Published
May 22, 2025

MONTRÉAL INVIVO

Scientific watch for new therapies indicated in the treatment of rare conditions as well as cell and gene therapies

Updated version as of June 4, 2024

In Collaboration with Synergyx Consulting Inc. and KPMG

This study was conducted with the financial support of Takeda, Pfizer, Vertex Pharmaceuticals (Canada) Inc., AstraZeneca Canada Inc. and Roche Diagnostics

AstraZeneca VERTEX Pfizer Takeda Roche Diagnostics

Note to readers

- This is the second monitoring report on new therapies indicated in rare conditions and on cell and gene therapies produced by Montréal InVivo.
- There are many programs to develop new treatments in the fields of rare diseases and cell and gene therapies, with thousands of clinical trials underway worldwide. The research conducted for this report was conducted using selection criteria and therefore this tool cannot be considered exhaustive. The methodology used is detailed in the appendix.
- The list of therapies includes a number of therapies that have already been approved by Health Canada. These therapies remain interesting because they are the subject of ongoing clinical trials for new indications that are being examined.

Objectives of this report

Montréal InVivo's mission

Montréal InVivo is an economic development organization whose mission is to promote the growth and competitiveness of organizations in the life sciences and health technologies sector in Montreal. One of Montréal InVivo's strategic focuses on research and innovation, of which rare diseases are a priority theme.

The objectives of the report

This monitoring of new drugs* related to rare diseases and new cell and gene therapies* aims to inform stakeholders, particularly in a context where resources are limited. As more therapeutic options with significant potential benefits for patients become available, the goals are:

1. **To ensure rapid and optimal access for Quebec patients to therapeutic and diagnostic innovations.**
2. **To strengthen the competitiveness of Quebec's innovation ecosystem to attract investment and develop skills based on international best practices.**

Challenges in evaluating and implementing cutting-edge innovations in the healthcare system can lead to sometimes critical **access delays** for patients with rare conditions or who could benefit from cell and gene therapies. The importance of these issues is highlighted in the 2022-2025 Quebec Life Sciences Strategy and the 2023-2027 Quebec Action Plan on Rare Diseases.

*Due to the nature of the monitoring tool, many treatments have not yet been evaluated by Health Canada.

Executive Summary

- The report contains 196 new therapies in development, of which 139 are for rare diseases and conditions (71%), 37 are for cell and gene therapies (19%), among which 23 are for rare diseases and conditions (12% of all treatments).
- For rare diseases and conditions:
  - Targeted therapies represent 43% of therapies in development.
  - More than half of the therapies are in phase 3 clinical studies.
  - Most clinical studies are conducted in adults.
  - Nearly half (49%) of the new therapies in development can be administered by subcutaneous or oral injection, meaning they can be obtained in a community pharmacy and administered at home.
- For cell and gene therapies:
  - Gene therapies account for 58% of therapies in development.
  - 40% of the therapies are in phase 3 clinical studies and most studies are carried out in adults.
  - 62% are for rare diseases and conditions.
  - 32% are for rare and ultra-rare cancers.

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Background and Introduction

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Rare diseases – a global crisis

±300M

People impacted by rare diseases around the world.[1,2]

Of rare diseases have approved treatments.[1,2]

+7 000

Known rare diseases.[1,2]

80%

of rare diseases have a genetic cause.[1,2]

$20M

Canada's investments in 2024 to improve the health of children living with rare diseases.[3]

70%

of rare disease manifest in childhood.[1,2]

Of children with rare diseases die before age 5.[1,2]

4,8 years

The average time to get an accurate diagnosis.[1,2]

517 000$

As avoidable expenses due to delayed diagnosis for a patient living with a rare disease.[4]

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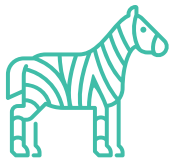
[1] *The Lancet, « The landscape for rare disease in 2024 »*
[2] *Parexel International, Inc., « Rising to the challenges of developing rare disease treatments »*
[3] *Instituts de recherche en santé du Canada, Le gouvernement du Canada investit 20 millions de dollars en vue d'améliorer les résultats cliniques des enfants vivant avec une maladie rare*
[4] *Global Genes, « Rare Disease Facts »*

Provincial and national responses



In **June 2022**, the Quebec Ministry of Health and Social Services (MSSS) unveiled the Quebec Policy on Rare Diseases, a comprehensive strategy to:[1]

- Strengthen **awareness and training**
- Ensuring **equitable access** to diagnosis and care
- Foster **research and innovation** in the field of rare diseases.

In **March 2023**, Canada's first-ever national strategy for rare disease drugs was launched, supported by a government investment:

- **$1.5 billion over three years** to improve access and affordability of treatment nationally.[2]
- **$1.4 billion** in **additional** funding **for provincial and territorial agreements** focused on access to new drugs, early diagnosis and rare disease screening.[1,2]

As of **November 2024**, bilateral agreements were signed between the federal government and the following provinces and territories to distribute the amount of $1.4 billion:

- Alberta, British Columbia, Manitoba, New Brunswick, Newfoundland and Labrador, Ontario, and Saskatchewan have already signed.[3]
- Quebec is still in negotiation.

Quebec's proactive and collaborative approach to rare diseases is marked by the early adoption of a specific policy, and a strong collaboration with experts and institutions to develop these resources. It improves the dissemination of knowledge, equips health professionals, and positions **Quebec as a leader in innovation in rare diseases.**

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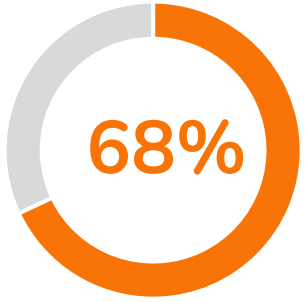
1. *Fasken, « Minister of Health Unveils Quebec's Rare Diseases Policy »*
2. *Health Canada, Canadians Invited to Provide Their Views on a National Strategy for High-Cost Drugs for Rare Diseases.*
3. *Health Canada, Bilateral Agreements for Drugs for the Treatment of Rare Diseases*

Cell and gene therapies

2 848

Cell and gene therapies (CGT) in development globally as of June 2024.[1]



Des TCG approuvées sont des produits cellulaires non génétiquement modifiés en date de juin 2024. [1]

100

Globally approved CGT products as of June2024. [1]

77

Number of products in Phase III clinical trials as of June2024.[1]

11

Number of CGT approved by Health Canada since 2012.[5]

North America

Leading region in CGT trials, with the highest number of ongoing clinical trials(987).[4]

