Quebec Policy and Action Plan for Rare Diseases

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Disclosure - I have nothing to disclose

Within the last 24 months, I have not had any type of affiliation, financial or otherwise, with a for-profit or non-for-profit organization that may have a connection to the content of my presentation and would require disclosure.

Presentation plan

1. Context on rare diseases in Quebec
2. Policy and action plan; the main points
3. Conclusion and what is next

CONTEXT

A little historic

2011 ||| 2018 2019 2020 2021 2022 2023 2024

- 2011: Publication du rapport de l'INESSS *Prise en charge des maladies rares*
- 2018: Création du Groupe de travail québécois sur les maladies rares **(GTQMR).**
- 2019: Le **GTQMR** remet au MSSS un rapport comportant plusieurs recommandations.
- 2020: Mise en place du Comité consultatif québécois pour les maladies rares **(CCQMR) (première rencontre)**
- 2022: **Consultation organismes en maladies rares**
- **Publication de la Politique québécoise pour les maladies rares**

POUR UNE MEILLEURE RECONNAISSANCE ET PRISE EN CHARGE DES PERSONNES ATTEINTES DE MALADIES RARES

- **Publication du Plan d'action québécois pour les maladies rares 2023-2027**

POUR UNE MEILLEURE RECONNAISSANCE ET PRISE EN CHARGE DES PERSONNES ATTEINTES DE MALADIES RARES

Début de la mise en œuvre →

Consultation with rare disease organizations

What we heard

- The challenges

Diagnostic and medical odyssey

Multiple consultations without results

Inequity of access

Lack of coordination of care

Few therapeutic options, or access to clinical studies

Non-fluid pediatric-adult transition

Psychological distress, psychiatricization, little support

Consultation with rare disease organizations
What we heard

- Why these challenges?

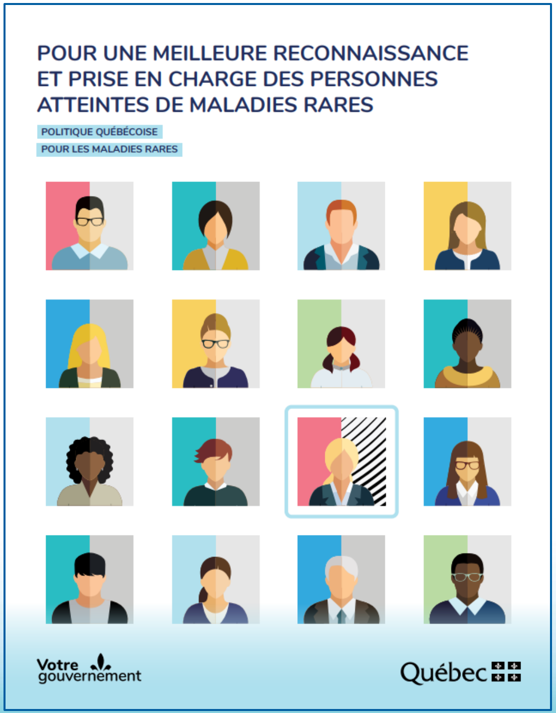
- **Lack of knowledge** of rare diseases among doctors and health care professionals
- **Limited resources,** particularly in distant regions
- **Lack of standardization** of care
- **Lack of structure** in the organization of care and services – **absence of a continuum** of care
- **Lack of synergy** between levels of care
- **Lack of research** and **innovation**

Policy and Action Plan

How to enhance our efforts to improve the health of people with rare diseases?

- 2022: Publication of the **Quebec Policy for Rare Diseases**
- 2023: Publication of the **Quebec Action Plan on Rare Diseases 2023-2027**

“For better recognition and care of people with rare diseases.”



POUR UNE MEILLEURE RECONNAISSANCE ET PRISE EN CHARGE DES PERSONNES ATTEINTES DE MALADIES RARES

POLITIQUE QUÉBÉCOISE
POUR LES MALADIES RARES

Votre gouvernement

Québec

POUR UNE MEILLEURE RECONNAISSANCE ET PRISE EN CHARGE DES PERSONNES ATTEINTES DE MALADIES RARES

PLAN D'ACTION QUÉBÉCOIS
SUR LES MALADIES RARES
2023-2027

Votre gouvernement

Québec

Votre gouvernement

Québec

What are rare diseases for the MSSS?

