RARE DISEASE FRAMEWORK FOR ALBERTA

SUMMARY REPORT - SEPTEMBER 2025

bio
alberta
Association for Life Sciences Industry

INNOVATIVE
MEDICINES
CANADA

MÉDICAMENTS
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CANADA

ABOUT BIO ALBERTA

BioAlberta is the voice and champion for the life sciences and health innovation industry in Alberta. As a member driven industry association, our mandate is to convene and connect within our network to promote, advocate and drive growth in Alberta's life sciences industry.

ABOUT IMC

Innovative Medicines Canada represents Canada's innovative pharmaceutical industry. IMC helps its members discover, develop, and deliver innovative medicines and vaccines, and contribute to the life sciences ecosystem across Canada. Guided by a strict Code of Ethical Practices, IMC members work with governments, private payers, healthcare professionals, and stakeholders to contribute to the total health of Canadians.

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OVERVIEW

Rare diseases collectively affect a significant number of Albertans, despite each individual condition being uncommon. Defined as conditions that impact fewer than 1 in 2,000 people, rare diseases encompass a wide range of genetic, metabolic, acquired, and degenerative disorders. Although they are classified as rare, 1 in 12 Canadians are affected by a rare disease.[1]

As the Government of Alberta works to address systemic healthcare challenges such as long wait times, limited primary care access, and healthcare workforce shortages, it is essential to also recognize and address the unique needs of the rare disease community. With over 350,000 Albertans living with a rare disease, patients and their families face disproportionately prolonged diagnostic journeys, fragmented care, and inequitable access to specialized treatments and support networks. Moreover, healthcare providers face significant challenges in delivering care for rare disease patients without the necessary infrastructure, resources and coordination tools. This lack of support not only makes it more difficult to provide timely and comprehensive care but also contributes to inefficiencies and added strain on the broader healthcare system.

To explore the current and future states of rare disease care within the province, Bio Alberta and Innovative Medicines Canada hosted a two-day multi-stakeholder workshop with support from the Pivot Group, an Alberta-based design consultancy. This collaborative event, or "design lab," brought together patients, caregivers, advocates, industry partners, and government to develop a made-in-Alberta approach to rare disease care. Thirty-one pre-event qualitative interviews with representatives of all stakeholder groups provided insights into existing challenges and opportunities within Alberta's rare disease landscape, informing the structure and focus of the workshop.

By bringing patients, clinicians, advocates, industry partners, and government together, we can better understand the current experience within the rare disease care system and ensure meaningful, people-centred solutions that address the current deficits in the Alberta system and that allow Alberta the opportunity to show leadership towards meaningful change.

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[1] Government of Canada improves access to affordable and effective drugs for rare diseases

KEY THEMES FROM RARE DISEASE STAKEHOLDER DISCUSSIONS

Three key themes emerged throughout the course of the pre-session interviews and the design sprint. These should serve as foundational guiding principles for any action that is taken to improve rare disease care in the province.

1. **Co-designed solutions:** Patients, families, caregivers, researchers and healthcare providers need to be co-design partners in all efforts to improve the system in Alberta. Policy and decision-makers need to ensure that policies and programs are designed and that decisions made with meaningful engagement from those who are most impacted.
2. **Equity-focused care:** Any system must ensure equity in access for all Albertans facing the rare disease journey, regardless of geography, socioeconomic status, or background.
3. **Innovative transformation:** Approaches to improving rare disease care in the province of Alberta require new partnerships and different ways of thinking about how to improve the system. The government needs to be open to new partnerships and collaboration to ensure that Alberta can show real leadership in bringing innovative care pathways, rare disease research and treatment options to Alberta patients.

RECOMMENDATIONS FOR ACTION

Several strategic recommendations were identified to address the unique and complex challenges associated with rare diseases in Alberta. The table below outlines the overarching goal for action, as well as short and long-term actions that can be taken to improve rare disease care in the province. More details about these recommendations can be found in the Recommendations and Insights section of this report.

1. IMPROVE COORDINATION OF CARE FOR PEOPLE LIVING WITH RARE DISEASE

SHORT-TERM ACTION	OUTCOMES	BENEFITS
• Establish multi-stakeholder rare disease advisory group of patients, families, clinicians, researchers, industry partners, and policymakers	• A structured mechanism/ framework to actively engage all relevant stakeholders in co-design, ensuring their involvement in both solution development and implementation	• Ensures all rare disease health systems improvement initiatives are practical, readily adopted, sustainable, effective, and tailored to meet the needs of those who rely on and deliver these services • Improved health outcomes • Increased school/workforce participation

2. CREATE NAVIGATION TOOLS AND SUPPORTS FOR PATIENTS, PHYSICIANS AND CARE PROVIDERS

SHORT-TERM ACTION	OUTCOMES	BENEFITS
• Map existing resources and develop a centralized directory of rare disease specialists and support programs	• More efficient and accurate referrals • Improved care coordination among healthcare providers • Reduced duplication of services	• Lower healthcare costs • Improved healthcare outcomes • Increased school/workforce participation

3. REMOVE BARRIERS TO INNOVATIVE AND CUTTING-EDGE TREATMENTS

SHORT-TERM ACTION	OUTCOMES	BENEFITS
• Update drug listing processes to formally involve patients, families, and clinicians in decision-making	• More efficient and accurate referrals • Improved care coordination among healthcare providers • Reduced duplication of services	• Lower healthcare costs • Improved healthcare outcomes • Increased school/workforce participation

4. REDUCE RED-TAPE AND ADMINISTRATIVE BURDEN

SHORT-TERM ACTION	OUTCOMES	BENEFITS
• Update forms and leverage AI to find inefficiencies and streamline workflows	• Reduced administrative overhead • Increased continuity of care; timely care	• Reduced red tape • Cost savings and resource optimization • Reduced burden on healthcare providers • Improved health outcomes • Increased school/workforce participation

5. INCREASE ACCESS TO CLINICAL RESEARCHERS, TRIALS AND PATIENTS

SHORT-TERM ACTION	OUTCOMES	BENEFITS
• Establish a province-wide clinical trial network	• Equitable access to clinical trials • Improved link between researchers and patients	• Increased industry investment and economic growth • Increases Alberta's national leadership and global competitiveness • Improved health outcomes • Increased school/workforce participation

CONCLUSION

This work represents the collective voice of the Alberta rare disease community, including patients, caregivers, families, advocates, clinicians, researchers, and industry partners. By centering the lived experiences of those impacted by rare diseases and the healthcare providers who support them and by continuing to foster strong collaboration among diverse stakeholders, we can develop practical, people-centered solutions to address gaps in diagnosis, access to treatment, and coordinated care. Addressing these gaps is critical to improving the quality of life for the 350,000 Albertans living with rare diseases and achieving better healthcare outcomes.

Alberta's recent signing of the $162 million National Strategy for Drugs for Rare Diseases marks a positive step toward improving access to life-changing therapies. Building this momentum and integrating rare disease care within the province's healthcare reforms will contribute to a more sustainable, efficient, and responsive healthcare system—one that delivers equitable access, supports innovation, and meets the needs of all Albertans. A stronger, more effective healthcare system will enable individuals to more fully participate in daily activities, including school, work, and community engagement, fostering a healthier and more resilient Alberta.

BACKGROUND

Alberta's current rare disease care system is well positioned to leverage existing strengths, including engaged patient and advisory communities, internationally recognized researchers and clinicians, and data and infrastructure supports. However, despite these inherent strengths, many challenges remain for patients and their families living with rare diseases.