105 000

Projected population eligible for CGT in 2032 (patients per year).[6]

11,3 B$

Amount invested in H1 2024 for CGT. [2]

12x

Expected growth of the CDMO market for CGT by 2034, reaching 74 B$. [3]

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[1] International Society for Cell & Gene Therapy (ISCT), « Cell and Gene Therapy Global regulatory report H1 2024»
[2] Alliance for Regenerative Medicine, « Investment Data – 2024 Q3 »
3 Precedence Research, « Cell and Gene Therapy CDMO Market Size and Forecast 2025 to 2034 »
4 Alliance for Regenerative Medicine, « Clinical Trials – 2024 Q3 »
5 Signals, Insider's perspective on the world of stem cells and regenerative medicine « Cell and gene therapies approved by Health Canada and global regulators »
6 Tufts Medicine, « Cell and Gene therapy (CGT) pipeline deep dive »

Analysis of the results

Analysis period: 10/2022 – 06/2024

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Main selection criteria

Rare diseases and conditions

- Affect less than 1/2,000 people
- Phase 2+ clinical trial

Cell and gene therapies

- All Diseases/Conditions
- Phase 1 and + clinical trial*

For both areas of interest, clinical trials had to be sponsored by a biotechnology or pharmaceutical company and take place in the United States, Europe or Canada.

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* Phase 1 clinical trials are included since several clinical studies in cell and gene therapies are combined studies (e.g. phase 1/2/3 study.

Main sources used

Only public data was used.

The main sources are provided here, and a detailed bibliography is in the appendix.

Commercially available drugs (updated)

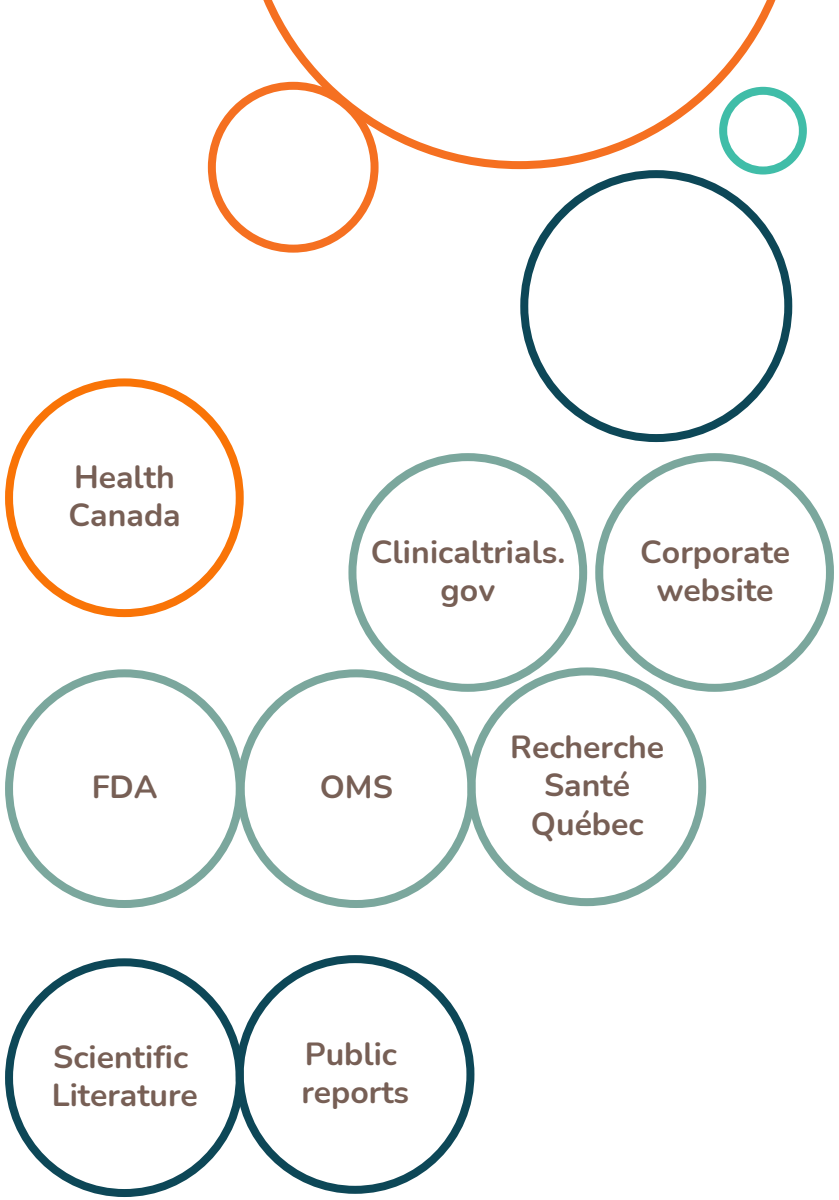
This source was used to gather information on recent approvals and therapies under review.

Products in clinical trials

These sources were used for information on ongoing clinical trials.

Detailed therapeutic data

These sources provided additional information on therapies and clinical trials.