- Definition adopted by the Ministry of Health and Social Services of Quebec:

Disease affecting less than 1/2000 person

Other figures recognized by the MSSS

- Between **5000** and **8000** rare diseases listed (6417 on Orphanet)
- Up to **8%** of the population is affected (1/12 in Canada)
- **80%** are genetic diseases
- They also include autoimmune, infectious, toxic diseases, etc.

Approximately 700,000 Quebecers

Intervention Axes

01

1. AWARENESS AND TRAINING

02

2. FACILITATED AND EQUITABLE ACCESS TO DIAGNOSIS, CARE, AND SERVICES

03

3. PROMOTION OF RESEARCH, INNOVATION, AND DATA COLLECTION

Policy and Action Plan

Axis

01

1. AWARENESS AND TRAINING

4 objectives

- **1.1** Raise awareness and encourage knowledge transfer
- **1.2** Improve training for clinicians and health care professionals
- **1.3** Support the development of clinical tools that can guarantee the quality of care
- **1.4** Facilitate access to information

8 actions

Policy and Action Plan

Axis

01

1. AWARENESS AND TRAINING

8 actions

Notably:

- Conducting awareness activities,
- Involvement of patient partners and organizations in reference centers and in the continuum of care (defining needs, promoting community services, etc.)
- Creation of training modules;
- Establishment of a community of practice;
- Development of clinical tools
- Develop a web component to centralize information
- …

Policy and Action Plan

Axis

02

2. FACILITATED AND EQUITABLE ACCESS TO DIAGNOSIS, CARE, AND SERVICES

4 objectives

- **2.1** Continue the **improvement and development of screening and diagnosis** of rare diseases by reducing the time needed to access a diagnosis.
- **2.2 Organize and structure care and services**, ensuring that access to services and their coordination are improved.
- **2.3** Improve access to care for people with a rare disease causing **oral health consequences**.
- **2.4** Continue efforts to improve access to care and **pharmaceutical services**.

20 actions + sub-actions

Policy and Action Plan

Axis
02

2. FACILITATED AND EQUITABLE ACCESS TO DIAGNOSIS, CARE, AND SERVICES

20 actions + sub-actions

Notably:

- Cover preimplantation genetic tests ✦
- Reduce waiting times for preconception genetic counseling and prenatal diagnosis services
  - Increase recruitment in university programs
- Support the evolution of the prenatal screening program and the neonatal screening program ✦
- Support the development of genomic medicine in Quebec (Quebec Network for Molecular Diagnostics) ✦
- Designate reference centers and regional competence centers ✦
- Implement a National Clinical Navigation Network for Rare Diseases (Navi-Nat) ✦
- Support the autonomy of health professionals in the field of rare diseases
- Establish an oral care program for rare diseases
- …

