



Canadian Organization
for Rare Disorders

Webinar 1: Canada's Rare Disease Drug Strategy – Sep 29, 2026

How Readiness Is Built — and What Happens When it is Broken

Hemophilia • Thalassemia • Sickle Cell Disease

Panel:



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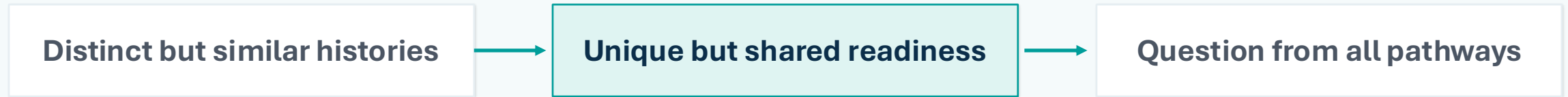


From Phase 1 learning to Phase 2 readiness

Purpose: guide a synthesis-first conversation with patient/community representatives.

Lessons from Synthesis-Three Rare Blood Disorders

A comparative frame helps the audience understand the lessons before hearing disease-specific examples.



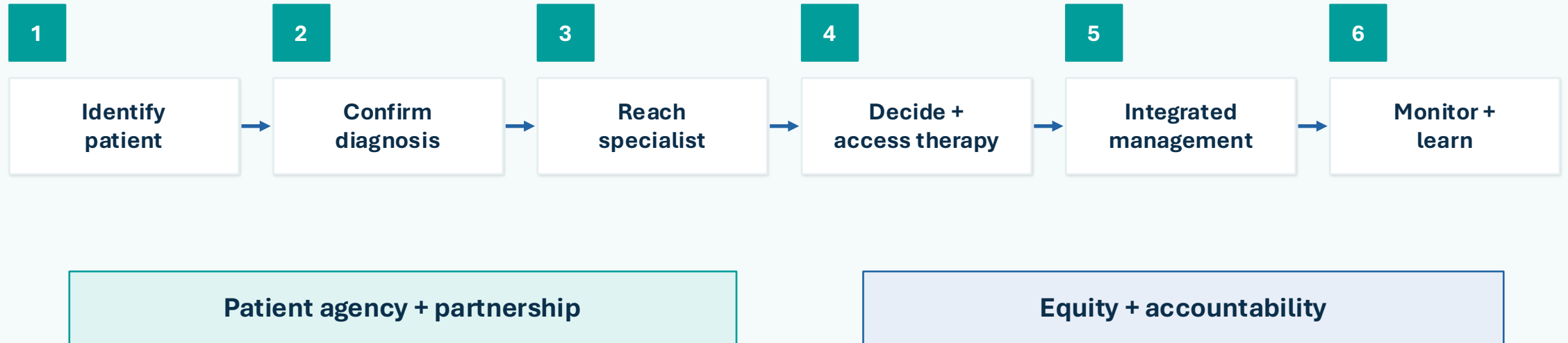
Core question

What must Canada be ready to do around a funded therapy so the right patient can receive the right treatment, at the right time, with the care and learning needed to optimize benefit?

This is a Phase 2 RDDS planning conversation: build on what Phase 1 made possible to identify what should be strengthened next.

End-to-end readiness: the pathway we are testing

Drug access is essential, but it sits within a larger patient and system pathway.



Map of repeated lens: every disease may have different gaps, but the question is whether the pathway connects for patients.

Three pathways, three readiness archetypes

The diseases are not models to copy; they make different system lessons visible.

Hemophilia

Accumulated capability becomes reusable infrastructure

Centres, home care, product systems, standards, data and patient–clinician partnership allow successive innovations to enter an organized pathway.

Thalassemia

Clinical sophistication can coexist with burden and life-course gaps

Scheduled chronic treatment, chelation burden, transition, changing communities and advanced therapies expose what conventional metrics may miss.

Sickle Cell Disease

Readiness must work beyond the specialist centre

NBS and specialist expertise matter, but acute care, transition, pain credibility, trust and equity determine whether the pathway reliably reaches patients.

Panel task: test the archetypes. What do they capture well, what do they miss, and what do they teach us for Phase 2?

Hemophilia: turning points that built cumulative capability

Chronology provides context for discussion

1953–54

Clinical + patient organization

Montreal programs and the Canadian Hemophilia Society develop together: patient–clinician co-development.

1964–66

Cryoprecipitate changes delivery

High-potency replacement makes treatment more effective and more distributed.

1970s

Home infusion + self-management

Treatment moves from repeated hospital attendance to patients/families as active co-managers.

1976–78

National coordination + Winnipeg I

CHS clinical advisory structures, national survey and comprehensive-care consensus make gaps visible and actionable.

1980s–90s

Blood crisis + governance rebuild

Safety, surveillance, public participation and accountability become pathway capabilities.

1998–2020s

Standards, data, new modalities

Winnipeg II, national standards, registry/centre assessment and non-factor/gene therapies test whether the mature pathway can adapt.

Discussion anchor: Which capability lasted because patients and clinicians built it together?

