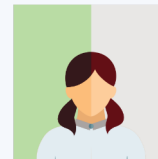
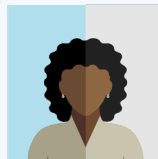


CORD
April 29th, 2025

NAVI-NAT:

Province-Wide Navigation for Timely Diagnosis of Rare Diseases

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CHU Ste-Justine and Université de Montréal



Rare diseases in Quebec

Current services in rare diseases include:

Multiple provincial programs (PQDNSU, RQDM, PAQTMMH, etc.)

Up to 300 clinics diagnosing and treating patients with rare diseases

Multiple programs to access drugs for rare diseases

Despite the existence of programs and clinics, many challenges need to be addressed:

Lack of standardization

Lack of common overarching structure in programs and services

Lack of synergy across the health care system

Challenges for individuals living with rare diseases in Québec

Diagnosis and management

Diagnostic odyssey

Psychiatrization

Inadequate care

Inequitable access

Lack of coordination/
fluidity of care

Access to treatments

Few efficient treatments
available

Few clinical trials/limited
research on rare
diseases

Access to social services

Lack of psychosocial
support

Psychological distress

Time for action in rare diseases in Quebec

Politique québécoise pour les maladies rares (2022)

Health care and services in rare diseases

Quality, safe, equitable, inclusive, adapted and culturally sensitive care and services

Patient-centered approach

Fast and facilitated access to care and services

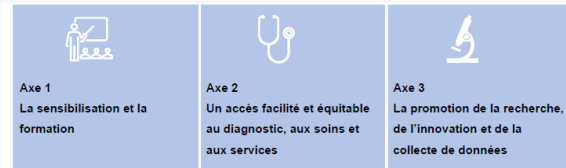
Learning approach



<https://publications.msss.gouv.qc.ca/msss/fichiers/2022/22-916-01W.pdf>

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

1. Raising awareness and training
2. Equitable and facilitated access to diagnosis, care and services
3. Promotion of research, innovation and data collection



Axe 1
La sensibilisation et la
formation

Axe 2
Un accès facilité et équitable
au diagnostic, aux soins et
aux services

Axe 3
La promotion de la recherche,
de l'innovation et de la
collecte de données

<https://publications.msss.gouv.qc.ca/msss/fichiers/2023/23-916-02W.pdf>

MSSS names Reference Centers in rare diseases



5 centres de référence interdisciplinaires pour les soins et la recherche en
en maladies rares et un centre de référence régional désignés

CHUM

CHU de Qc - UL

CIUSSS Estrie
(CHUS)

CUSM

CHU Sainte-
Justine

CIUSSS SLSJ
(Centre de
référence
régional)

Census of rare disease expertise and services in Quebec



Recensement québécois des soins et services disponibles en maladies rares

Started by the MSSS in May 2024

Method: Survey addressed to each reference center

Objective: Dress an accurate and detailed portrait of the currently available expertise and services in rare diseases in the Quebec health care system

Rare disease themes proposed by MSSS

1	Syndromes dysmorphiques ou polymalformatifs avec ou sans troubles neurodéveloppementaux associés
2	Autismes, déficience intellectuelle et autres troubles neurodéveloppementaux
3	Démences précoces, maladies neurodégénératives et autres atteintes rares du système nerveux central
4	Maladies cardiaques héréditaires isolées
5	Immunodéficience, maladies auto-immunes et auto-inflammatoires systémiques rares
6	Maladies vasculaires rares accompagnées d'une atteinte multisystémique
7	Maladies hépatiques rares
8	Maladies neuromusculaires et autres maladies rares du système nerveux périphérique
9	Maladies rares en dermatologie
10	Maladies rares endocriniennes
11	Erreurs innées du métabolisme
12	Maladies constitutionnelles de l'érythropoïèse, de l'hémostase et autres conditions rares hématologiques
13	Mucoviscidose et affections liées à une anomalie de cystic fibrosis transmembrane conductance regulator (CFTR)
14	Maladies respiratoires rares
15	Maladies rénales rares
16	Maladies rares de l'os, du calcium et du cartilage
17	Surdités congénitales
18	Maladies rares de la rétine et autres maladies rares de l'œil
19	Maladies rares de la tête, du cou et des dents
20	Non applicable ou autre

NAVI-NAT: a component of the action plan

Through a **network of clinical experts** and with the support of **nurse navigators**, patients will get access directly to the appropriate expertise.