To explore the current and future states of rare diseases within Alberta, a two-day collaborative design lab was hosted by Bio Alberta and Innovative Medicines Canada, with support from the Pivot Group. The lab aimed to learn from the unique experiences and expertise of patients, families, caregivers, health care providers, researchers, and other key stakeholders to develop a made-in-Alberta approach to rare disease care.

This report aims to:

1. Summarize the process and activities undertaken before and during the event.
2. Present key findings and themes emerging from discussions.
3. Offer actionable recommendations for improving care and support systems for individuals impacted by rare diseases.
4. Provide a foundation for future policy development and innovative solutions within the Alberta health care system.

DESIGN LAB APPROACH

The rare disease design lab brought together key stakeholders on September 30 and October 1, 2024, in Calgary, Alberta (Appendix A). Over the course of two days, participants collaboratively explored the unique challenges and opportunities associated with rare diseases in Alberta. The primary purpose was to gain a deeper understanding of the needs and experiences of those affected by rare diseases and to identify actionable solutions that could enhance the delivery of care, increase access to innovative treatments, and strengthen support systems across the province.

The event aimed to:

1. Provide a platform for patients and clinicians to share their experiences and articulate the obstacles they face.
2. Create opportunities for dialogue and partnership among diverse stakeholders to align efforts in addressing challenges in rare disease care.
3. Utilize design thinking methodologies to develop creative and forward-thinking approaches to rare disease care in Alberta.
4. Gather insights and recommendations that could guide provincial strategies, leverage federal initiatives (e.g., DRD strategy), influence policy development, and promote systemic change to improve the quality of life, including mental health, for individuals impacted by rare diseases.

This event brought together a diverse group of stakeholders—patients, families, clinicians, researchers, policymakers, and industry partners—each contributing unique perspectives and expertise to the discussions.

This collaborative approach ensured that the insights and recommendations generated were well-rounded and reflective of the complex ecosystem surrounding rare disease care in Alberta.

To ensure that the two-day lab was grounded in stakeholder experience and informed by a wide range of perspectives, a series of key informant interviews with patients, physicians, government, and industry partners were conducted (See Appendix B for interview guide). These 31 interviews were then distilled into six personas. Each persona described an individual's goals, behaviours, challenges and frustrations, and motivations. These were shared and evaluated during the lab to help participants understand the differing experiences and needs of the user groups and to form the basis for challenge recognition and idea generation. Feedback and suggestions for modifications and additions to the personas were incorporated into the updated versions found in Appendix C.

THE DISTINCT NEEDS OF RARE DISEASE

Before outlining the challenges and ideas that were generated by the participants, it is important to explore why rare diseases require a different approach from the one traditionally applied to other health service planning. These conditions pose unique challenges for patients, families, caregivers, health care providers, government decision-makers, and planners. It is only by approaching policy and planning with these considerations in mind that meaningful improvements will take place.

COMMON HEALTH CONDITION	RARE DISEASE
There are treatments available; in many cases there are multiple options.	Only 5% of the 9,000 rare conditions identified today have treatment options, and of those, only approximately 60% have been approved in Canada.
The condition is well known and the disease process is well understood.	Most rare diseases are unknown outside of a few specialists, and for many, the disease mechanisms are still being discovered. As recognition of rare diseases grows, so does the need for support within that community.
Testing and diagnosis for the condition is relatively straightforward and accessible.	Diagnosis often takes many years, and for some conditions, requires sophisticated testing methodologies such as specialized genetic or genomic testing.
Medical and allied health education covers the diagnosis, treatment, and management of the condition.	Very little education on rare diseases is given in medical and allied health education due to limited curriculum time.
Depending on the condition, treatment can be provided in primary care, or with access to specialists.	Most care is provided by highly specialized providers.
For many of the common conditions, treatments can be assessed using traditional approval and reimbursement processes.	Rare disease treatments often do not have the large numbers required to run random-ized controlled trials, and traditional phar-macoeconomic assessments do not fully capture their value
For many of the common conditions, costs can be managed by private insurance plans and/or public formularies using traditional contracting methods	The costs of rare diseases can be prohibitive for both public and private plans, resulting in reduced coverage.

SHARED CHALLENGES

Challenges to the rare disease system were identified throughout the design lab process, both through the key informant interviews that took place in advance of the session and during the two-day workshop itself. These challenges exist across the patient journey and are experienced in different ways, depending on where patients are in the diagnostic and treatment process, and what level of care/treatment is available for their conditions. A patient journey map was developed (Appendix D) to help outline the various steps in the process and identify areas for action. It should be noted that this map does not capture the specific complexities that individual patients or their caregivers or families experience. However, it does highlight the complexity of the current system and the various points where patients can experience care breakdowns or delays.

Despite the variability in patient experience, there were some common challenges identified by multiple groups.

CARE NAVIGATION

All stakeholders agreed that one of the biggest challenges for the group continues to be the ability to navigate the various parts of the healthcare system that are required to obtain a diagnosis and access treatments. The healthcare system in its current state is siloed and difficult to navigate, which can lead to increased utilization of the system, decreasing patient trust in the healthcare system, and extending a patient's time to reaching a diagnosis. While participants acknowledged the strengths in care navigation at the dedicated children's hospitals, they emphasized the major barriers that exist for adolescents and adults seeking diagnosis and treatment. Further, participants highlighted the lack of integration between primary care and specialty care and the lack of resources for patients as they seek a diagnosis. While there are fantastic rare disease patient groups (both national and provincial) that can help patients, these alone are not enough to ensure effective navigation.

ACCESS

Stakeholders identified challenges in access on multiple fronts. Patients often face multiple delays in accessing required screening and testing in order to obtain a proper diagnosis. This is compounded by the delays in accessing specialists that can order and interpret the results from these tests. Further, once a diagnosis is realized, there are challenges in accessing treatments, if they are available. This is due to administrative burdens and delays, as well as decisions around which treatments are covered under private or public insurance plans. This lack of access leads some patients to access treatments outside of the country at significant personal and financial costs. Finally, access to necessary support services is often challenging for patients, due to the complexity in the application process, as well as in some cases the need to have a confirmed diagnosis, a major challenge given the length of the diagnostic odyssey for many patients.

UNDERSTANDING OF RARE DISEASE

Participants noted that there is a lack of understanding about rare diseases among health care providers and the public. This can lead to feelings of mistrust and self-doubt as patients' experiences are often discounted or ignored by providers who don't understand these conditions, resulting in delays in treatment and worsening of health outcomes. Further, the lack of understanding of rare diseases among government and program staff means that patients are often challenged to access needed support, even though they are eligible. There is a strong call for greater awareness and education about rare diseases, both within the medical community and among the public, to improve diagnoses and care pathways.

ADMINISTRATIVE HURDLES TO CARE

Patients and providers alike identified the burdensome administrative processes in place to access needed services and supports. Providers noted spending hours on paperwork and forms, taking time away from direct patient care. Patients and families noted that systems are often set up to gatekeep access rather than providing supportive care. These hurdles can be even worse for those who haven't yet received a diagnosis.

LACK OF CONNECTIONS BETWEEN RESEARCH AND TREATMENT

Rare disease treatments often face significant barriers in clinical trial development due to small patient populations, geographical dispersion, and limited research supports. Researchers, both in academia and industry, are often challenged to enroll sufficient patient numbers to gain approvals under the current regulatory and health technology assessment processes. Patients often struggle to identify clinical trial opportunities in the province, sometimes seeking out of country options instead. Clinicians in the province are interested in offering these clinical trials locally, and have the research capacity to do so, but they struggle to become clinical trial sites. Once back at home, access to treatments that patients may have had in an out-of-country trial can be limited.