Hemophilia experience: test of the archetype

Patient/community response: start with the archetype, then offer one or two concrete examples.

1

Does the archetype fit?

Does “accumulated capability becomes reusable infrastructure” capture hemophilia in Canada? What does it miss?

2

What is the key lesson?

Which capability taught by this profile matters most for other rare diseases: centres, home care, product systems, standards, data, governance or partnership?

3

What is your reflection or experience?

What concrete patient, family, clinician or community experience proves, complicates or challenges the archetype?

Thalassemia: transitions in treatment, care and populations

The pathway is clinically sophisticated, but readiness depends on life-course fit and patient-defined value.

1980s–90s

Community infrastructure

Ontario Thalassemia Association/TFC, family networks and TIF links create visible community capacity.

1990s–2000s

Population shifts

Readiness shifts from known Mediterranean family networks to newcomer identification, language and navigation needs.

Transfusion era

Chelation burden becomes central

Outcomes depend on repeated transfusion plus demanding home chelation; adherence reflects burden, not simply willingness.

2006–16

Oral chelation expands choice

Deferasirox and deferiprone make treatment burden and psychosocial autonomy part of value.

2009–24

Guidance + transition

Canadian guidance and transition/adult programs make comprehensive life-course care an explicit requirement.

2020–26

Reblozyl + Casgevy

Reduced transfusion burden and potential cure test funding, cellular therapy capacity, monitoring and patient choice.

Discussion anchor: What did patient experience reveal that clinical measures alone did not?

Thalassemia experience: test the archetype

Patient/community response: start with the archetype, then use one or two concrete examples.

1

Does the archetype fit?

Does “clinical sophistication can coexist with burden and life-course gaps” capture the thalassemia pathway in Canada?

2

What is the key lesson?

What does this profile teach about burden, adherence, transition, newcomer navigation, treatment choice or advanced-therapy readiness?

3

What is your reflection or experience?

What lived or community experience shows what clinical measures alone missed?

Sickle cell disease: pathway challenges beyond the centre

Specialist expertise matters, but acute-care reliability, equity and trust determine whether the pathway reaches patients.

Recognition

Population + equity lens

SCD visibility is linked to racialized and newcomer communities; readiness includes credibility and health-system response.

2005–06

Advocacy creates NBS capability

Ontario adds SCD to newborn screening after sustained patient/community and clinician advocacy.

Prevention

TCD, transfusion, hydroxyurea

The pathway moves from crisis response toward prevention and disease modification.

2014–17

Transition + adult infrastructure

Joint transition clinics and Ontario investment recognize red-blood-cell disorder care as specialized infrastructure.

Ongoing

Acute-care interface remains fragile

Pain disbelief, ED delays and non-specialist settings can undermine even strong specialist care.

2022–26

Registry, Bill S-201, Casgevy

National data/policy efforts and gene editing expose funding, delivery, equity, decision and long-term follow-up needs.

Discussion anchor: Where does the formal pathway differ most from what patients actually experience?

Sickle cell disease experience: test the archetype

Patient/community response: start with the archetype, then use one or two concrete examples.

1

Does the archetype fit?

Does “readiness must work beyond the specialist centre” capture the SCD pathway in Canada?

2

What is the key lesson?

What does this profile teach about newborn screening, acute care, transition, pain credibility, trust, equity or long-term monitoring?

3

What is your reflection or experience?

Where does the formal pathway differ most from what patients actually experience?

Lesson 1 — Readiness is cumulative

Pathways are built over time; a new drug enters what already exists.

Funding a therapy does not automatically create the pathway for using it well.

Hemophilia

Home treatment + comprehensive centres + national coordination became adaptable infrastructure.

Thalassemia

Repeated transfusion and chelation helped build expertise, but the pathway remains dependent on institution and life-course factors.

Sickle Cell Disease

NBS and specialist care improve prevention, but wider system expertise and responsiveness remain uneven.

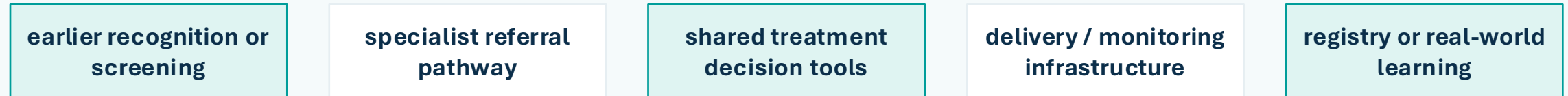
Prompt to panelists: What capability in your pathway took years to build, and what did it make possible later?

Lesson 2 — Innovation requires and generates capability

A therapy can benefit from existing readiness, but implementation should also leave something durable behind.



Examples of durable capability:



Prompt: What did a treatment change in your disease that remained useful after the initial access decision?

Lesson 3 — Patient agency builds the pathway

Patient communities do more than provide input; they identify gaps, create urgency, define value and help build capability.

Patient agency is not an add-on to the pathway. It is important way the pathway gets built.

Name the gap

make invisible burden
visible

Shape value

define benefit beyond
narrow endpoints

Build capacity

registries, education,
navigation, advocacy

Hold accountable

ask what changed for
patients

Prompt: What did patients or families understand that the formal system initially missed?