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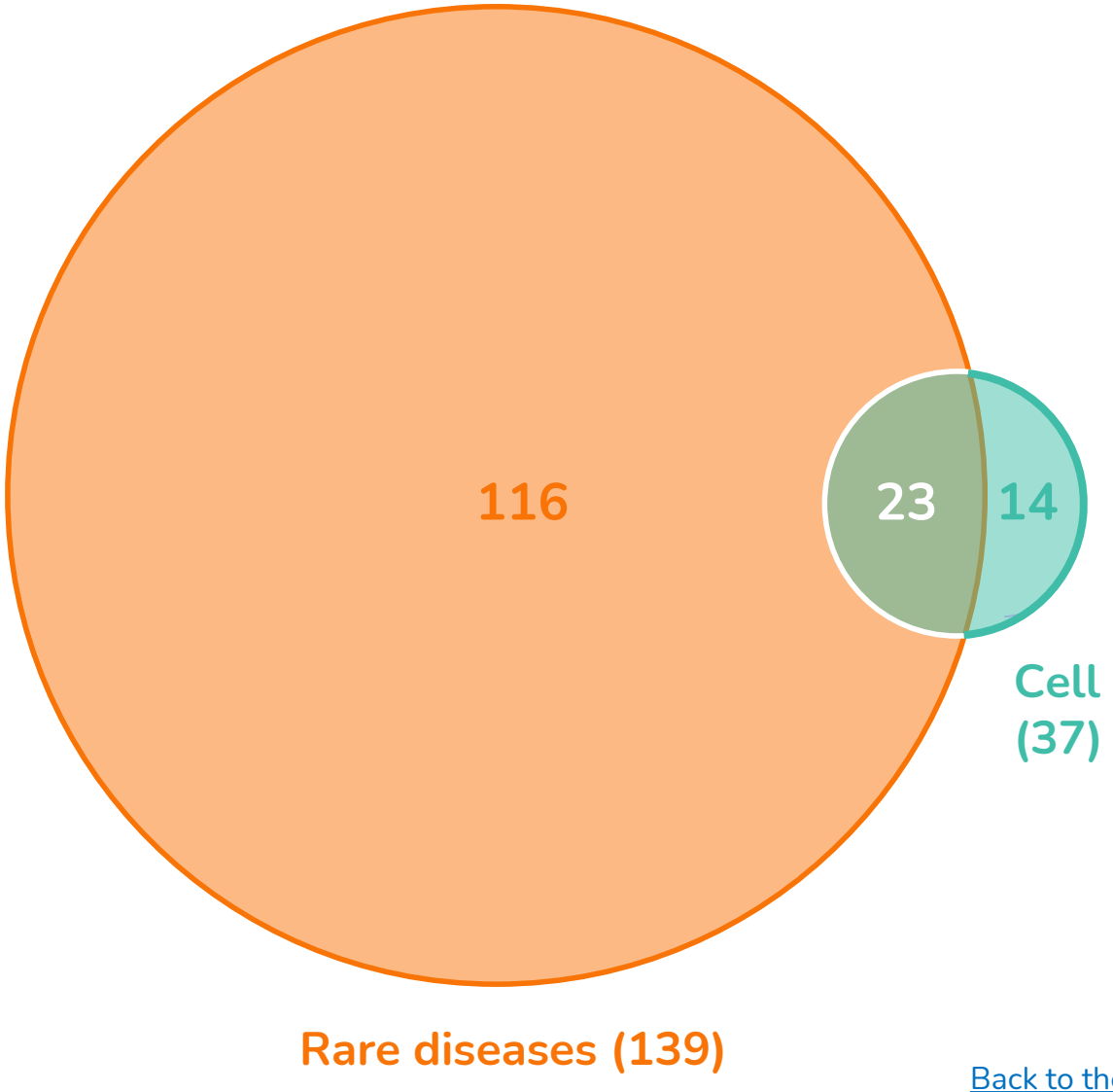
Rare Diseases and Cell and Gene Therapies

Of the 196 treatments included in this report, 139 are for rare diseases and conditions, 37 are cell and gene therapies, 23 of which are for rare diseases.



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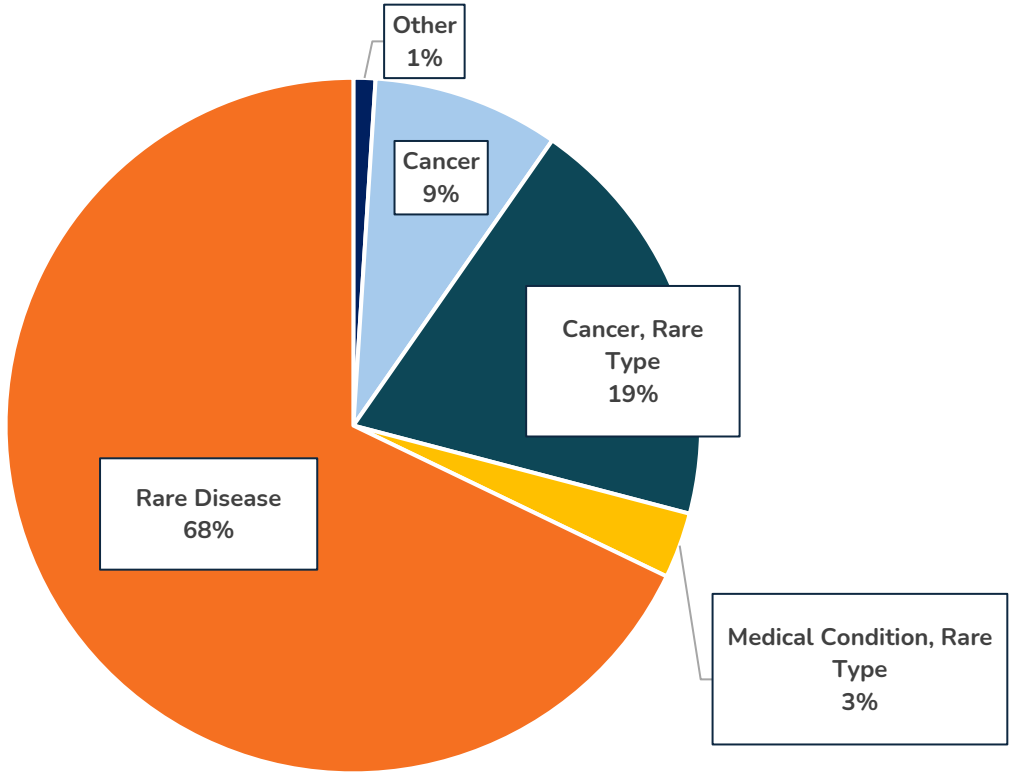
Breakdown of new therapies in development by therapeutic area (N=196)

Nearly two-thirds of the new therapies in development are for rare diseases.



Note: The rare disease category is composed of all treatments for conditions considered rare and ultra-rare.