GUIDING PRINCIPLES

Throughout the course of the consultations, a few key themes emerged, which should serve as foundational guiding principles for any action taken to improve rare disease care in the province. While not relevant for rare disease care alone, these were identified as being particularly important for a community that has experienced multiple challenges in accessing appropriate care and support services. These should be considered as part of any program or policy development that takes place.

1. **Co-designed solutions:** Those who directly experience, navigate, or deliver rare disease care provide invaluable personal and professional insights into care gaps, system inefficiencies, and opportunities for innovation. Without early and meaningful collaboration with these stakeholders, healthcare policies and interventions risk being disconnected from real-world needs, leading to inefficiencies and unfocused healthcare expenditures. Integrating co-design principles at the foundation of healthcare planning ensures that solutions are people-centered, operationally feasible, and resource-efficient, ultimately driving more effective and sustainable improvements across the healthcare system.

2. **Equity-focused care:** Given the challenges in care coordination, and difficulties in accessing treatments and support services, there are inequities across the province. System improvements must be designed with all Albertans in mind, ensuring that timely, coordinated, and needs-based care is accessible to all regardless of geography, location, socioeconomic status, or background.

3. **Innovative transformation:** There have been many advancements in rare disease treatments and care; however these don’t always reach patients and their families. Approaches to improving rare disease care in the province of Alberta require new partnerships and different ways of thinking about how to improve the system. By modernizing processes, embracing flexibility and fostering collaboration with industry, Alberta can not only establish itself as a national leader in rare disease care but also ensure that Albertans receive timely, equitable, and high-quality care that aligns with evidence-based best practices.

RECOMMENDATIONS & INSIGHTS

Several actionable strategies were identified for the Government of Alberta to enhance the care and support available for individuals affected by rare disease. These initiatives focus on building on Alberta's existing strengths and leveraging funding available through the National Strategy for Rare Diseases. These recommendations are grouped by overarching category with suggestions for short and longer-term actions.

RECOMMENDATION 1

Improve coordination of care for people living with rare disease

All stakeholders agreed that one of the biggest challenges continues to be the ability to navigate, organize, and deliver services for patients who are dealing with complex symptoms. Properly addressing this challenge will take time and resources, but there are short-term actions that could be initiated to help build capacity.

Establishing a comprehensive, publicly accessible directory of specialists with expertise in rare diseases would significantly improve care coordination and access to appropriate expertise. Such a directory could include detailed profiles of specialists, their areas of focus, and contact information, enabling both physicians and families to quickly identify and connect with the right providers. This resource would reduce the time spent navigating the healthcare system and expedite access to care.

In the longer term, there is a need to explore a centralized and collaborative hub model for rare disease patients and their families. Alberta has successfully built a cancer care system that is internationally recognized for the level of care provided, and the ease of navigation for patients and families. Alberta should make efforts with its partners to replicate aspects of this successful model for rare disease patients and their families.

Participants noted that the interdisciplinary hub model used at the two children's hospitals in the province could be expanded upon, with the need for similar hubs for adult care. These hubs should include specialty care, allied health, mental health and social support services and programs, and be well integrated with primary care. By fostering collaboration among patients, clinicians, researchers, and policymakers, these types of hubs could drive innovation and ensure comprehensive care.

Additionally, there is a need for dedicated navigators to help patients and families as they move through different aspects of their care journey. Costs saved from unnecessary duplication of services and unneeded system utilization could offset the costs necessary to fund the navigators and services required to support the expanded hubs.

Finally, Alberta has already demonstrated a commitment to improving the use of technology, including AI, to improve care for patients in the province. The province should examine ways to expand telehealth and other technologies to increase access to specialists. Given that most specialty clinics are located in Edmonton and Calgary, this could be particularly helpful for rare disease patients and their families who live in rural and remote communities.

RECOMMENDATION 2

Create navigation tools and supports for patients, physicians and care providers

For patients

Accessing specialists and health care treatments is not the only support that is required for patients and their families living with rare disease. Most patients who have a rare diagnosis don't have curative treatment options available and only supportive care is provided. As a result, other meaningful and accessible supports must be provided.

In the short-term, Alberta can review current programs such as the Family Support for Children with Disabilities (FSCD) and Persons with Developmental Disabilities (PDD) initiatives. The province should convene a committee of experts, including those with lived experience, to review the way the current programs are accessed, delivered, and administered, with an eye to improving care and making these valuable supports more accessible.

In the longer term, the province can work with patients, families and caregivers, program providers, and policymakers to implement the changes that were suggested by the committee, and increase coverage to better support patients and families.

For physicians and providers

Participants acknowledged challenges faced by clinicians, both in diagnosing and managing rare diseases. Many clinicians struggle with identifying the appropriate care pathways due to the complexity and rarity of these conditions, compounded by a lack of centralized information on specialists and treatment options specific to each disease.

In the short term, a comprehensive and accessible directory of specialists with expertise in rare diseases – whether locally, nationally, or even internationally – could be developed. Such a resource would help physicians quickly connect patients with the right experts, reducing delays in diagnosis and treatment. The iRARE centre, which now has a dedicated support person in Edmonton, has been created to help patients navigate the system. It could serve as a model for a similar approach for resources and supports for physicians.

In the longer-term, a centralized digital platform offering detailed, condition-specific guidelines and care pathways could empower clinicians with the knowledge to manage these conditions more effectively. This platform could be supported with decision-support tools developed by clinicians and researchers in either the Alberta rare disease hubs, or national networks of researchers and clinicians, such as the Canadian Rare Disease Network. By investing in navigation tools and support systems, the healthcare system can empower physicians to deliver more timely, accurate, and coordinated care for rare disease patients, ultimately improving outcomes and reducing the burden on all stakeholders.

RECOMMENDATION 3

Remove barriers to innovative and cutting-edge treatments

Screening and diagnosis testing

Alberta already has a strong diagnostic testing and screening system. However, challenges remain for patients in accessing these resources and in obtaining a rare disease diagnosis in a timely manner. This is particularly true for those whose symptoms arise in adolescence or adulthood. Alberta has the opportunity to continue to build on its existing system to reduce the time to diagnosis for rare disease patients and their families. The province of Alberta should look at what can be done in the community and invest in a larger framework for rare disease care. This will lead to better health outcomes for patients and reduce costs to the health care system through a reduction in health care utilization.

There is an opportunity to use funds provided through the National Strategy for Rare Diseases to work with key stakeholders in the province to identify ways to increase capacity and improve system efficiencies. Collaboration and information sharing with other similar jurisdictions is also encouraged to capitalize on work being done across the country.

In the longer term, investments in infrastructure, training, and patient supports, such as clinic geneticists and genetic counselors, will help build the foundation for a system that identifies patients early and gets them to the necessary supports.

Access to innovative treatments

Building a screening and diagnostic testing infrastructure that supports rare disease patients, is one way to ensure better access to innovative and cutting-edge treatments. However, there are other steps that the province can take to ensure greater access to these potentially life-saving medications.

In the short-term, the province should work with patients and providers in the province to identify the most meaningful way to spend the funds that have been allocated for rare disease treatments under the national strategy. As part of this work, the province can establish a process to ensure participation from patients, families, and clinicians in the drug listing processes.

Patients and their families have a wealth of experience and expertise in living with rare diseases. They can provide insight and support for decision making in a way that is unlike any other group. The government should use some of the funding available under other eligible expenses to establish a framework for decision-making about rare disease treatments and other support services where patients, families and caregivers are meaningful and engaged partners. This framework should be publicly available so that rare disease patients, families, and clinicians, and researchers understand how drug listing decisions are made.