Policy and Action Plan

Axis

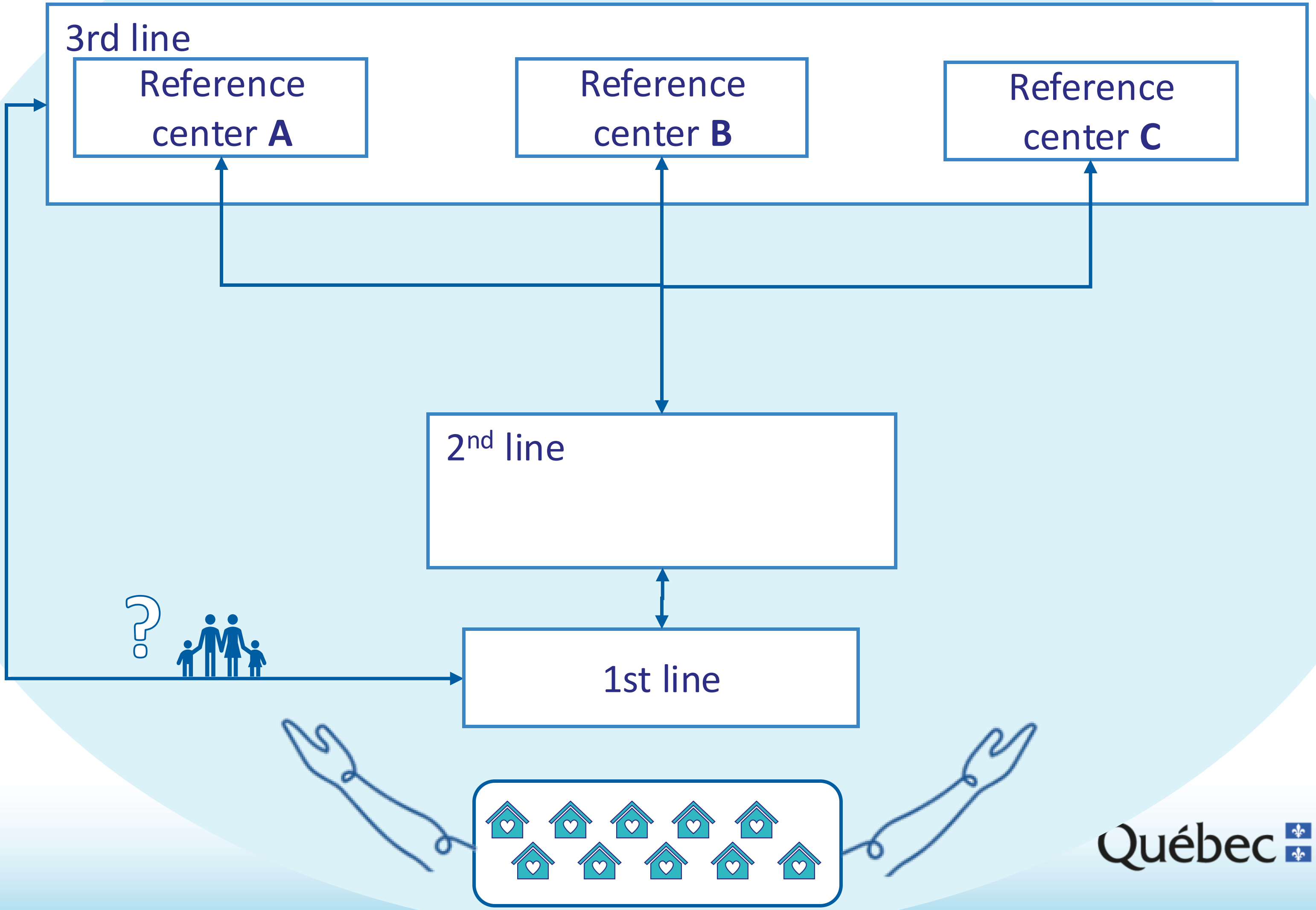
02

Légende:

Already existing element

Upcoming elements

🏠 Organizations, patient and parent associations, support groups for rare diseases.