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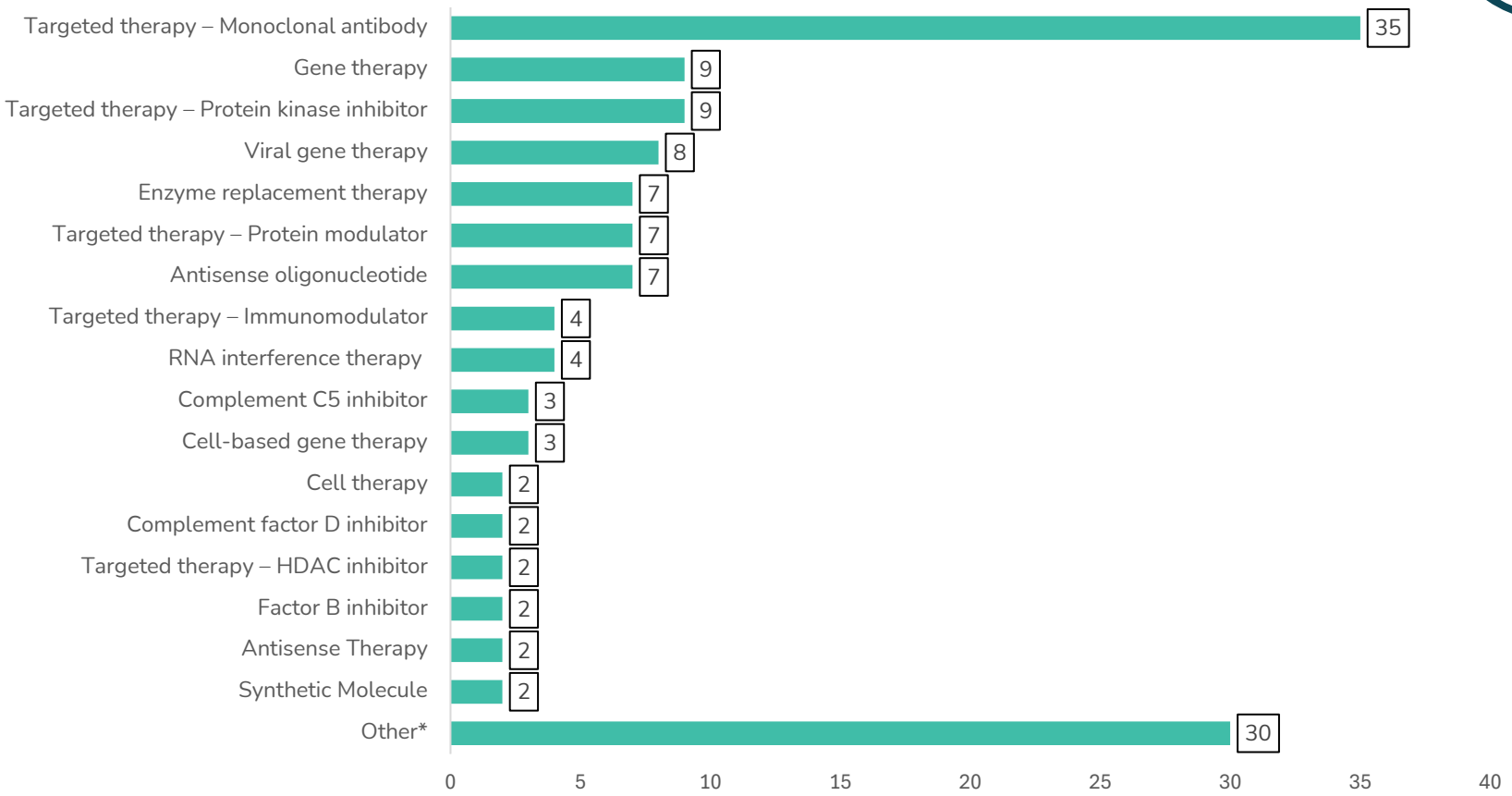
Breakdown of new therapies in development for rare diseases and conditions by type of treatment (n=138)

Targeted therapies make up 43% of all 138 treatments for rare diseases and conditions.

Cell and gene therapies make up 17% of the total.



** Other refers to any therapy that appears only once in the monitoring tool*

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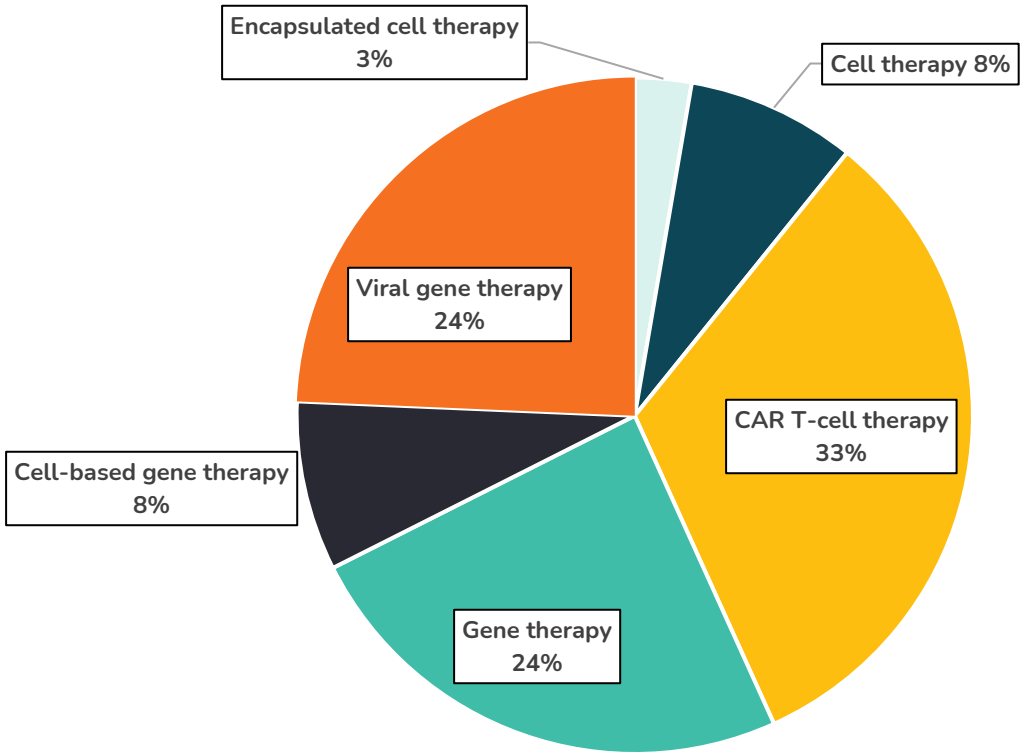
Breakdown of new cell and gene therapies in development by type of treatment (n=37)