In the short-to-medium term, the province should work to evolve the short-term exceptional drug therapy program (STEDT) into a Specialty High-Cost Drug Access Program with a clear, future-proof process for accessing medications, including those still emerging in development. This evolved program would integrate a more streamlined and transparent framework that prioritizes social and ethical considerations while addressing patient needs, ensuring equitable access to treatments, and supporting the healthcare system's ability to manage high-cost medications effectively. Bringing together key stakeholders to examine the drug access system would be an effective use of some of the federal funds, recognizing that the national strategy is not designed to have any impact on speeding up approval and reimbursement decisions.

In the medium term, there is an opportunity for Alberta to show national leadership by pioneering the use of innovative agreements for rare disease treatments. The province already has significant data capacity to undertake such work, and local experts and researchers such as the Institute of Health Economics are well placed to lead pilot work. The government should work with industry partners to identify and test agreements for some of the new and emerging rare disease products. This approach will help ensure greater access for patients, while allowing the government to meet financial obligations. Groundwork has already been laid for the launch of such an initiative.

Finally, the government of Alberta should use its leadership position among the provinces to advocate for faster timelines from agencies such as the Pan-Canadian Pharmaceutical Alliance to ensure that Alberta patients have access to necessary treatments. Reducing these delays would not only improve patient outcomes, but it would reduce health system costs and make Canada a more attractive destination for investment and product launches from industry.

RECOMMENDATION 4

Reduce red tape and administrative burden

One of the biggest challenges identified by rare disease clinicians was the administrative burden and the amount of paperwork required to get access to medications and supportive services for patients, including disability tax credits. This administrative work takes time away from patient care and further adds to increased wait times for specialty services and treatment options. It also contributes to clinician burnout.

It is recommended that the Minister of Red Tape Reduction convene a group of expert stakeholders to review the current forms and processes with the eye toward finding efficiencies and identifying ways to improve the process. Minimal funding would be required to support the committee of experts, and necessary system updates (i.e., incorporating forms in Connect Care), but no ongoing funding would be required. This foundational work would reduce the administrative burden on clinicians, freeing up more patient time and reducing wait times for specialty care.

In the longer term, utilizing AI tools to streamline administrative processes could free up valuable time for clinicians and healthcare providers, allowing them to focus more on patient care. AI applications could assist with automating routine tasks such as patient referrals, data entry, and billing, as well as supporting diagnostic processes by flagging potential rare disease cases for further investigation. This would not only improve efficiency but also help clinicians prioritize complex cases more effectively.

RECOMMENDATION 5

Increase access to clinical researchers, trials and patients

Alberta is home to world-leading clinicians and researchers focused on rare diseases. Additionally, the province has prioritized making Alberta a destination for the launch of industry clinical trials. Efforts to establish a better connection between clinical research and treatment in the province would allow more patients to access new and emerging therapies and make Alberta a more attractive destination for investment.

One major challenge in the rare disease research is the small patient populations for some conditions. This makes it difficult for researchers and industry to identify the right people to enroll and also creates challenges for patients in accessing trials as there are not always easily accessible resources about which trials are available. Some organizations, such as the MAGIC clinic in Calgary, have developed registries for patients to identify potential clinical trial participants, and the children's hospitals in the province have a program called Phenotips that enables them to find patients that have a specific disease and therefore recommend them for a trial. For patients and researchers, not affiliated with these programs, there are limited options for identifying trials outside of the US run clinicaltrials.org. The province should work with researchers and clinicians to expand and improve way-finding for clinical trial access.

In the longer term, the province could focus on enabling more clinical trials in the province and supporting efforts to expand clinical trials that address the needs of adult rare disease patients. Alberta researchers are already involved in multiple national programs aimed at supporting paediatric research, such as the National Paediatric Rare Disease and Clinical Trials and Treatment Network, and RareKids-CAN. Leveraging the researchers involved in these networks to launch trials in Alberta would strengthen research capacity in the province and ensure greater access to innovative treatments for Alberta patients.

While Alberta has a great deal of strength in paediatric clinical trials, adults often struggle to access to clinical research. Efforts to create a dedicated rare disease clinical trial network in Alberta for adult patients might help address this gap. Bringing together and fostering collaboration across research institutions, healthcare providers, and industry partners, could help address the challenges often faced with small patient populations. This network could centralize and coordinate clinical trials within the province, streamlining recruitment efforts and ensuring that patients have equitable access to participate. By integrating patient registries, genetic data, and health records, the network could efficiently match eligible participants to trials, overcoming one of the most significant hurdles in rare disease research.

Additionally, the network could support researchers by offering shared resources such as data analytics platforms, biobanking, and standardized protocols. For healthcare providers, the network would offer training and resources to better understand ongoing clinical trials, so they can discuss opportunities with their patients. For patients and families, it would provide clear pathways to participate in trials, giving them hope through access to emerging therapies while also contributing to the advancement of rare disease treatment as a whole. These efforts would not only accelerate the pace of clinical discovery but also ensure that Alberta remains a leader in rare disease innovation.

CONCLUSION

The insights and recommendations generated during the design lab underscore the transformative potential of collaboration between government, patients, families, caregivers, researchers, health care providers, and the life sciences industry. Each stakeholder brings unique strengths to the table—patients' expertise and lived experience, the health system's deep understanding of patient care, industry's capacity for innovation, and government's ability to enact policy and allocate resources. By aligning these strengths, the opportunity to create a more inclusive, efficient, and compassionate rare disease system is vast.

To enable that opportunity to emerge, significant effort must be assigned to breaking down functional, disciplinary, and knowledge silos, streamlining processes, and fostering a patient-centred approach. From improving navigation for patients and families to navigation tools for clinicians to enhancing access to innovative treatments, the recommendations offer actionable pathways to better outcomes. Most importantly, these solutions are not merely technical in nature; they reflect the lived experiences of patients and families, advocating for a system that meets their needs with dignity and respect.

Looking forward, there is a shared optimism that Alberta is well-positioned to be a leader in rare disease care by embracing collaboration, innovation, and co-design. Through continued partnership and commitment, a future can be built where Albertans with rare diseases are not just supported but empowered to lead healthier, fuller lives.

APPENDIX A

AGENDA - DAY 1

Rare Disease Pathway Development

TIME	ACTIVITY
08:15	Front Doors Open
09:00	Welcome and Land acknowledgement
09:15	Introduction To Design and each other
09:45	Discovery – Persona Reviews
10:30	Break
10:50	Discovery – Journey Map Review
11:40	Journey Map – Report Out
12:00	Lunch
12:45	Systemic Design – The Iceberg
13:30	The Iceberg – Report Out
14:00	Challenge Identification
14:30	Creative – The Analogy
15:00	BREAK
15:15	Creative – Ideation
15:45	Final Day 1 Reflection – Report Out
16:30	Closing

AGENDA - DAY 2

Rare Disease Pathway Development

TIME	ACTIVITY
08:15	Front Doors are open, breakfast and coffee
09:00	Welcome to Day 2 and Land Acknowledgement
09:10	Impromptu networking
09:20	Current State
09:50	Future State
10:30	BREAK
10:35	Ideal Future State
11:05	Ideal Current State
11:40	Effort/Impact Matrix
12:00	Lunch
12:45	Policy Canvas
13:45	Break
14:00	Presentation
14:20	Policy Canvas Report out
14:45	Closing

APPENDIX B

INTERVIEW PROTOCOL

Introduction script:

"Hello, (name)

My name is _________, and I am from the Pivot Group, and I will be the one interviewing with you today. I am working with Innovative Medicines Canada and Bio Alberta to engage in work around rare diseases within the province; to better understand the process, what works for patients/families/providers, and what does not work. Our first step in understanding the current state is to talk to those individuals who do engage with the rare disease system. You were one of the individuals identified as an expert to share with us the current state experience."