Patients will be directed to an expert in the NAVI-NAT network and offered the appropriate investigations, for a **more efficient path to diagnosis**.

In the absence of a diagnosis, their case will be presented to a provincial NAVI-NAT case discussion meeting to recommend future steps in their evaluation.

Patients accepted in the NAVI-NAT network **remain active in the network until a diagnosis of rare diseases is established or ruled out**.

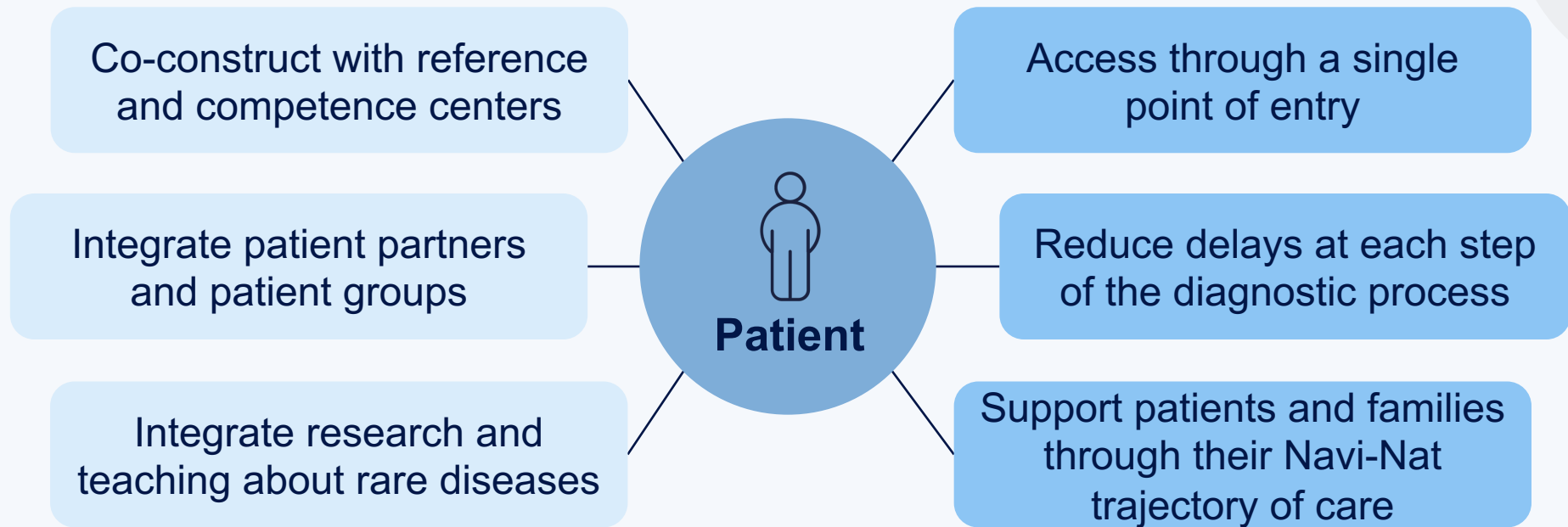
NAVI-NAT:

Provincial navigation network in rare diseases

The objectives of NAVI-NAT are:

- Assist patients in their path to a diagnosis
- Coordinate efforts to shorten the diagnostic odyssey, for individuals with rare diseases to attain their full health and well-being potential.
 - Build on the expertise in the health care system, by working closely with the reference and competence centers.
 - Act as the entryway and as a resource to guide patients more quickly to the most relevant care and services
- Create a community of practice in rare diseases, to share best practices and benefit from the expertise of all stakeholders