3rd line

Reference center **A**

Reference center **B**

Reference center **C**

2nd line

1st line

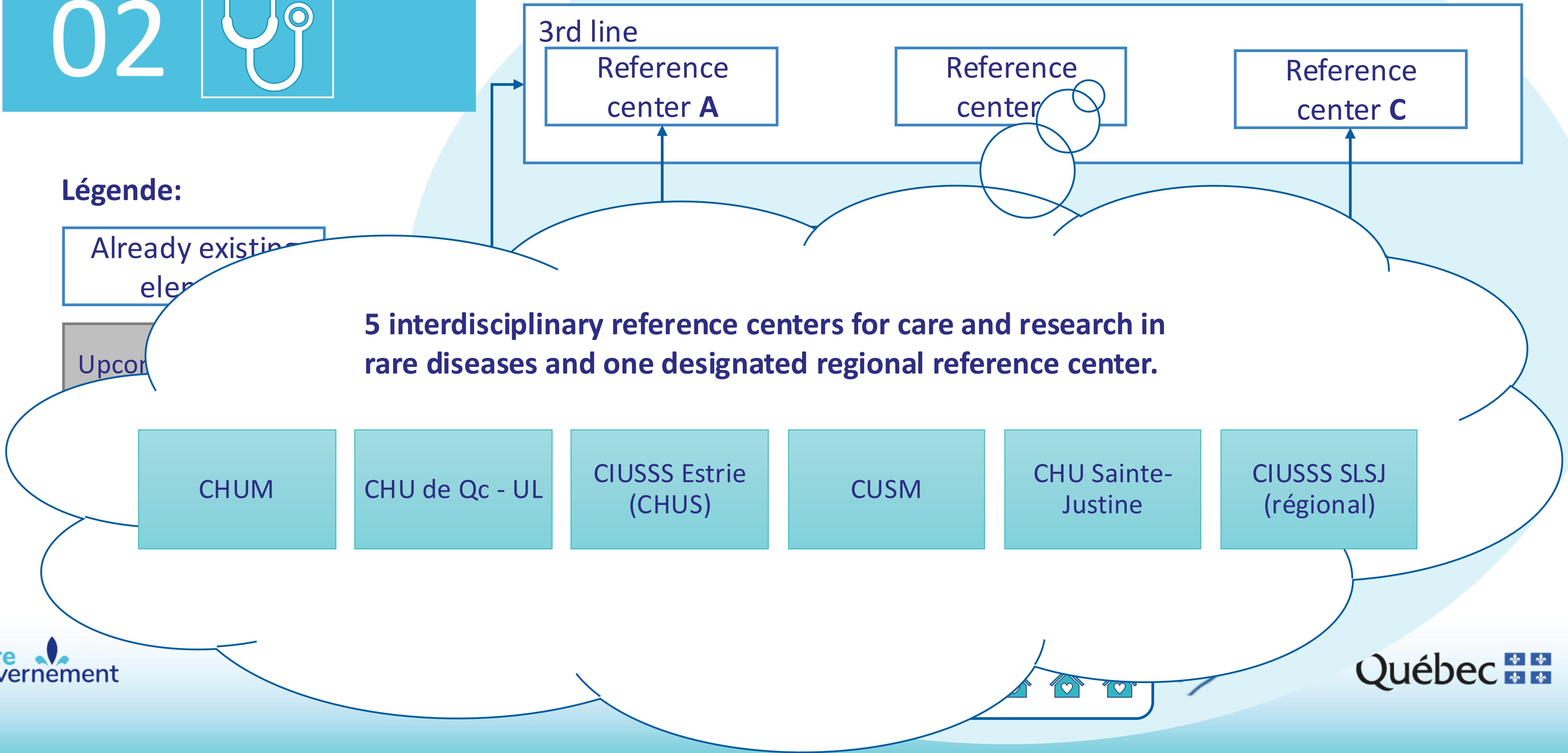
Policy and Action Plan

Axis
02

Légende:

Already existin
ele

Upcor



3rd line

Reference center **A**

Reference center

Reference center **C**

5 interdisciplinary reference centers for care and research in rare diseases and one designated regional reference center.

CHUM	CHU de Qc - UL	CIUSSS Estrie (CHUS)	CUSM	CHU Sainte-Justine	CIUSSS SLSJ (régional)