Confidentiality Notice

Our notes will be stored on a project platform that only our project team members have access to, and any identifying information will be removed. After the interview, we will take some time to compile our notes and then we will send them back to you so that you can correct any factual errors or add details that you feel are missing. Your participation is entirely voluntary, and you are free to revoke it at any time, and you also do not have to answer any questions you do not feel comfortable with. Do you have any questions or concerns regarding these confidentiality measures before we proceed?

INTERVIEW GUIDE: QUESTIONS

Patients/families/caregivers

1. Can you tell us a little about your condition and/or the condition of the person for whom you are providing care?
   a. How does it impact your daily life?
   b. What are the challenges you/they face as a result of the condition?
2. How long ago were you/they diagnosed?
3. Can you speak to the journey to diagnosis? How long did it take? What were the steps in the process? The challenges you/they faced?
4. What have been your experiences with clinical teams (i.e. primary care, specialty care etc.)?
5. Any barriers or opportunities you see for clinical teams to support you/they in the provision of care?
6. What has been your experience with care navigation in the province?
7. Is your/their condition one that has an available treatment option? If so, have you been able to access treatment?
8. What, if any barriers have you encountered in accessing treatment?
9. What other services/supports have you utilized or alternatively wish were available to support you/them in managing their/your disease?
10. What do you wish policymakers understood about living with rare disease?
11. What do you think are the most important changes necessary to support people living with rare disease in Alberta?

Clinicians/researchers

1. What area of rare disease treatment/research are you currently involved in? How did you get into this type of practice/research?
2. Can you describe briefly how a patient may come to be referred to you?
    a. Are there significant delays to access your clinic?
    b. Do patients see multiple specialists before they are referred to you?
3. What have been your experiences in terms of the diagnostic pathway in the province? From your experience, what does it look like for a typical patient
4. In your experience are there sufficient diagnostic/screening processes in place? If no, where are there needs for improvement?
5. Can you briefly describe the system of care for rare patients? Is there one?
6. Does your area of practice/research have available treatment options? If so, are they accessible for your patients?
7. What other services or supports do you think would benefit your patients or their caregivers?
8. What do you think policy-makers need to understand about the experience of patients living with rare disease?
9. What do you think are the most important changes necessary in order to support people living with rare disease in Alberta?

Government/program managers

1. Can you briefly describe your role?
2. How are you involved in the rare disease system in Alberta? What programs/policies do you oversee or support?
3. What do you see as the biggest challenges faced by the province in terms of providing care for these patients? i.e care navigation, HHR, costs of treatment etc.

Note all questions will not be asked - these are used as conversation starters, and the individual being interviewed will guide the process, and the interviewer will follow their lead

APPENDIX C

PERSONA: PATIENTS (years before diagnosis is not covered)

Goals and needs

- Access to timely, specialised care from doctors who understand their rare diseases (or who are willing to learn)
- Clear information on treatment options, trials, and emerging research
- Support network, both medical and emotional
- Flexible work accommodations to balance health and career.
- Access to treatment options, clinical trials and emerging research
- Improved Quality of life and mental health, and to be listened to.

Behaviours

- Frequently researches own condition but has unanswered questions
- Relies heavily on a network of online support groups depending where they are on their own journey (denial)
- Want to be the central decision maker of my own care team.
- Makes decisions don't always follow
- Does their own research

Frustrations + pain points

- Multiple appointments, that are months to years away
- Lack of clear communication between different specialists and primary care
- High-cost medication that is not always covered, or is delayed in being covered by either government or insurance
- Long wait times
- Sometimes needing to travel out of the country for necessary treatments or support
- There is more to the cost of these rare diseases, it affects everyone within the family, caregiving, spousal support, child support, missing work, need more support than just around treatment. (Deals with all aspects of the social determinants of health).
- Unclear treatment or access to treatment
- Limited access to my EMR
- Pain management
- Limited access to patient owned data – EMRs

Motivations

- Wants to live the best possible life
- Finding a healthcare team that understands their condition and advocates for their needs.
- Wants to be informed about new treatments or advancements

> **"It feels like I suffer every day, there are so many hurdles that I have to jump over, but I can barely make it through"**

PERSONA: CARE PARTNERS/FAMILIES

Goals and needs

- Awareness about the issues and challenges as a caregiver
- Being believed, listened to, and understood
- Want best care and quality of life for the person they care for
- Simple and easy ways to transition from pediatrics to adult
- Need to be heard and have coordinated care
- Need Treatments need easier access to treatments

Behaviours

- Giving their full time and support as a caregiver (Lack number of resources including funding that is needed, loss of personal life as you become a caregiver or take over a caregiver role)
- Primary advocate for the family members they care for
- Investigation and research on their own because resources and information isn't usually centralised or available
- Driving open conversations
- See a system that is reactive vs proactive

Frustrations + pain points

- Lack of awareness about specific diseases
- Being Listened too
- Outside of specific disease treatments, there are supports needed for mental health, social services, work/lifestyle management – Need funding for this
- Wanting to improve full quality of life, considering all of the knock-on effects people living with rare diseases face (i.e. mental health, social services, disability, physical health and exercise)
- Waiting times during the diagnosis phase creates so much fear and uncertainty – not enough human resources

Motivations

- Building relationships with people involved in the rare disease ecosystem
- Independence
- We need work to be acknowledged as an expert on the rare disease of the person they are caring for.
- Encouraging a more holistic approach to care delivery outside of direct symptom or treatment management: Primary care, paediatrician, neurologists, cardiologists, dentists, EENT, general surgeon, specialized clinics, mental health services which include counselling, psychiatric services in support and evaluations to get assessments done to access care. Educational supports, special ed teachers, aid support, assisted technology. Financial assistance piece, AISH, PDD, employment supports, specialized services. Occupational therapy, speech and behavioural support. community supports, specialized sports, day programs. (This is so well done at the alberta children's hospital but there is nothing for adults)
- Change the "disability" conversation
- To find treatments/diagnosis, access to treatments, anything to help their person.

PERSONA: PATIENT ADVOCATES

Goals and needs

- Bring to light what patients are going through, and support people they have met throughout their journey.
- Provide knowledge, resources, and information that isn't available through traditional or primary care network channels
- Bring overall general awareness to rare disease through their advocacy
- Trying to lead change but people need to listen

Behaviours

- Community building, influencing decision makers and policy
- Fundraising for sustainability of advocacy networks
- Time management, resiliency, stakeholder engagement
- Helping to guide patients through care journey
- Education is important lots of advocates are also volunteers
- Volunteer management

Frustrations + pain points

- Lack of data to understand incident rates or to identify patients that could benefit from interventions such as clinical trials
- Groups are not always well connected to organizations
- Frustrated with lack of supports, wayfinding, and information available to support people in the rare disease network.
- Wishes there was grant funding set up for patient advocacy organizations.

Motivations

- Moving the needle around awareness and advocacy
- Breaking through barriers for challenges that seem insurmountable
- Stakeholder engagement
- Treatments are an investment not a cost

"People need to realise when dealing with rare disease it's not going to be one solution, we need to come at it from different pillars, or targets. Understanding there needs to be more awareness about rare diseases. It's tough, there are many patients that haven't been diagnosed, but no one has any idea what the prevalence rate is. Public awareness, education, starting earlier with medical teams, if you think about medical training, put those people in front of us."