The NAVI-NAT vision



Increased value for the patient and for the health care system



Coûts



Qualité



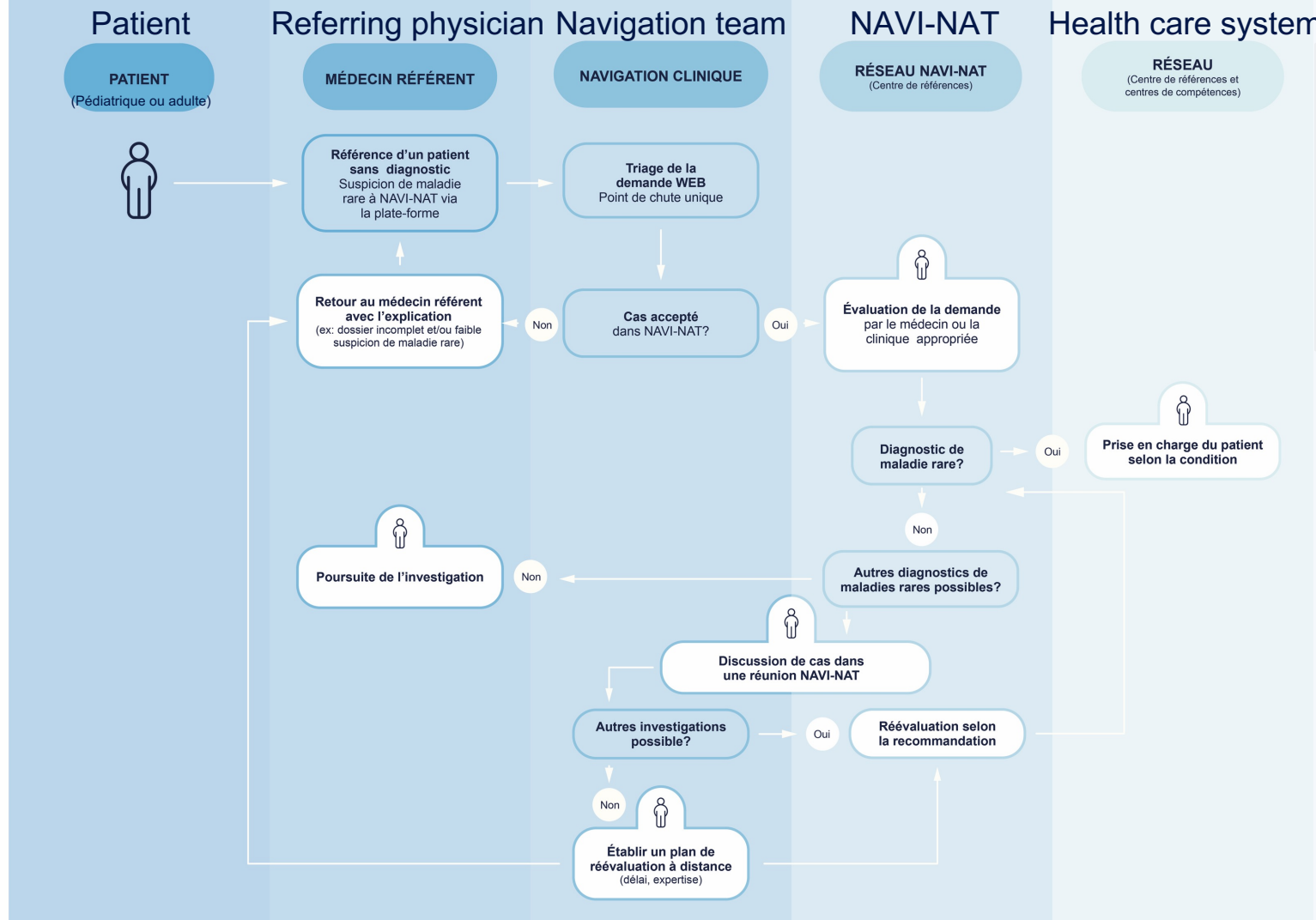
Résultats

- **Reduce waiting times de délais d'attente :**
 - Referral is evaluated and directed to the specialist/clinic with the appropriate relevant expertise within 5 days
 - First clinical evaluation→ Objective : 4 weeks
- **Delay to diagnosis**
 - Target : 3 to 6 months, depending on the complexity of the situation
- **Orientation of patient to appropriate care and services, with expert recommendations**
- **Reduction of costs due to the diagnostic odyssey**