Votre gouvernement

Québec

Policy and Action Plan

Axis

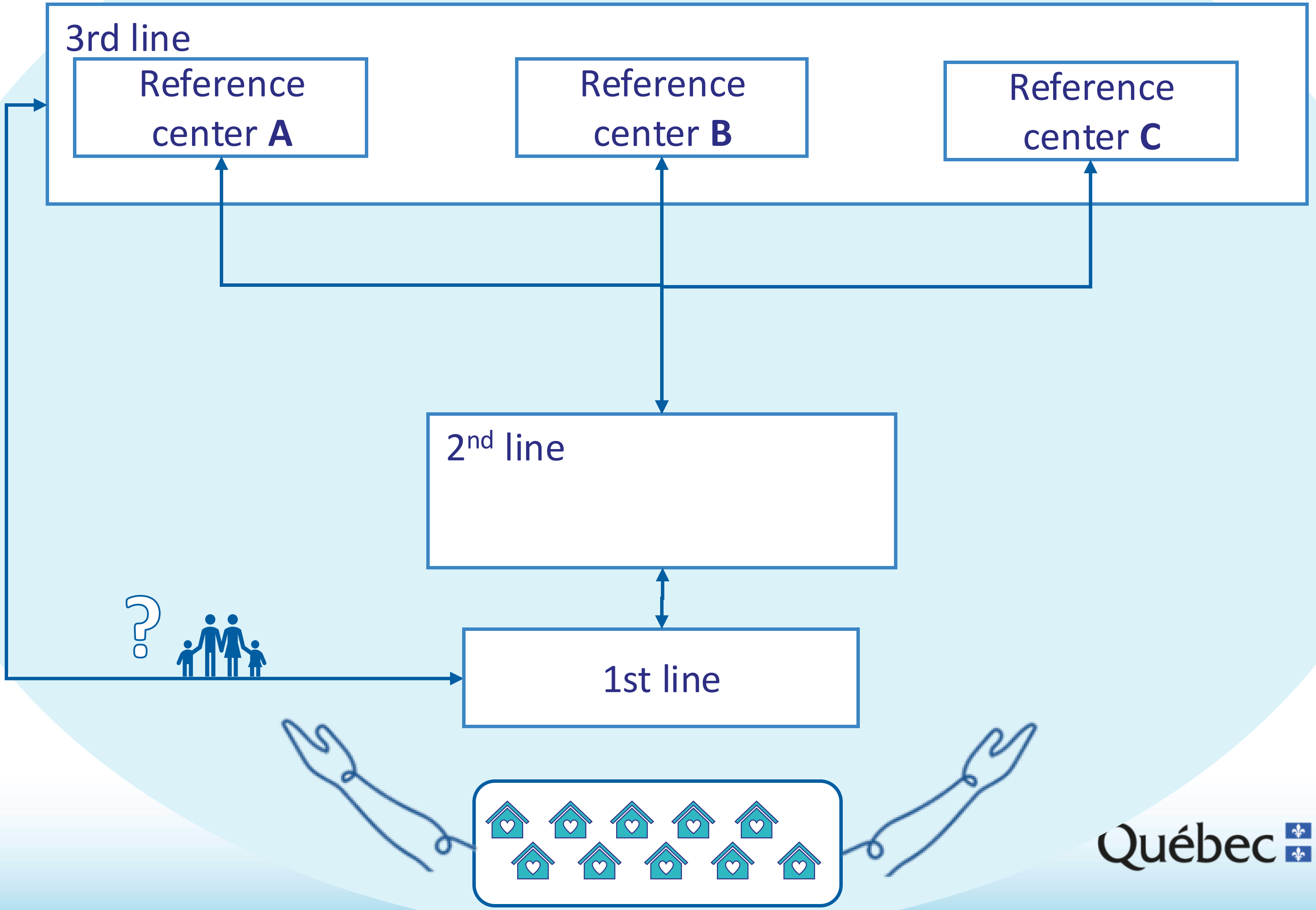
02

Légende:

Already existing element

Upcoming elements

Organizations, patient and parent associations, support groups for rare diseases.