"You don't want to be diagnosed with a rare disease between the ages of 18 to 64, paediatrics we have coordinated care, and over 65 there are more supports"

PERSONA: CLINICIANS & RESEARCHERS

Goals and needs

- Developing a specialization through a meaningful career pathway
- Building awareness about their area of research and specialization
- Registries for access to potential patients for clinical trial research
- Administrative support to help use patient time more effectively
- Holistic support of the patient with referrals to the appropriate spaces

Behaviours

- Building relationships with referral networks – no real knowledge on who to refer to docs need education or clear pathways **
- Care, compassion, understanding of unique patient needs
- Over Communication with patients so that they fully understand their condition and treatment options – which is not paid for within the current remuneration models

Frustrations + pain points

- Stretched to capacity, can see a limited number of patients per year
- Siloed care provider teams
- Getting patients approved and covered for treatment is extremely difficult
- Data focused investigative work with no finish line, constant and ongoing
- Too much paperwork that is not covered in a fee for service model
- Inadequate awareness of the funding needed for research, clinical trials, direct treatments

Motivations

- Trying to do the best for patients
- Focusing on how to make patients live healthier happier lives throughout their health journey
- Building relationships with other specialists to ensure there is more awareness in the diagnostic pathway

> **"If we have to grade in levels of importance - drug coverage cost is a huge thing that I can't do anything about, or patients, it really comes down to government policy makers. Everything else is secondary to that."**

> **"I see a day in the future where newborn screening will be full genome sequencing. It's only about $300, cheaper than 80% of tests we do daily. The biggest problem is what we do with the data."**

> **"If someone has a rare disease, it can take years before they get a final diagnosis. From primary care to specialist, and possibly back to primary care or another specialist. These wait times add because of our lack of coordinated care"**

PERSONA: GOVERNMENT

Goals and needs

- Want to ensure timely access to primary care and other health services
- Reduce bureaucracy and operational silos
- Ensure effective use of health care resources
- Develop a system that is supportive of patients, families, and physicians.
- Continuity of care

Behaviours

- Reviewing health systems from across the globe
- Meeting with Albertans to hear their challenges with the healthcare system
- Moving from a single system to 4 operational pillars of care delivery
- Hear vs listen

Frustrations + pain points

- Outdated remuneration models Triage to find a way forward
- Overly bureaucratic system that delays access to care and access innovative medicine
- System change resistance while advocating for new and novel access points to healthcare (i.e. Nurse Practitioners
- Reduce/stop the increase in budgets for the healthcare system while still meeting patient needs. Better value for money

Motivations

- Want Albertans to have access to the most innovative medicine – I don't think they do
- Ensuring Albertans are getting value for their tax dollars within the healthcare system
- Improved Access for rural Albertans
- Faster access to treatment, before a progressive disease puts them in hospital
- Want to reduce administrative burden and red tape

PERSONA: INDUSTRY PARTNER

Goals and needs

- Develop innovative treatments (health solutions) for rare disease/repurpose drugs
- Build partnerships within the Alberta health care system and with patients and clinicians
- Expand clinical research and early diagnostic programs (increased screening) (There is no or limited funding available)
- Opportunities for clinical trials and development of centres of excellence – more access to clinical trials (funding is an issue)
- Decreased outcome measures
- Increase family representation and patient representation
- Increase access to treatments
- Advocacy for better health solutions for patients.

Behaviours

- Meeting with the government and generating pathways to care. (need to involve other stakeholders)
- Identifying Federal funding that can be leveraged for improved provincial healthcare
- Advocacy for improved access to rare disease treatments and system navigation
- Investing in assistance programs that help with systems navigation and medication coverage (clinicians needs resources to help with this)
- Measurement of outcomes needs to change

Frustrations + pain points

- Balancing Government, insurance, pharmaceutical, patient healthcare providers and health system need
- Bureaucratic system that complicates processes and slows access to innovative medicine
- High costs for developing rare disease treatments, which decreases access to treatments
- Limited patient populations for clinical trials, hard to find and engage in Canada and not organized in a way that is easy to access
- Lack of genetic screening
- Lack of payer-HTA for rare disease

Motivations

- Desire to be (a trusted partner for innovative solutions) at the front of innovative treatments that make an impact on patient lives
- Strengthening industry reputation
- Many engage with families outside of the box – increase ability to do so
- Newborn screen, could industry push for that?
- Bring people together for better collaboration.

"We have patient support programs, and we will bridge you, provide the drug to you for free for 3 months until we can ensure coverage."

APPENDIX D

ABOUT THIS JOURNEY MAP

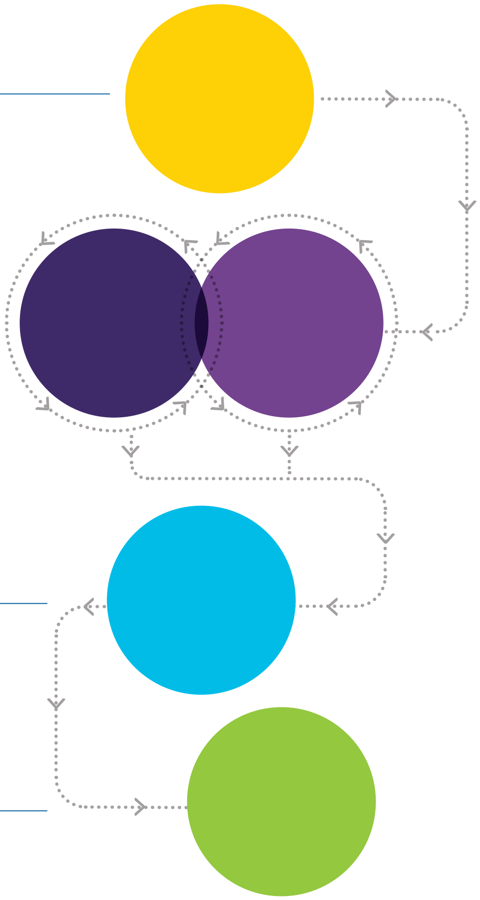
This journey map provides a high-level overview of what someone might experience when navigating a rare disease. While it outlines common stages, it's important to note that every individual's journey is unique. Not everyone will go through each stage, and many may face delays, roadblocks, or detours along the way.

The overlapping circles in the middle reflect the reality that many people move back and forth between primary care and specialist care—sometimes repeatedly—without receiving a diagnosis or clear path forward. These transitions are rarely linear and often involve uncertainty and waiting.

The stages highlighted in this image include:

- **AWARENESS**: When symptoms first begin.
- **SEEKING HELP**: Starting with initial screenings and, when possible, connecting with specialists
- **UNDERSTANDING OPTIONS**: Receiving a diagnosis and exploring possible treatment paths.

The pages that follow break down each stage in more detail, providing insight into the steps and experiences that people living with rare diseases may encounter as they seek answers, support, and care.