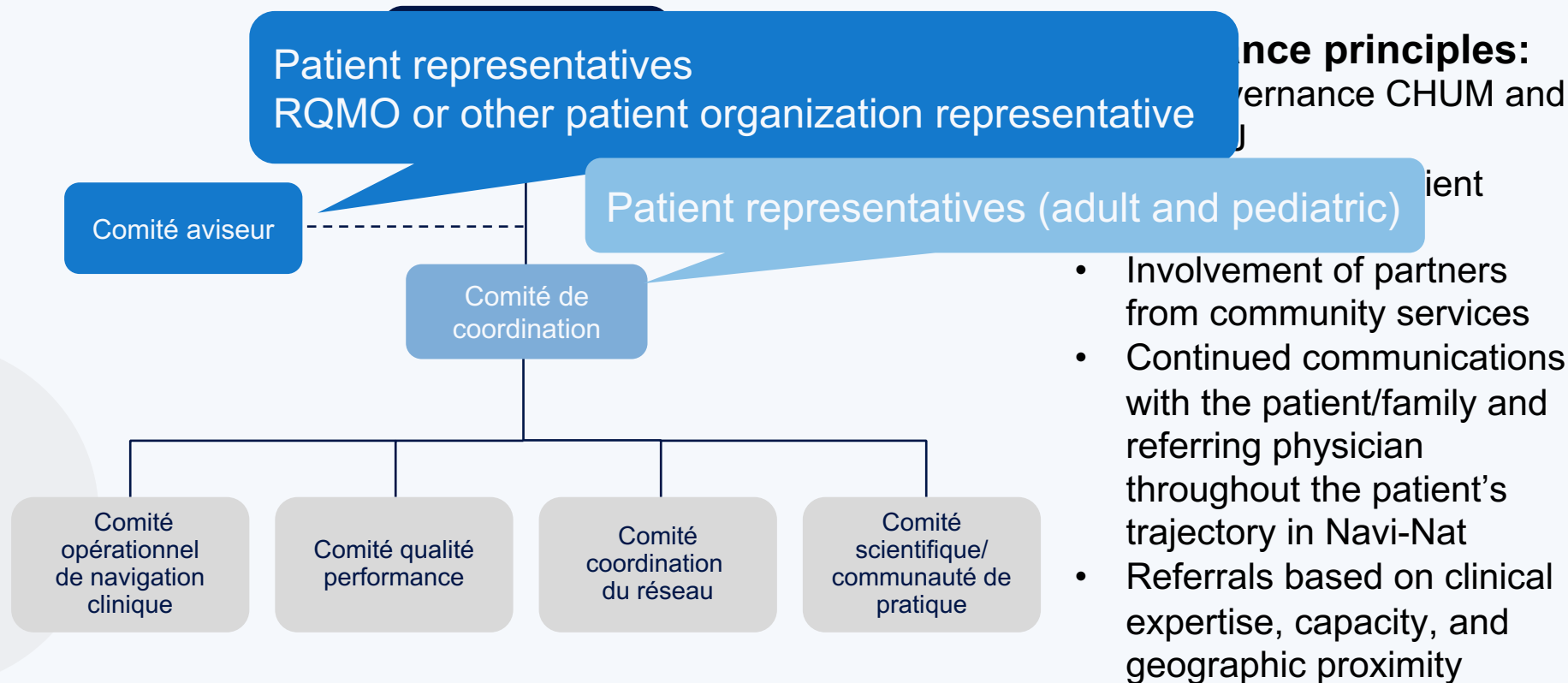
What NAVI-NAT **is not**

- NAVI-NAT **is not** a substitute to existing trajectories of care
 - It uses, improves and connects to existing trajectories and structures
- NAVI-NAT **is not** an addition to the patient load for reference centers
 - It improves the orientation of patients more directly to the right expertise
- NAVI-NAT **is not** a centralized approach to care
 - It improves communication before and after a referral, to facilitate better collaboration in the health care system.
- NAVI-NAT **is not** the only strategy in the rare disease action plan
 - It is one of many actions planned to improve care in rare diseases
- NAVI-NAT **is not** comprehensive patient care for individuals with rare diseases
 - It focuses on access to a diagnosis