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Reference center **A**

Reference center **B**

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2nd line

1st line

Policy and Action Plan

Axe

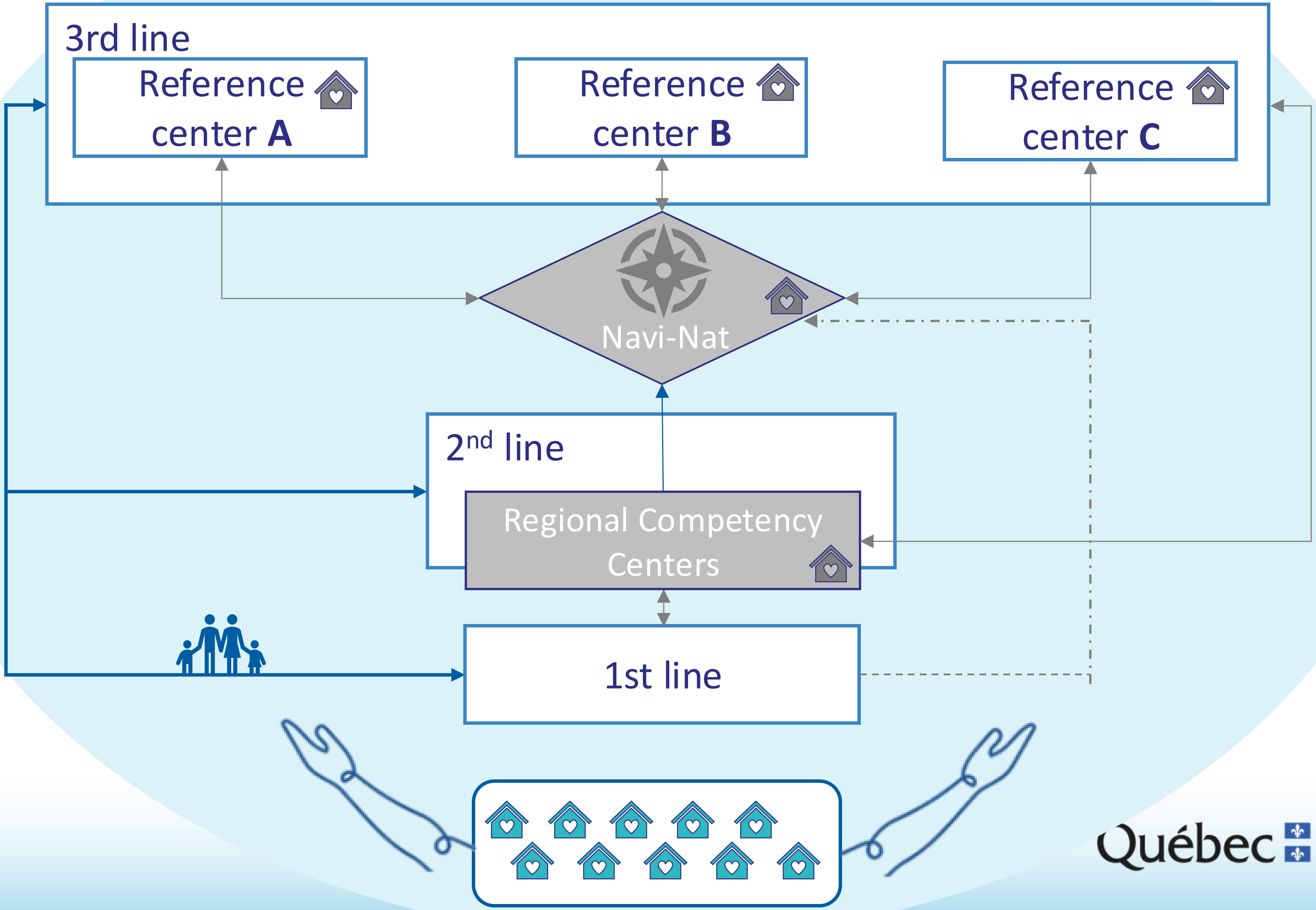
02

Légende:

Already existing element

Upcoming elements

Organizations, patient and parent associations, support groups for rare diseases.