STAGE 1: AWARENESS

SYMPTOMS START

STAGE 2 & 3: SEEKING HELP

INITIAL SCREENING
SPECIALISTS

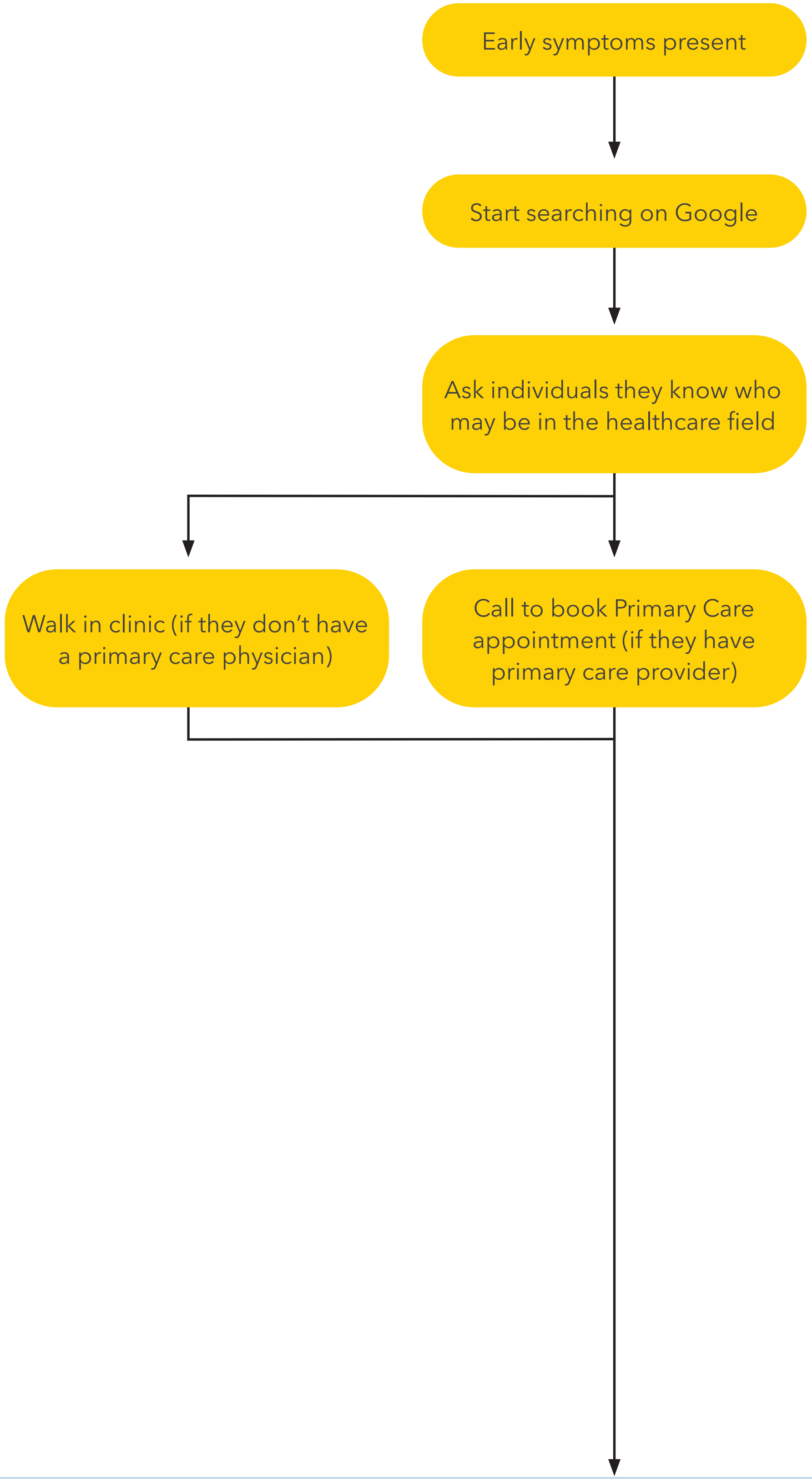
STAGE 4: UNDERSTANDING OPTIONS

DIAGNOSIS

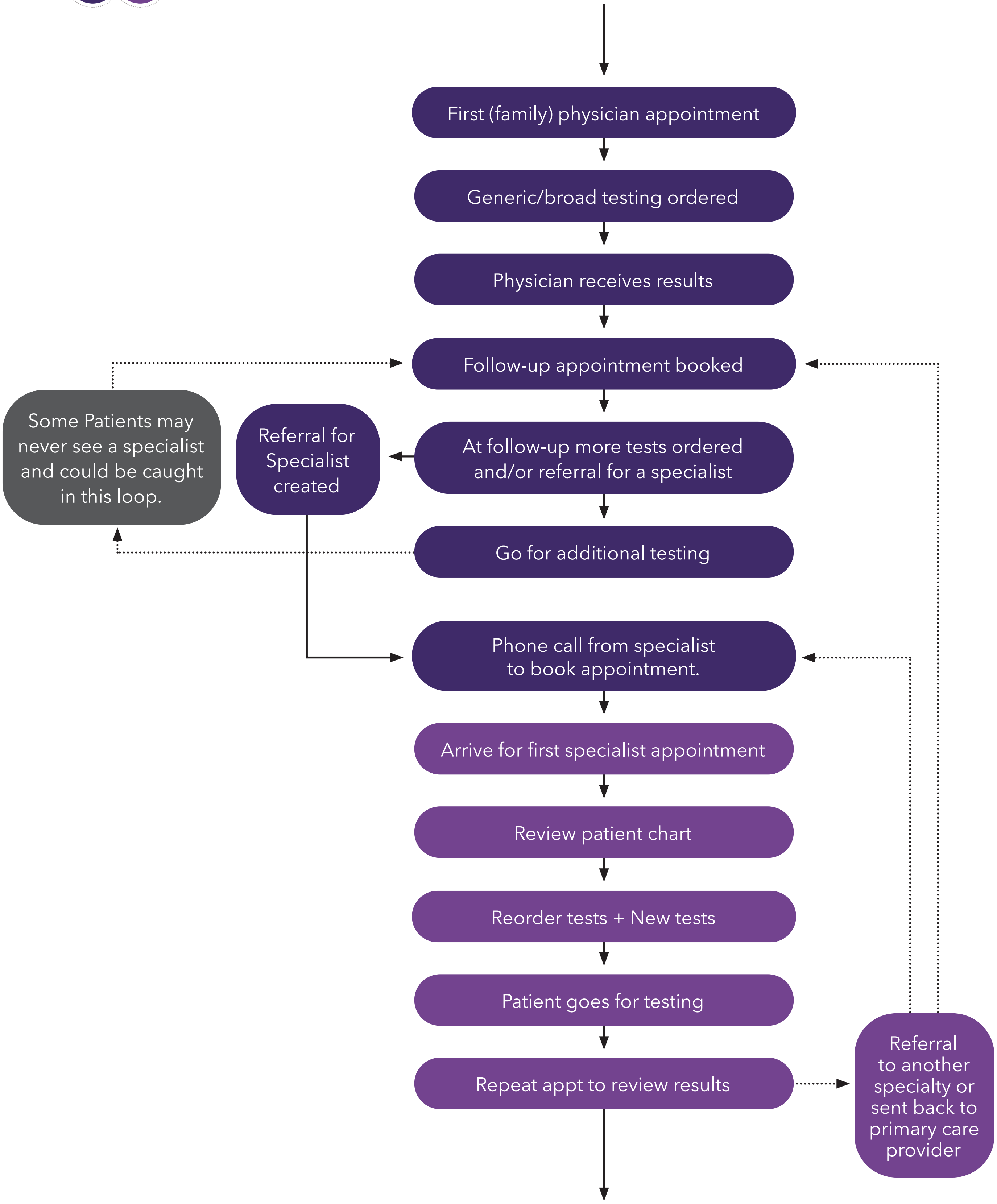
STAGE 4: UNDERSTANDING OPTIONS

TREATMENT

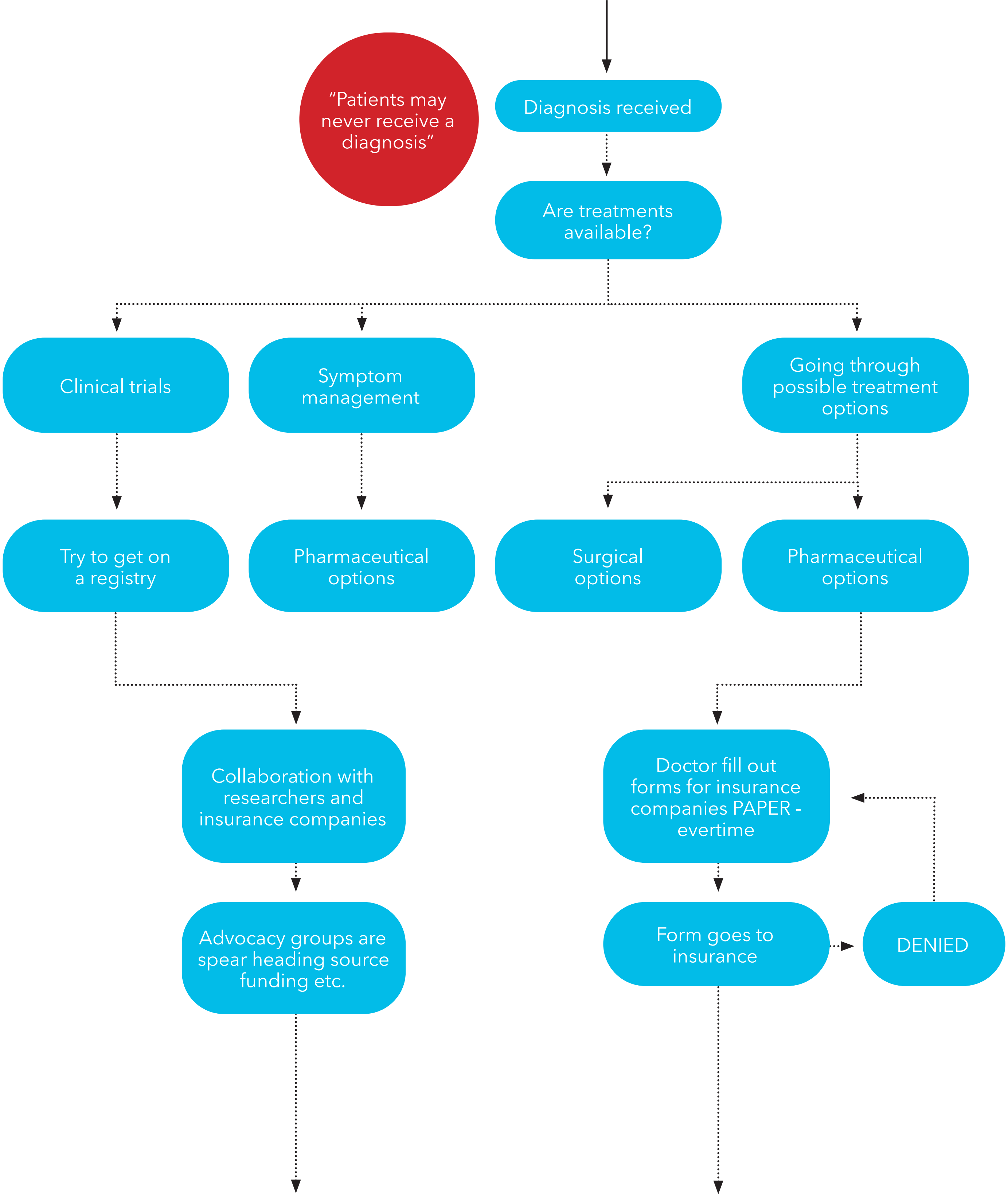
STAGE 1 - AWARENESS



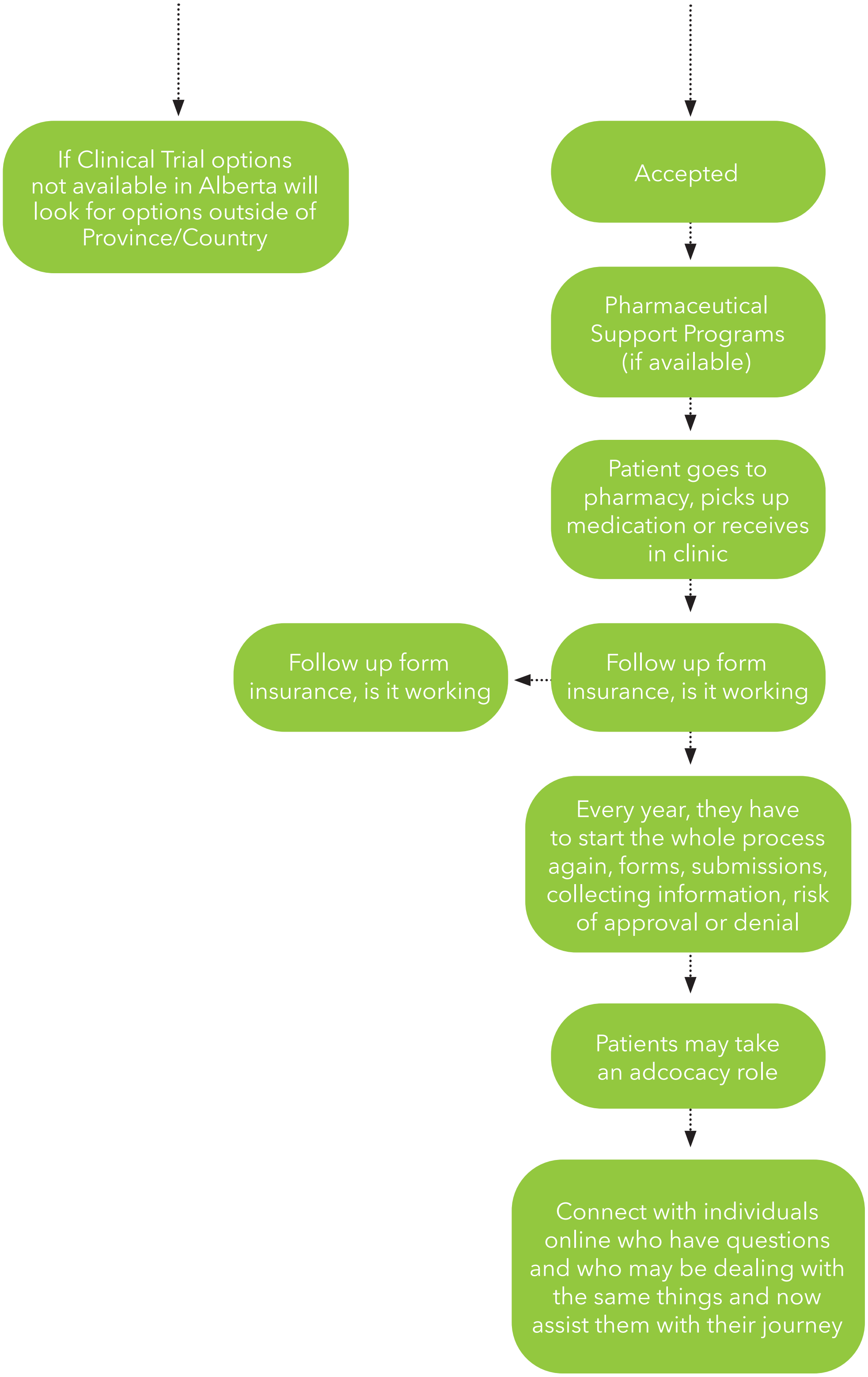
STAGE 2 &3 - SEEKING HELP



STAGE 4 - UNDERSTANDING OPTIONS



STAGE 5 - NEW NORMAL



If Clinical Trial options not available in Alberta will look for options outside of Province/Country

Accepted

Pharmaceutical Support Programs (if available)

Patient goes to pharmacy, picks up medication or receives in clinic

Follow up form insurance, is it working

Follow up form insurance, is it working

Every year, they have to start the whole process again, forms, submissions, collecting information, risk of approval or denial

Patients may take an adcocacy role

Connect with individuals online who have questions and who may be dealing with the same things and now assist them with their journey

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