Lesson 4 — Readiness is multidimensional

Different parts of the pathway can be ready or unready at the same time.

Diagnosis

Is the right patient found?

Funding

Can access be secured?

Decision

Can patients choose with support?

Clinical

Is expertise available?

Delivery

Can treatment be given safely?

Data

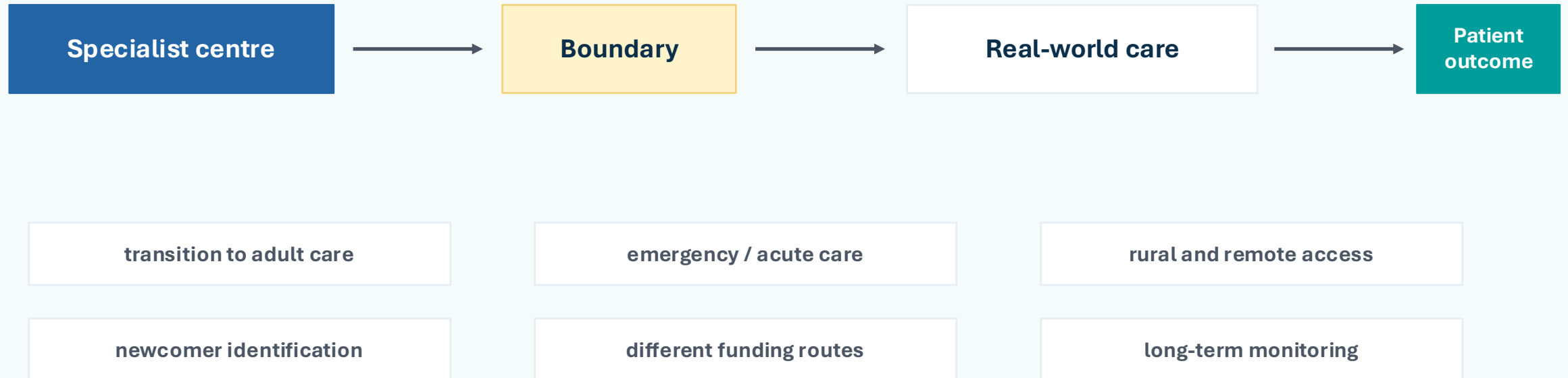
Can outcomes be learned?

A single “ready / not ready” label hides the mechanism that matters.

Prompt: In your pathway, which part is most ready — and which part would fail first if a new therapy arrived tomorrow?

Lesson 5 — Boundaries are where pathways break

A pathway can look strong inside the specialist centre but fail at hand-offs and interfaces.



Prompt: Which hand-off or boundary most undermines the pathway in your disease?

What current gene therapies expose

Gene therapy makes historic readiness visible because it depends on the whole pathway.

Hemophilia

Mature infrastructure may support eligibility review, counselling and follow-up — but gene therapy can sit outside usual blood-product pathways.

Thalassemia

Cellular therapy capacity, conditioning, travel, fertility, monitoring and patient choice matter as much as approval or listing.

Sickle Cell Disease

Curative potential intersects with equity, trust, pain experience, access to centres and long-term follow-up obligations.

Cross-case prompt: what needs to be ready before a therapy arrives, and what should access leave for the next patient?

Conference bridge: applying the lessons to current therapy pathways

Preview of workshop cases to be discussed at CORD Conference Nov 12-13, 2026

FCS	Redemplo + Tryngolza	genetic confirmation • pancreatitis-risk criteria • treatment choice
PH1	Oxlumo	earlier diagnosis • genetics • nephrology / metabolic referral
VHL	Welireg	hereditary identification • family testing • surveillance
FOP	Sohonos	ultra-rare recognition • expert management • first therapy
NF1	Koselugo	eligibility • specialist assessment • longitudinal care

Conference bridge: these cases will be used to stress-test whether the five lessons transfer — and where a new archetype is needed.

What this could mean for Phase 2 Rare Disease Drug Strategy

Not a wish list; it is a pathway-based way to define what the next investment should enable.

1

Preserve dedicated rare-disease drug access

Do not lose the access gains made possible by Phase 1.

2

Assess the surrounding disease–drug pathway

When a therapy changes diagnosis, eligibility, delivery or management, evaluate the whole pathway.

3

Support enabling capabilities

Testing, specialist networks, delivery, integrated care and monitoring should be funded when needed for appropriate use.

4

Embed patient agency and partnership

Patients should help design, implement and review the pathway, not only comment on reimbursement.

5

Report access, outcomes and durable capability

Phase 2 should ask what changed for patients and what capacity remained for the next innovation.

Closing question: what should Phase 2 RDDS be expected to make possible around the drug?

What should we carry into November?

- 1 Which lesson held most strongly across the three diseases?
- 2 Which lesson needs to be changed or sharpened?
- 3 What would make the next therapy fail to deliver its full benefit?
- 4 What should Phase 2 RDDS be expected to make possible around the drug?