Patient trajectory in Navi-Nat



Governance structure

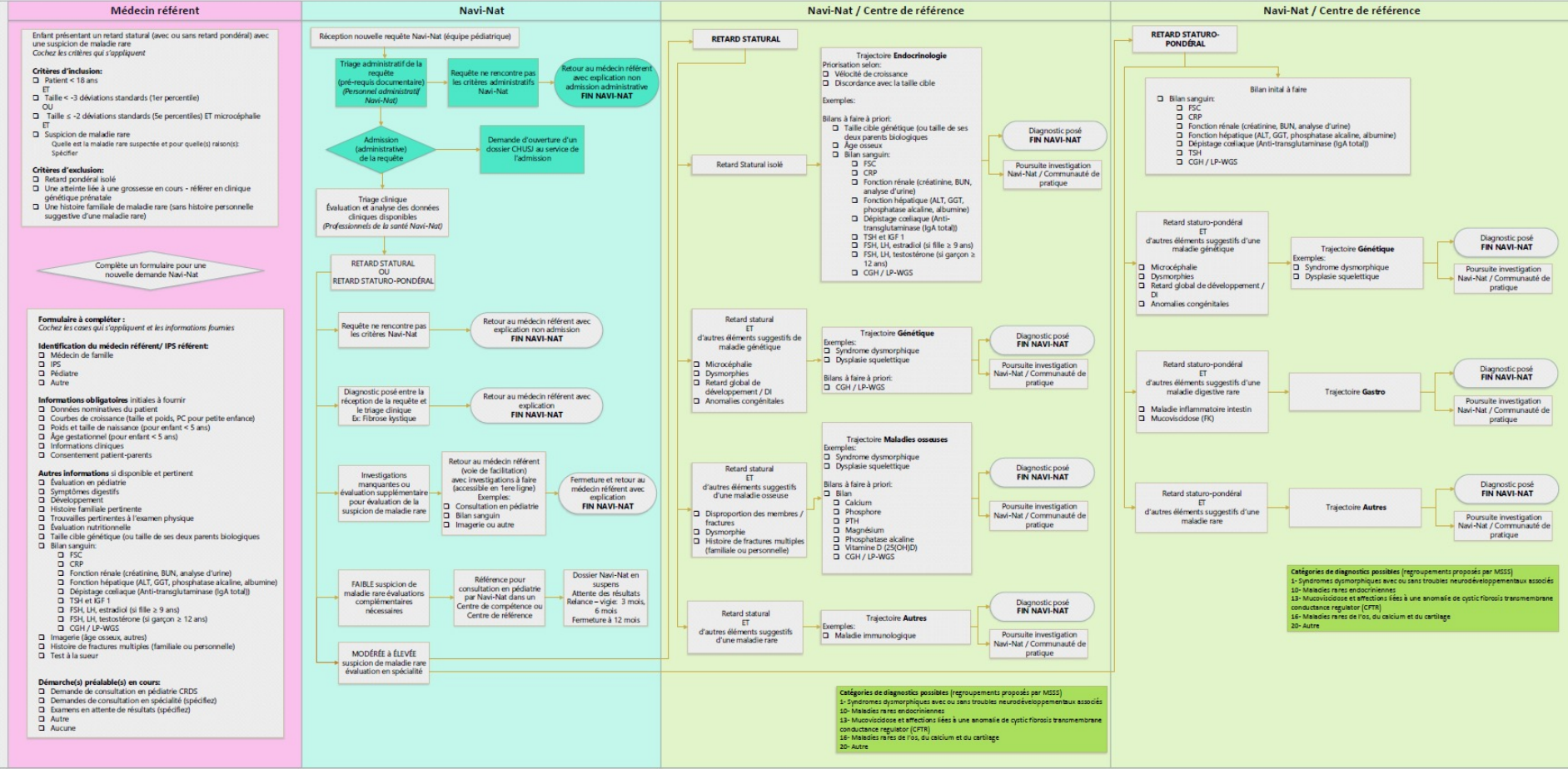


Piloting NAVI-NAT

Pilot triage algorithms developed, based on common presentations of rare diseases.

- For adults:
 - Diagnostic odyssey (> 3 specialist evaluations in last year without a diagnosis) and suspected rare disease
 - Hypermobility and suspected rare disease
- For children
 - **Short stature/failure to thrive and suspected rare disease**
 - Overgrowth and suspected rare disease
 - Diagnostic odyssey (> 3 specialist evaluations in last year without a diagnosis) and suspected rare disease

Navi-Nat Algorithmhe décisionnel: Retard statural et suspicion de maladie rare- pédiatrie - DOCUMENT DE TRAVAIL

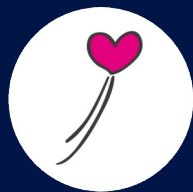


Strengths of NAVI-NAT

- Patient-centered provincial approach to facilitate access to a diagnosis
- Advisory committee including diverse stakeholders, including both patient representatives and patient group representatives
- Navigation team already in place (2 nurses, 3 genetic counselors, 2 medical geneticists, 1 pediatrician)
- Support from the MSSS
- Engagement from the leadership and clinical teams in the Reference Centers

Next steps for NAVI-NAT

- Keep census of expertise in rare diseases across centers in Quebec up to date and complete
- Ensure tools used for tracking patient trajectories in NAVI-NAT are efficient and secure
- Navigate the transition to Santé Québec
 - Make sure roles and responsibilities of all parties and stakeholders are clearly defined
- Raise awareness about NAVI-NAT
- Build a community of practice in rare diseases in Quebec
- Measure impact of NAVI-NAT for patients with rare diseases



Merci!

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