed diagnosis must trigger specialist access, local care planning, and navigation.
Access + learn	Therapies, monitoring, RWE, and patient-reported outcomes need to be ready after diagnosis.

The family did not need a test alone. They needed a system that could recognize, test, interpret, refer, support — and keep learning.

Ema today

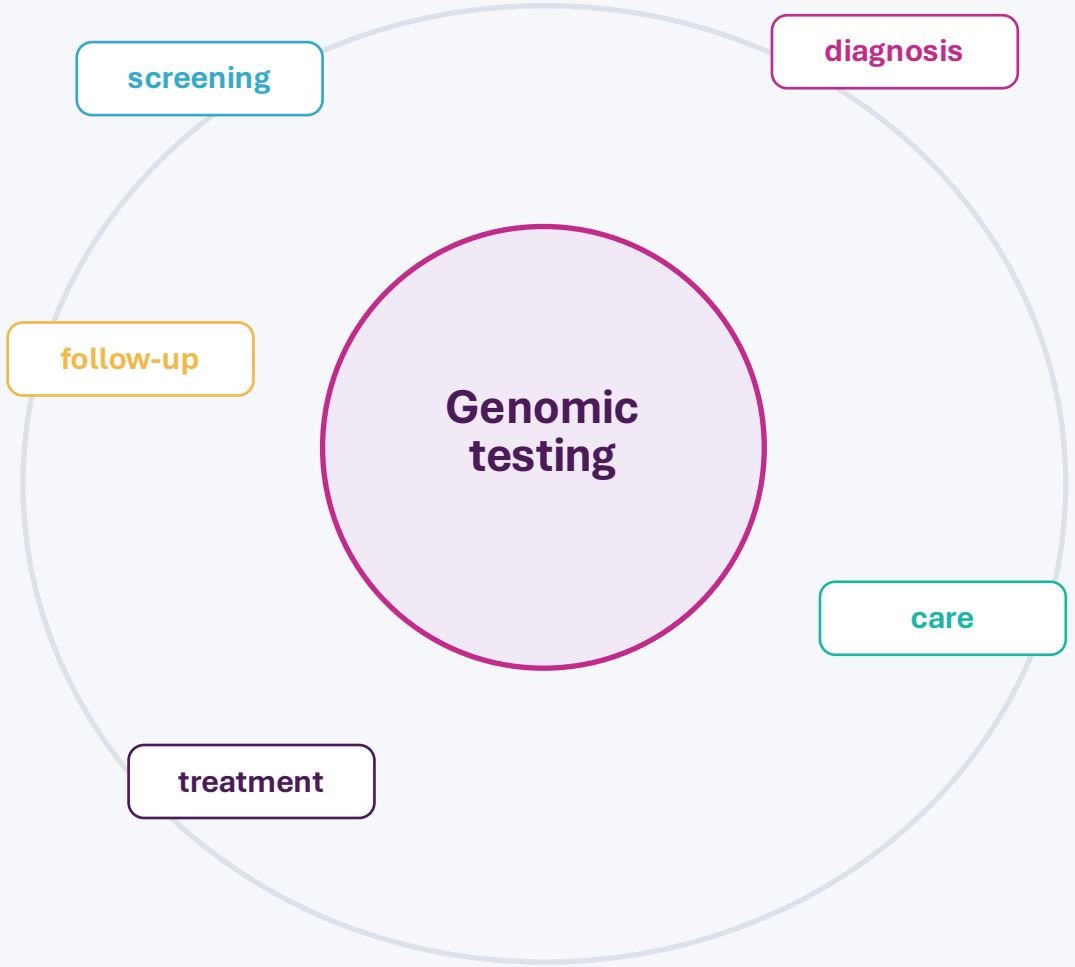
High school graduate — a milestone that matters.



What readiness is for: better pathways, stronger supports, and meaningful milestones.

Rare disease readiness is broader than genomic capability

Many rare diseases are not genetic; many genomic answers still require care, treatment and support systems.



The strategic move

Start with the integrated rare disease system. Use genomic testing readiness as a concrete example of how to assess capacity, gaps and action.

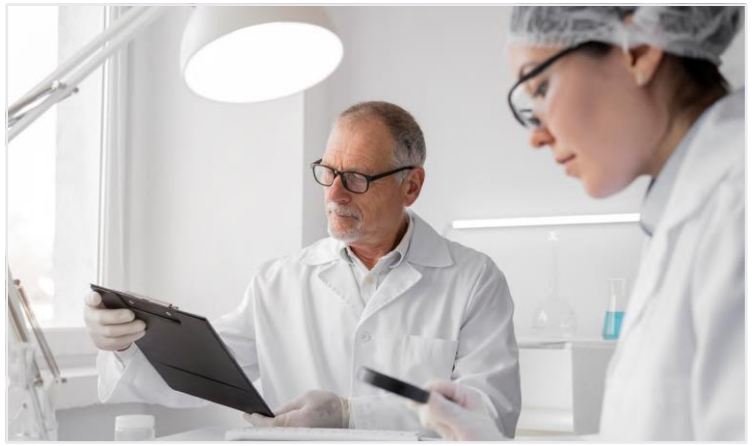
This avoids over-crediting genomic gaps while still preserving the value of the readiness report: it demonstrates a method, a vocabulary and a policy opening.

