RARE DISEASES

COMMITTED STAKEHOLDERS, DETERMINED TO WORK WITH THE GOVERNMENT TOWARDS A QUÉBEC POLICY FOR RARE DISEASES.

Montréal InVivo's Project Committee on Rare Diseases.

- SEE NOTE TO READER ON PAGE 2 -

NOTE TO READER

This brief is the result of work carried out since 2019 by Montreal InVivo and its Rare Disease Project Committee. This working group brings together several stakeholders including patient groups, clinicians, researchers, representatives of paragovernmental organisations and industry representatives.

The objective of this strategic plan is to **inform** and **inspire** the Quebec government to act quickly and seize the opportunity to be a pioneer in Canada in the field of rare diseases. Our guiding principles have been to place the patient and the economic development of Quebec at the heart of our thinking. Inspired by **international best practices**, the concrete recommendations proposed are complementary to and respectful of the future Quebec Policy on Rare Diseases and are in line with the Quebec Life Sciences Strategy (QLS) and the Quebec Research and Innovation Strategy (QRIS).

Montréal InVivo’s involvement in the area of rare diseases

In early 2019: creation of the Québec Working Group on Rare Diseases, mandated by the Ministère de la santé et des services sociaux to issue recommendations on organizing services to manage rare diseases.

In addition, Montréal InVivo is creating a Specialized Project Committee on Rare Diseases representing the entire ecosystem, to propose a national plan that is well aligned with the issues in the field.

This Committee’s objectives are to:

- Allow Québec patients suffering from a rare disease to have rapid and optimal access to therapeutic and diagnostic innovations;
- Improve the competitiveness of Québec's innovation ecosystem to attract investment and develop our skills by drawing on international best practices.

Work has been underway since 2019

Members of the Project Committee on Rare Diseases

President of the Project Committee

Thérèse Gagnon-Kugler, PhD – Vertex Pharmaceuticals (Canada) Inc.

Members of the Project Committee

Frédéric Alberro – Médicaments novateurs Canada
Hermance Beaud – CERMO-FC
Frank Béraud - Montréal InVivo*
Sylvain Bussière – Biogen Canada*
Sophie Bernard, MD, PhD – IRCM
Marie-Hélène Bolduc, MBA – Dystrophie musculaire Canada
Diane Brisson – Ecogene 21
Hélène Delerue-Vidot – ESG-UQAM
James Doyle, PhD – Modelis
Mira Francis, PhD – Alexion
Angela Genge, MD, FRCPC – NEURO
Diana Iglesias, PhD – Génome Québec
Olivier Jérome – CATALIS Québec*

Members of the Project Committee (continued)

Denis Laflamme – Takeda Canada*
Mélanie Langelier – IR CUSM
Norman MacIsaac – SLA Québec
Jacques L. Michaud, MD – Centre Québécois de Génomique Clinique
Vincent Mooser, MD – McGill
Gail Ouellette, PhD - Regroupement québécois des maladies orphelines
Nicolas Pilon, PhD – CERMO-FC
Vincent Raymond – Pfizer Canada*
Donald Vinh, MD, FRCPC, FACP – IR CUSM

*Membres du Comité de travail

A multi-disciplinary committee representing the entire ecosystem

We would also like to thank all the external contributors of this Committee for their involvement in the project.

Introduction

RARE DISEASES AFFECT MORE QUEBECERS THAN WE THINK.

6 to 8%

OF QUEBECERS ARE AFFECTED BY A RARE DISEASE.

690 000

This represents up to families affected by rare diseases in the province.

We all know someone affected by a rare disease... it's time to take action!

7 000 - 8 000
rare diseases in Québec

75%
begin in childhood

Main issues identified by the Project Committee

Care provided to patients

is sub-optimal and complex

Limited level of funding

for competitive research and development activities

Concerns about the ability to pay

for new value-added innovations

Business environment

Is significantly less competitive

Towards a Québec Policy for Rare Diseases
Possible concrete actions

1 **Accelerate diagnosis**

in order to limit diagnostic delays

2 **Improve care**

for patients' health

3 **Aggregate health data**

to optimize care pathways and research opportunities

4 **Support the development of innovations**

In Québec

5 **Attract more clinical studies**

to improve patient care and create economic wealth

6 **Improve access to approved treatments**

so that Québec patients benefit quickly from innovations

7 **Create a public-private fund**

dedicated to rare diseases

1 Accelerate diagnosis

in order to limit diagnostic delays



Recommendations

1.1 **Facilitate access to genetic testing to speed up diagnosis**

1.2 **Offer whole genome sequencing tests to all individuals without a diagnosis**

1.3 **Be proactive in updating the Programme québécois de dépistage néonatal (Québec newborn screening program) and immediately add rare diseases for which a treatment is available**

2 Improve care

for patients’ health

Recommendations

2.1 Create specialized care centres and centres of expertise in the regions that include multidisciplinary expertise

2.2 a) Develop the necessary expertise and b) increase resources to manage patients' health

2.3 Increase resources to improve the quality of life of people with rare diseases and ensure equity in medical and paramedical care

2.4 Develop and regularly update a directory of expertise in rare diseases

3 Aggregate health data

to optimize care pathways and research opportunities

Recommendations

3.1 **Create a registry that consolidates health data on rare diseases and facilitates access to it**