Gene therapies account for 58% of cell and gene therapies in development



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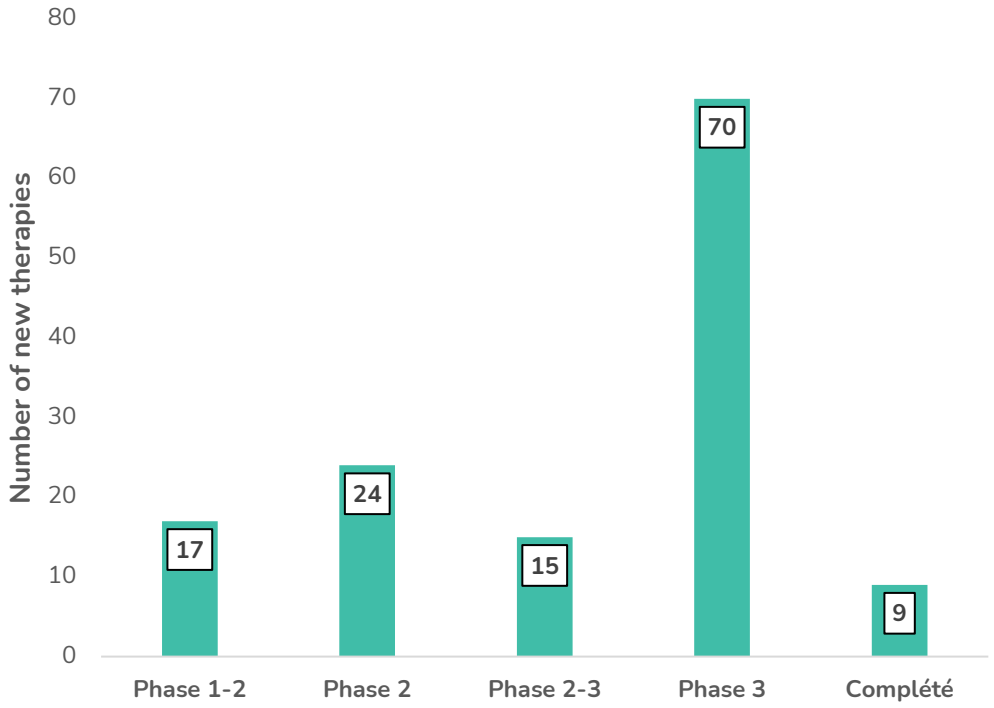
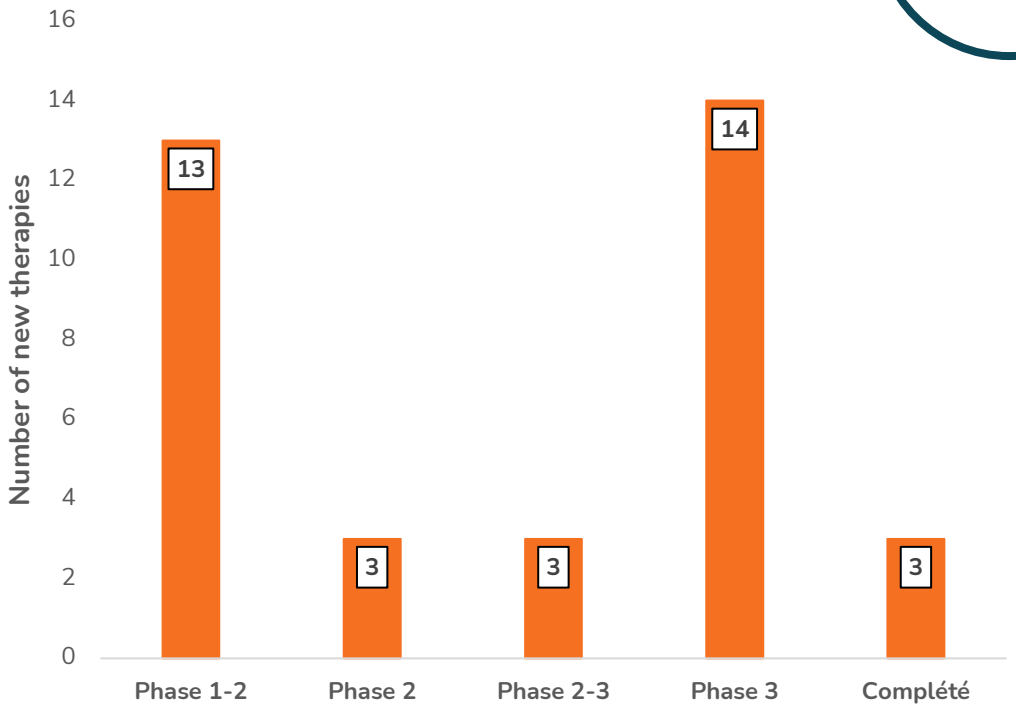
Breakdown of new therapies in development by clinical research phase

A. For rare diseases and conditions (n=135)



For rare diseases and conditions, more than half of the treatments are in phase 3 clinical studies.

B. For Cell and Gene Therapies (n=36)



Nearly 36% of new CGT are in phase 1-2 clinical studies and almost 40% are in phase 3.