From: one technology readiness report → **To: whole-pathway RD readiness**

Phase 1 RD Drug Strategy was right starting point — not the end point

A drug strategy must now be connected to diagnosis, eligibility, delivery, monitoring and outcomes.

Phase 1: medicines	**A historic start to address timely and equitable access to rare disease drugs.**
Phase 2: system readiness	**Equitable access to medicines depends on the system around them.**



Risk if Phase 2 is not defined

An access cliff, uneven provincial planning and lost momentum.

Identify → **Confirm** → **Fund** → **Deliver** → **Monitor** → **Learn**

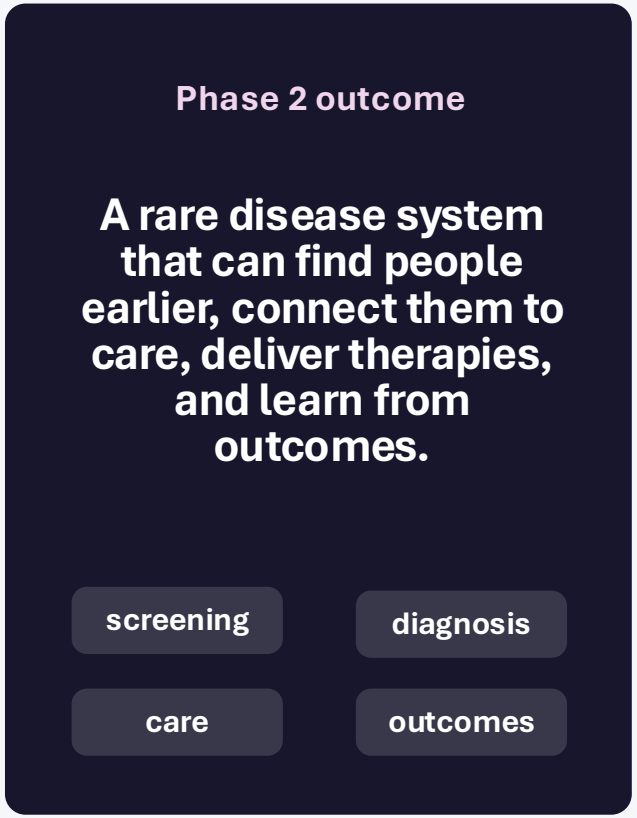
The drug pathway only works when each step is ready.

Source basis: CORD 2026 federal pre-budget submission.

Phase 2: build the whole rare disease ecosystem

A practical proposal: finance the components that make treatment access meaningful.

1. **Sustain medicines funding** — Protect and expand timely, equitable access.
2. **Federal leadership + oversight** — Align objectives, reporting and accountability.
3. **Provincial / territorial action plans** — Make implementation visible and measurable.
4. **Diagnosis, screening + data infrastructure** — Build early identification, registries, RWE and linked data.
5. **Innovation ecosystem** — Connect patients, clinicians, researchers, public agencies and industry.



Source basis: CORD 2026 federal pre-budget submission, Phase 2 proposal.

Implementation backbone: capacity → delivery → impact

The scorecard should ask whether investments change care and outcomes that matter.

Determinants

System capacity

policy • funding • workforce • expertise • infrastructure

Delivery

How care is organized

entry • referral • diagnosis • coordination • treatment

Outcomes

Patient + family impact

clinical • experience • psychosocial • economic • equity

Assessment question

Are the right system conditions in place — and are they improving care?

Source basis: CORD–CRDN workshop deck, IMPACTS logic model.

Canadian Organization for Rare Disorders & Canadian Rare Disease Network: complementary leadership for implementation

Patient-led urgency plus clinical, research, diagnostic, data and implementation expertise.

1

CORD

Patient-led advocacy
Provincial coalition-building
Public accountability
Patient/family outcomes + equity

2

CRDN

Clinical + diagnostic expertise
Genomics + data methods
Implementation science
Publication + knowledge translation

✓

Joint outcome

National implementation: Readiness assessment + scorecard
Pilot action plans
Evidence for Phase 2 investment

Governance: Patient-community led with clinical collaboration on scorecard and implementation evidence.

Source basis: CORD–CRDN consultation workshop deck and governance mandate.

Rare Disease Readiness Status Scorecard

A planning tool — not a ranking exercise. It identifies assets, gaps and where action can begin.

Anchor domains

- Governance + patient partnership
- Recognition, referral + diagnosis pathways
- Therapy access, monitoring + care coordination
- Data, registries + real-world evidence
- Equity, family burden + public reporting

Common scoring rubric

- 0 — No identifiable process
- 1 — Ad hoc / site-specific
- 2 — Defined in some pathways
- 3 — Province-wide accountability
- 4 — Integrated, measured, reportable

Scorecard Output **Provincial/territorial readiness profiles + national synthesis + 12–18 month action recommendations.**

Source basis: CORD–CRDN Rare Disease Readiness Status Scorecard proposal.