3rd line

Reference center **A**

Reference center **B**

Reference center **C**

Navi-Nat

2nd line

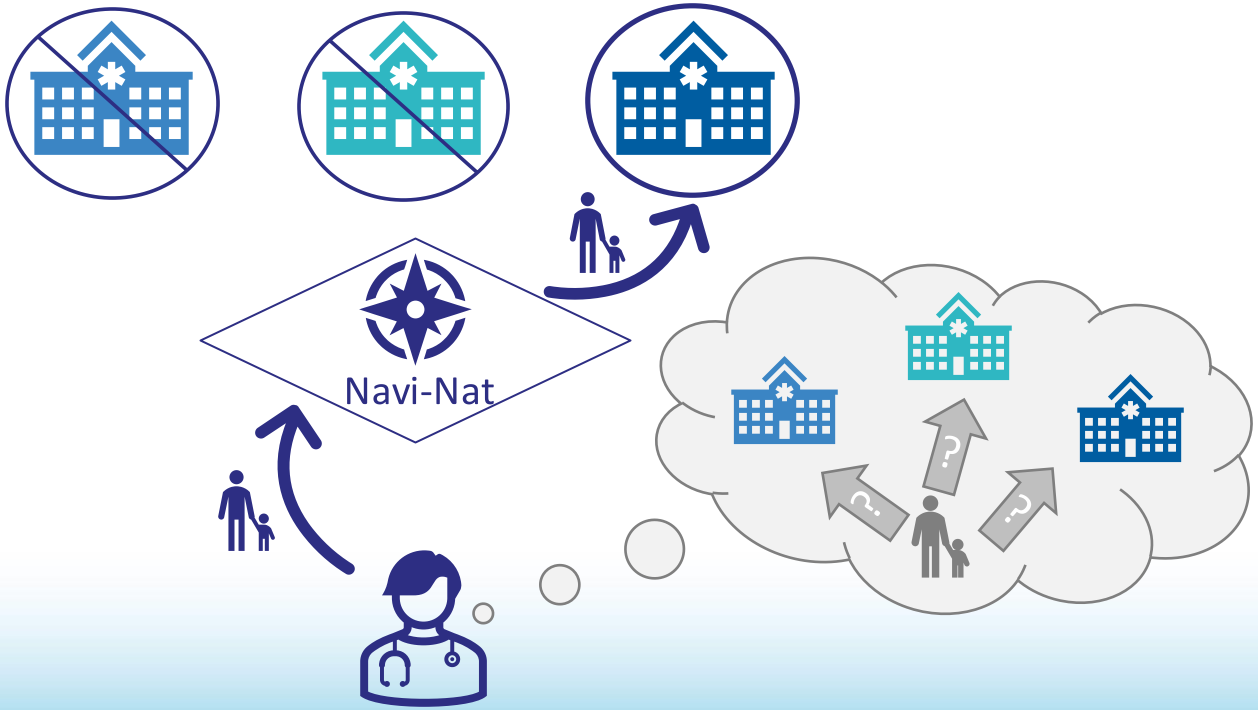
Regional Competency Centers

1st line

Axis

02

Upcoming creation of the National Clinical Navigation Network for Rare Diseases (Navi-Nat), an innovative and unique project in Canada



Policy and Action Plan

Axis
02

Upcoming creation of the National Clinical Navigation Network for Rare Diseases (Navi-Nat), an innovative and unique project in Canada

One of the first step:

Mapping of all care and service offerings for rare diseases realized in summer 2024

> 500 Specialized clinics   > 550 doctors

Axis

02

Health themes in rare diseases

1. Dysmorphic or polymalformative syndromes with or without associated neurodevelopmental disorders
2. Autism, intellectual disability, and other neurodevelopmental disorders
3. Early-onset dementias, neurodegenerative diseases, and other rare central nervous system conditions
4. Isolated hereditary heart diseases
5. Immunodeficiency, rare autoimmune and systemic autoinflammatory diseases
6. Rare vascular diseases with multisystem involvement
7. Rare liver diseases
8. Neuromuscular diseases and other rare peripheral nervous system disorders
9. Rare dermatological diseases
10. Rare endocrine diseases
11. Inborn errors of metabolism
12. Constitutional erythropoiesis disorders, hemostasis disorders, and other rare hematological conditions
13. Cystic fibrosis and other CFTR gene-related conditions
14. Rare respiratory diseases
15. Rare kidney diseases
16. Rare diseases of the bone, calcium, cartilage, and soft tissues
17. Congenital deafness
18. Rare retinal diseases and other rare eye conditions
19. Rare diseases of the head, neck, and teeth

Policy and Action Plan

Axis

03

3. PROMOTION OF RESEARCH, INNOVATION, AND DATA COLLECTION

4 objectives

- **3.1** Establish a Quebec **registry** of patients with rare diseases
- **3.2** Promote **fundamental, translational, and clinical research** to better detect and understand these types of diseases
- **3.3** Facilitate **knowledge transfer** between researchers, clinicians, and patients
- **3.4** Accelerate therapeutic **innovation**

9 actions

Policy and Action Plan

Axis

03

3. PROMOTION OF RESEARCH, INNOVATION, AND DATA COLLECTION

9 actions

Notamment:

- ✓ Develop a research network RARE.Qc
- Implementation of a registry
- Support the establishment of biobanks
- Call for projects to stimulate research
- Facilitate the integration of clinical trials into the continuum of care
- Support program for the development of innovation

Conclusion

In summary, the ministerial guidelines on rare diseases:

- **3 axes of intervention**
- **12 objectives**
- **38 actions (+ sub-actions)**
- **Timeline: 2023-2027**

Conclusion

For a health network:

- More sensitive, better trained, and better equipped
- Better structured: more fluid and standardized continuum of care and services

For a research and innovation environment: dynamic

For patients who:

- Reach their full health potential
- Feel better recognized and more supported

Acknowledgements

- The publication of these guidelines is a work of collaboration and multiple consultations

Support groups and patient associations

- More than 65 groups reached out
- 24 responded to the call and were consulted twice

Other groups working in rare diseases

Montreal InVivo

Catalis Québec

MSSS Divisions

Hospital Services Division

Pharmaceutical Affairs and Medication Division

Biovigilance and Medical Biology Division

University Affairs Division

Research Division

Maternal and Child Health Division

Clinical Prevention,

Dental Health, and Screening Division (Public Health)

Quebec Advisory Committee for Rare Diseases

Professional associations

- ACCGQ
- AMGQ
- FMOQ
- FMSQ
- APPQ
- Nurses in rare diseases
- First Nations and Inuit: First Nations CSSS of Quebec and Labrador, Atikamekw sipi

Recruitment of Patient partner and rare disease organizations

Please don't hesitate to contact me:

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Thank you

Questions?