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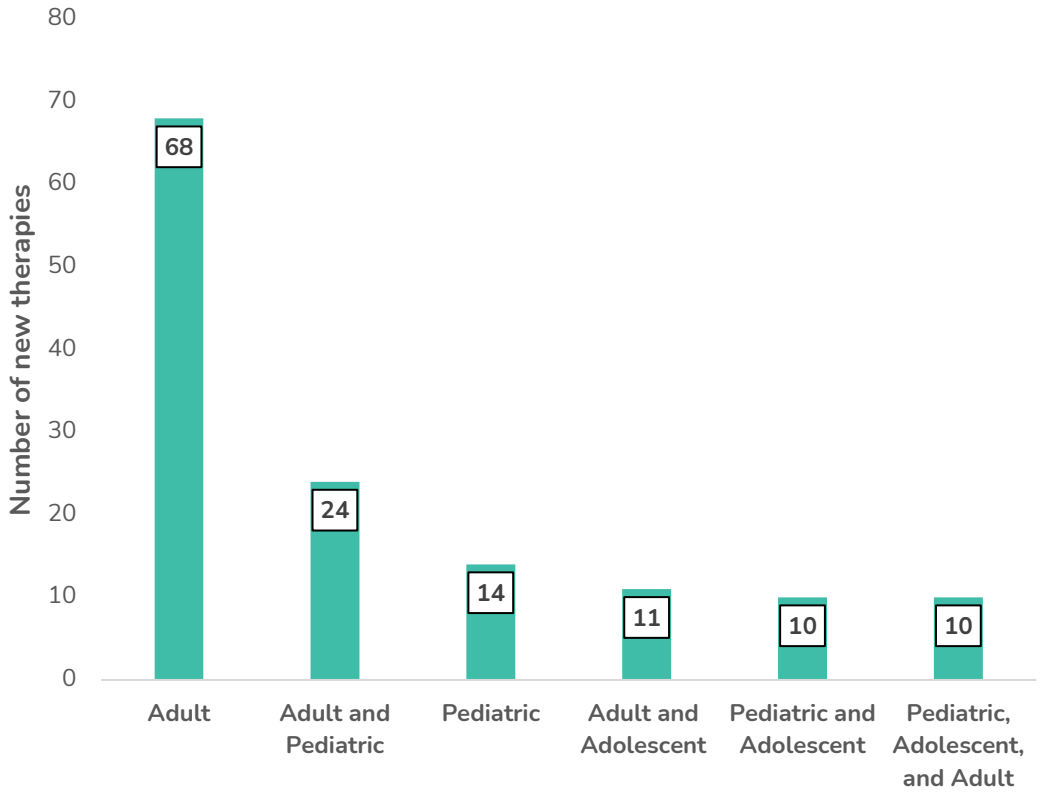
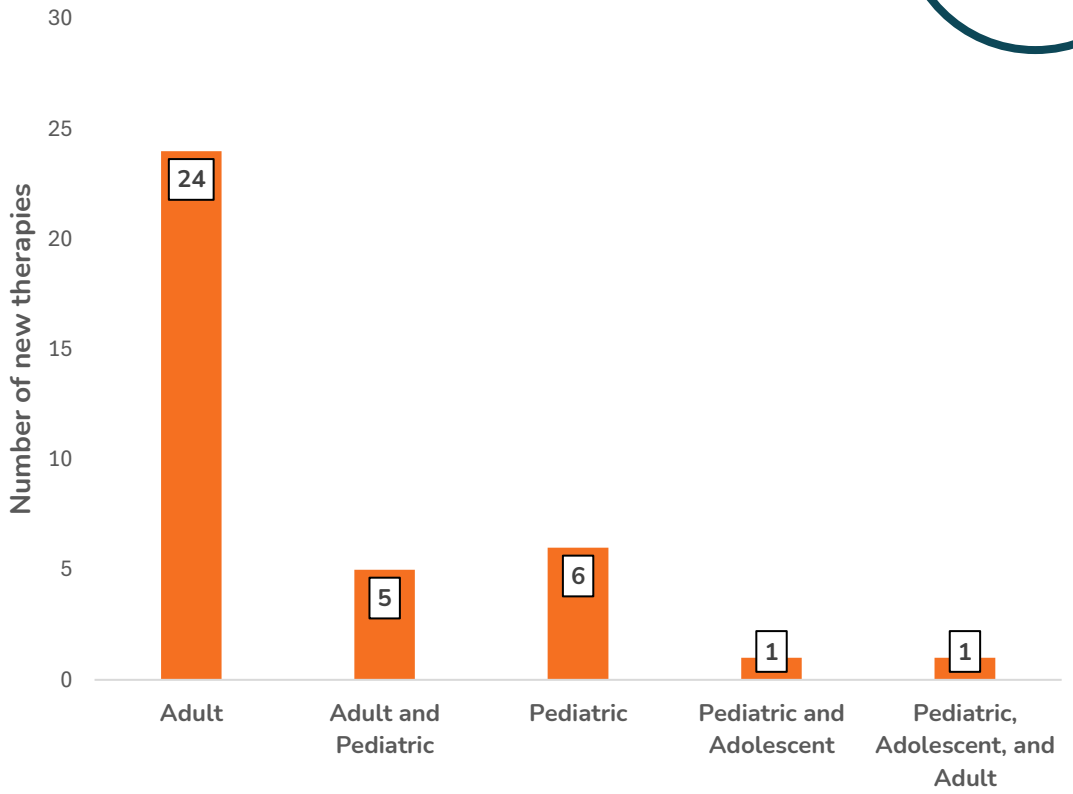
Breakdown of new therapies in development by target population

A. For rare diseases and conditions (n=137)



B. For Cell and Gene Therapies (n=37)



The majority of clinical studies of treatments are carried out in the adult population.

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Impact of routes of administration on patient management in the Quebec health care system

- The complexity of new therapies brings challenges in terms of implementation in the health and social services network.
- One of the priorities in Quebec is to relieve the burden of hospital care in the health care system and to increase the capacity and management of patients in outpatient clinics, community pharmacies or at home.
- Therefore, routes of administration such as oral or subcutaneous injection of new therapies in development can support this initiative and facilitate patient management outside of the hospital setting.

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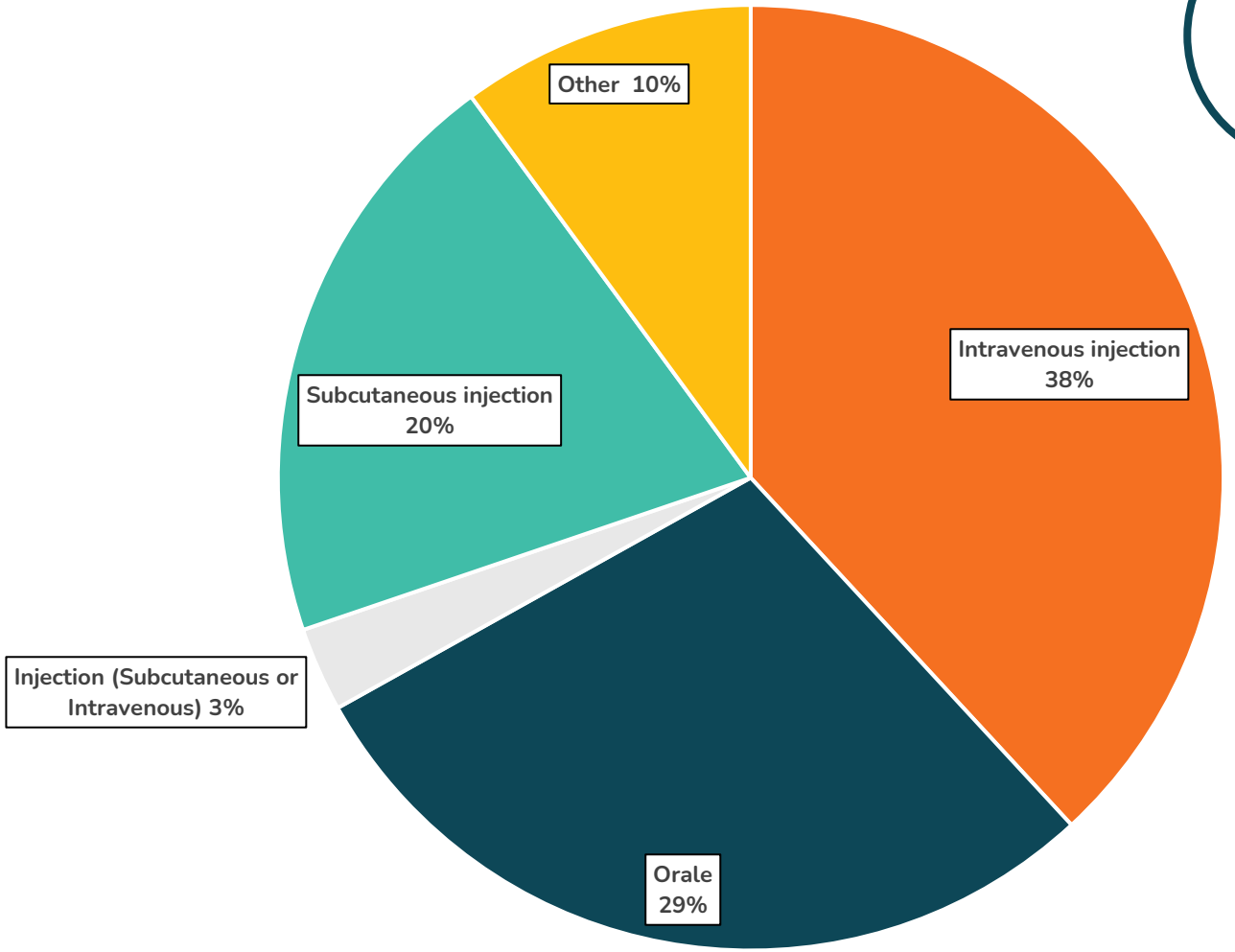
Breakdown of new therapies in development for rare diseases and conditions by route of administration (n=139)