3.2 **Develop the Québec reference genome and multi-genomics profile**

4 Support the development of innovations

In Québec

Recommendations

4.1 Stimulate collaboration to increase innovation development and investment attraction

4.2 Increase funding for rare disease research and provide incentives to attract the next generation of talent

4.3 Support expertise on rare diseases more specific to Québec to develop distinctive niches of excellence

4.4 Support the development of innovations by SMEs in the field of rare diseases

5 Leverage clinical trials

to improve patient care and create economic wealth

Recommendations

5.1 Position clinical research in the care trajectories for rare diseases

5.2 Attract more clinical studies in rare diseases by providing incentives

6 Improve access to approved treatments

so that Québec patients benefit quickly from innovations

Recommendations

6.1 Make representations to the Canadian government to optimize Health Canada's evaluation processes

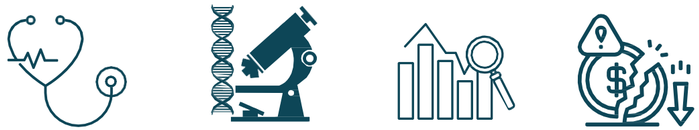
6.2 Supporting INESSS (Institut national d’excellence en santé et en services sociaux) assessment improvement processes for access to treatment

6.3 Make innovations accessible as soon as their therapeutic value is recognized by the INESSS for orphan diseases and rare diseases with unmet health needs

6.4 Implement innovative reimbursement arrangements

7 Create a public-private fund

dedicated to rare diseases



Recommendations

7.1 Make representations to the Canadian government so that the Québec government receives its share of the future federal program on rare diseases

7.2 Evaluate the existing Québec government support for rare diseases

7.3 Create a public-private support fund dedicated to rare diseases to leverage federal investment

Proposed schedule

The realization of the recommendations and the identification of the stakeholders involved

2022-2023	2023-2024	2024-2025
Lancement des activités de la table de concertation sur les maladies rares (MIV, parties prenantes)	**2.2b** Recruter plus de professionnels de la santé pour optimiser la prise en charge des patients (Gouv)	**2.1** Créer des centres de soins spécialisés et des Centres d'expertise en région qui incluent une multidisciplinarité d'expertise (Gouv)
1.3 Bonifier le programme de dépistage néonatal (gouv Qc, INESSS, RSSS)	**3.1** Créer un registre de données de santé québécois pour les maladies rares (Gouv, parties prenantes)	1.Faciliter l'accès aux tests génétiques pour accélérer le diagnostic (Gouv)
2.a Former plus de professionnels de la santé pour optimiser la prise en charge des patients (RQMO, MIV, Universités, Gouv)	**4.4** Mettre en place une série d'incitatifs concurrentiels pour soutenir l'innovations des PMEs (MEI)	2.Tests séquençage génome complet à toutes personnes soupçonnées d'avoir une maladie rare et leur famille (Gouv)
3.Développer et mettre à jour régulièrement un répertoire d'expertises (RQMO)	**5.1** Positionner la recherche clinique dans les trajectoires de soins pour les maladies rares (Gouv, Catalis et parties prenantes)	**3.2** Développer le génome de référence pédiatrique québécois (Gouv, FRQ-S, GQ, IRSC, Universités, Parties prenantes)
5.2 Attirer plus d'investissements privés en recherche clinique grâce à des incitatifs concurrentiels (Gouv, parties prenantes)	3.Rendre accessibles les innovations dès la reconnaissance de la valeur thérapeutique par l'INESSS et ajuster les prix à posteriori (Gouv. du Qc et Manufacturiers)	2.Augmenter le financement de la recherche en maladies rares (Gouv, FRQ-S, GQ, IRSC, Universités, Parties prenantes)
6.1 Faire des représentations auprès du fédéral afin accélérer les processus de Santé Canada (Gouv et parties prenantes)	4.Mettre en place des ententes de remboursement innovantes (Gouv et Manufacturiers)	3.Développer des créneaux d'excellence distinctifs sur les maladies rares plus communes au Québec (Gouv, FRQ-S, GQ, IRSC, Universités, Parties prenantes)
1.Faire des représentations auprès du fédéral pour récupérer la part du Qc au programme fédéral sur les maladies rares (Gouv Qc, parties prenantes)		**7.3** Créer un fonds de soutien public-privé dédié aux maladies rares (Toutes les parties prenantes)
2.Évaluer la valeur du soutien actuel du Qc pour les maladies rares (Gouv)		

Maintenir et renforcer les acquis: 4.1 Stimuler la collaboration entre les parties prenantes; **6.2** Soutenir l'amélioration des processus d'évaluation de l'INESSS

Conditions for success and conclusion

Conditions for success and conclusion

A national plan for rare diseases will require:

- A real willingness to open a dialogue and collaborate with the provinces and the different stakeholders of the rare disease ecosystem (patients, researchers, clinicians and companies);
- Build on international best practices to create a predictable and consistent regulatory framework with other federal initiatives;
- Innovate and implement a new financing mechanism to improve the entire ecosystem’s competitiveness.

Conclusion

The urgency to act demonstrated by the numbers and the mobilization of life sciences organizations leads us to this strategic action plan with recommendations in seven (7) major themes.

Canada is lagging behind. Québec can become a pioneer.

For this to happen, we must :

- Leverage the stakeholder players' very strong mobilization;
- Ensure that Québec takes the Canadian leadership in rare diseases and implements the proposed recommendations.
- More than just a policy

What Québec patients and their families need is an integrated plan for rare diseases articulated around the current major issues!

Our partners

LOOKING FORWARD TO THE DISCUSSION!

Frank Béraud
Président-directeur général
Montréal InVivo
fberaud@montreal-invivo.com