UK Evaluation framework provides an evidence bank (not a template)

Borrow tested readiness questions; adapt ownership, data and indicators to Canada.

Translation, not transplantation

- ✓ Does the underlying problem exist in Canada?
- ✓ Who would own the indicator here?
- ✓ Can evidence be collected across jurisdictions?
- ✓ Would it identify a feasible 12–18 month action?
- ✓ Does it reflect patient and family experience?

Starter indicator groups

- Governance
- Diagnosis
- Care
- Access + data
- Equity + learning

Source basis: Canada Advisory UK Action Plans Scorecard deck and England Rare Diseases Action Plan 2026 Annex A.

From scorecard to action planning

The first assessment should lead directly to feasible 12–18 month improvements.

1 → 2 → 3 → 4

Assess status

Map what exists and where readiness is uneven.

Find pathway breaks

Identify where patients get stuck between recognition, diagnosis, treatment and support.

Select priorities

Choose 2–3 feasible pathways or system gaps.

Measure progress

Track capacity, delivery, outcomes, linkages and equity.

Capacity → **Delivery** → **Patient + Family Impact** → **Public Reporting**

Action planning should be paired with public accountability and a learning cycle.

Source basis: CORD–CRDN readiness scorecard use model.

A focused 90-day path to a pilot-ready scorecard

Keep the first cycle manageable: adapt, test, learn, refine.

Days 1–30

Confirm scope

Confirm domains; map UK-derived indicators to Canadian readiness questions.

→

Days 31–60

Build v1.0

Select pilot indicators; identify evidence sources; draft scoring rubric.

→

Days 61–90

Test + refine

Pilot with 2–3 jurisdictions; refine method; prepare national assessment protocol.

Output: Canadian Rare Disease Readiness Status Scorecard v1.0 + pilot protocol

Source basis: Canada Advisory UK Action Plans Scorecard deck, 90-day pilot method.

NEXT STEPS

Move from commitment to readiness

1. Confirm Phase 2 rare disease funding beyond the first agreements.
2. Support provincial/territorial action plans that connect diagnosis, care, treatment, data and outcomes.
3. Authorize CORD–CRDN to pilot the Rare Disease Readiness Status Scorecard.
4. Use scorecard findings to prioritize 12–18 month implementation actions.

The white paper makes the case. The scorecard shows the status. Action planning defines what changes next.

Discussion

What would make this useful to CORD, CRDN, provinces/territories, and patients/families?

- ? Does the integrated system frame feel right?
- ? Which pathway breaks are most urgent to assess first?
- ? Which 2–3 jurisdictions should pilot the scorecard?
- ? What evidence can be collected consistently and quickly?

Source basis

2026 State of Readiness Progress Report • CORD 2026 Federal Pre-Budget Submission • CORD–CRDN Consultation Workshop Deck • Canada Advisory UK Action Plans Scorecard • England Rare Diseases Action Plan 2026 Annex A

CORD Rare Disease Readiness Dialogues

Reviewing Phase 1. Shaping Phase 2. Building a future-ready rare disease system.

A four-part CORD-led webinar and engagement series connecting Canada's Rare Disease Drug Strategy to system readiness: diagnosis, treatment access, care coordination, data, evidence, equity, outcomes, and readiness preparation.

Session 1 — **June 23 | 12 EDT**

From Genomic Testing Readiness to Rare Disease System Readiness

Uses genomic testing readiness as a concrete example of how system readiness can be assessed without making genomics the whole rare disease agenda.

RD Strategy + readiness:

Launches the readiness lens: Phase 1 opened access momentum; Phase 2 must connect diagnosis, care, data, equity, and outcomes.

Session 2 — **July 14 | 12 EDT**

From Burden to Value: What the CORD Survey Shows and Why Phase 2 Matters

Presents findings on diagnostic delay, 12-month reported healthcare use and spending, access barriers, travel, caregiving, and household impact.

RD Strategy + readiness:

Shows why the Drug Strategy needs systems that act earlier, match patients to the right intervention, support families, and measure value.

Session 3 — **July 28 | 12 EDT**

Measuring Readiness: A Canadian Rare Disease Readiness Assessment and Scorecard

Introduces a provincial/territorial readiness scorecard using Canadian priorities and international learning to identify assets, gaps, and action priorities.

RD Strategy + readiness:

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

Session 4 — **Aug. 11 | 12 EDT**

Readiness in Time of MFN: Preparing Canada for Rare Disease Innovation

Applies readiness to MFN-style global pressures, clinical-trial competition, launch sequencing, innovative therapies, managed access, RWE, and equitable benefit.

RD Strategy + readiness:

Prepares Canada to identify patients, activate sites, deliver access, generate evidence, and return value to patients, developers, and the health system.

Series through-line: Session 1 asks “Are we ready?” Session 2 shows why readiness matters. Session 3 defines how readiness can be measured. Session 4 applies readiness to access, innovation, trials, launches, evidence, and equitable patient benefit.

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

Add survey link here:

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

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

Canadian Organization for Rare Disorders

@raredisorders

raredisorders_cord

@raredisorders

@CORDRareDisorders

@canadian-organization-for-rare-disorders