Nearly half (49%) of the new therapies in development can be administered by subcutaneous or oral injection.

These treatments can be available at community pharmacies and administered at home – an important change in the patient care journey.

However, TCGs have more complex care pathways, they are considered as a whole to be offered in hospitals.



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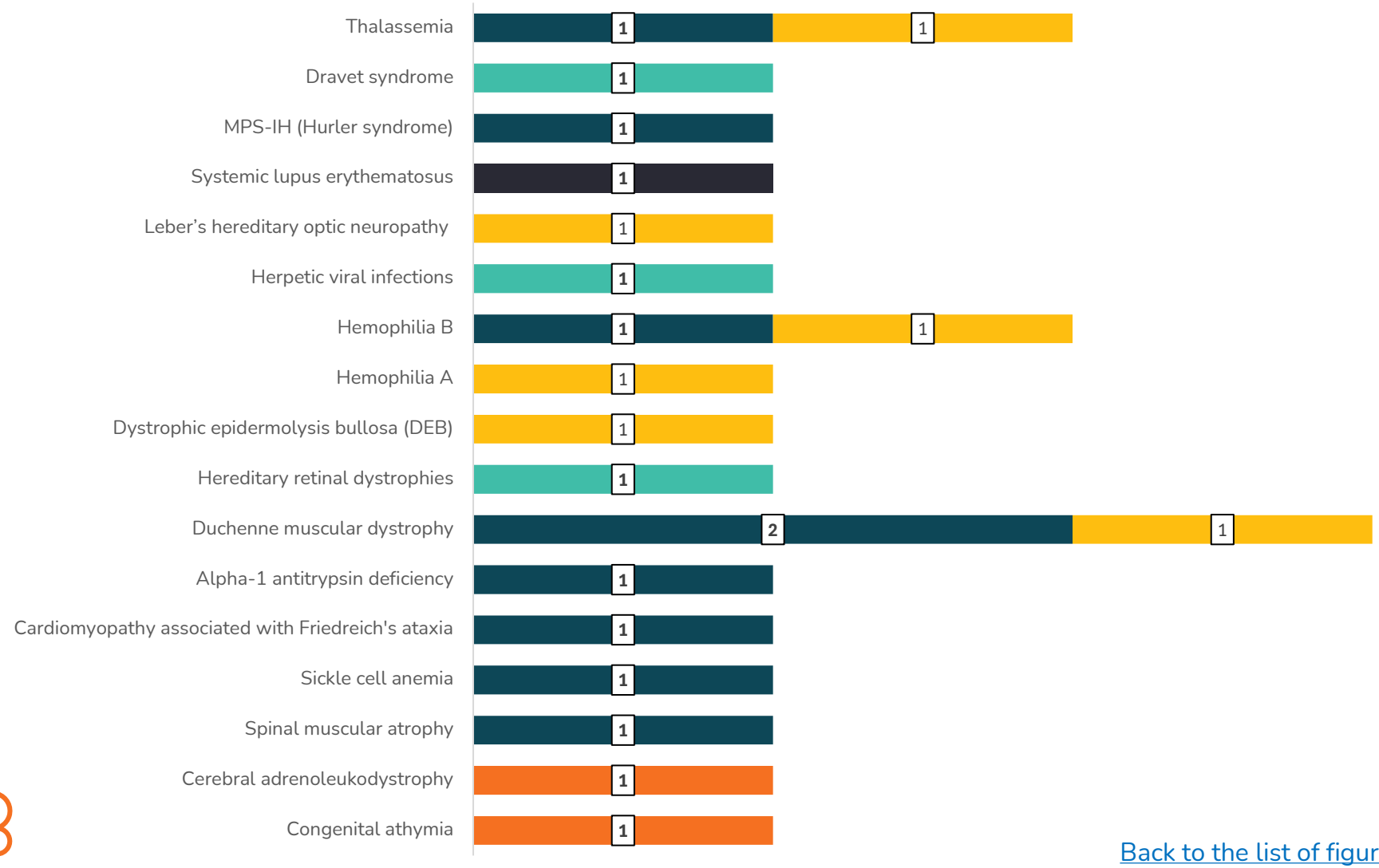
Breakdown of CGT used in rare and ultra-rare diseases (excluding cancers) by medical condition (n=23)

New cell and gene therapies are in development for 23 rare and ultra-rare indications (excluding cancers).



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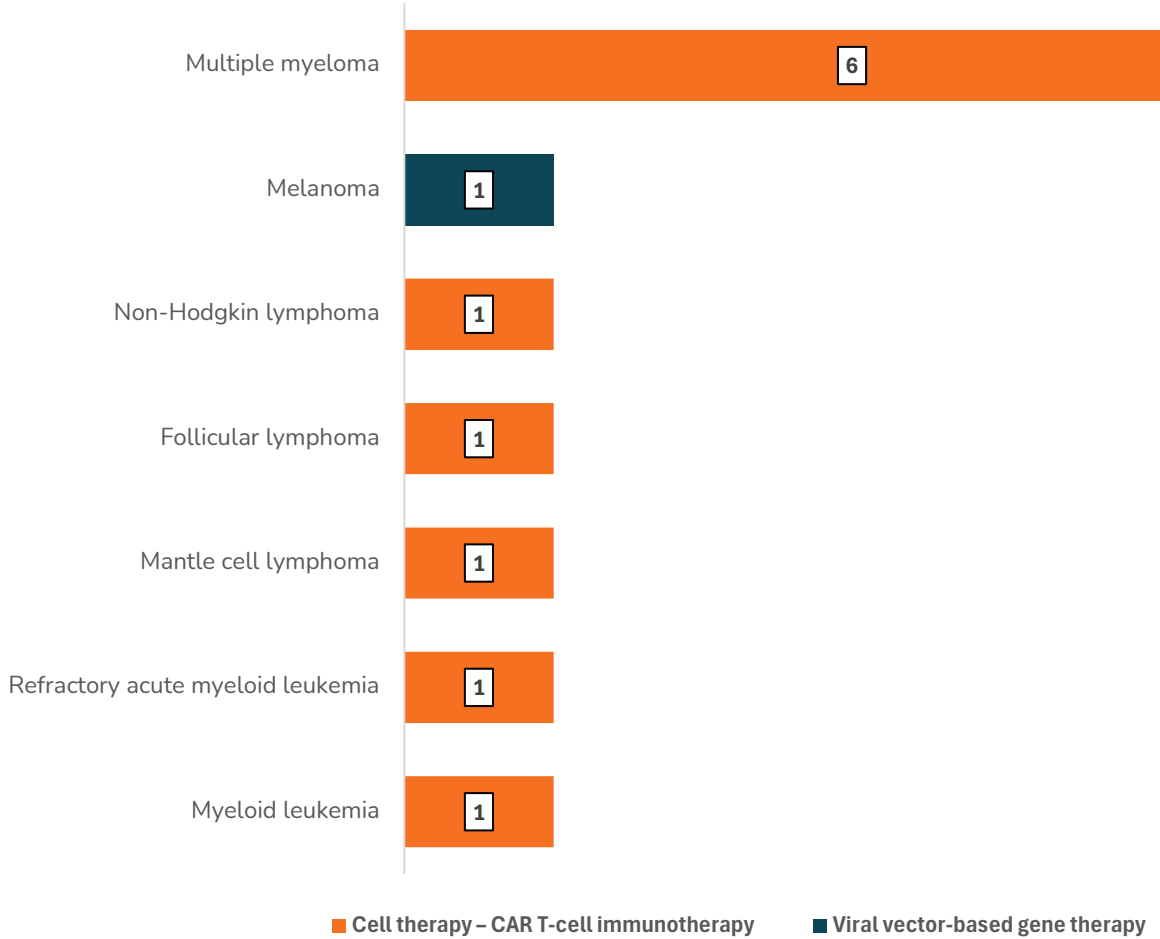
Breakdown of CGT in oncology by therapeutic indication (n=12)

New cell and gene therapies are in development for 12 rare and ultra-rare cancers.



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Future prospects

Strategic alignments to consider

- This monitoring is part of a short- to medium-term vision of the objectives of the Montreal InVivo Rare Disease Specialized Project Committee, which are to:
  1. To enable Quebec patients with a rare disease to have rapid and optimal access to therapeutic and diagnostic innovations;
  2. Improve the competitiveness of Quebec's innovation ecosystem to attract investment and develop our skills by drawing inspiration from international best practices.

- Opportunities to support the MSSS's 2023-2027 Quebec Action Plan for Rare Diseases and the Quebec Life Sciences Strategy (SQSV) through the actions of Montréal InVivo and its partners, including:
  - Training and awareness: data science programs, links with the next generation
  - Better access to care: committee projects value-based agreements, public-private collaborations for the development and commercialization of innovations
  - Better use of health data for research and innovation: AI in Health Innovation Hub

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Appendix

Methodology and approach

Common definitions

Keyword	Definition
Rare and ultra-rare diseases	For the purposes of this report, both terms are categorized as "rare diseases."
Targeted therapy	A treatment that focuses on genetic changes or mutations that turn healthy cells into diseased cells (cancer or other rare diseases).
Cell Therapy	Cell therapy products include cell immunotherapies, cancer vaccines, and other autologous and allogeneic cell types for certain therapeutic indications, including hematopoietic stem cells and adult and embryonic stem cells.
Gene therapy	Human gene therapy aims to modify or manipulate the expression of a gene or alter the biological properties of living cells for therapeutic purposes. The delivery of DNA into cells can be accomplished by multiple methods.
Viral Based Gene Therapy	Uses genetically engineered viral vectors to deliver therapeutic genes to cells with genetic dysfunction. These modified viruses act as vehicles, carrying the desired genetic material into specific cells.
Cell-based gene therapy	Consists of using genetically modified cells to treat diseases. Cells can be autologous (from the patient) or allogenic (from a donor).

Record of completed work

Date	Completed work	Performed by
December 2022	First watch of the 45 new therapies in an Excel document.	Synergyx Consulting Inc.
Spring 2023	Quality control and feedback collection from various stakeholders.	Montréal InVivo and stakeholders (members and patient groups)
January 12, 2024 (first version released)	Addition of 51 new therapies for a total of 96 new therapies (July 2023 update followed by enhanced data in November 2023 following feedback). Writing of the in-depth analysis report of all therapies.	Synergyx Conseils Inc.
June 2024	Added 60 therapies for a total of 156 new therapies.	KPMG
Fall 2024	Quality control and feedback collection from various stakeholders.	Montréal InVivo and stakeholders (members and patient groups)
Fall 2024	Added 32 new therapies.	KPMG
March 3, 2025 (second version released)	Quality control, addition of additional therapies for a total of 196 new therapies, improvement of the content and writing of the in-depth analysis report of all therapies.	Montréal InVivo and stakeholders (members and patient groups)

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Stage #1: Update

The methodology used by KPMG to manually update the 96 therapies recorded in the current monitoring tool is described below. These steps were executed in sequence for each therapy of interest to collect all new approvals, submissions, and clinical trial updates.

01 Approvals Health Canada

Collect all Health Canada approvals

Search Health Canada's Drug and Health Products Portal to identify approval decisions.

02 Submissions Health Canada

Include all pending submissions by Health Canada

Search Health Canada's Drug and Health Product Submissions Under Review (SUR) database.

03 International Approvals

Identify drugs with a high probability of submission

Search FDA orphan drug designations and approvals for marketing approvals.

04 Key Clinical Trials

Update key clinical trial information

Search the Clinicaltrials.gov database for clinical trial updates (e.g., new phases, completion, key milestones, etc.).

05 Focus on Quebec

Include information specific to Quebec

Search for information about Quebec in the public registry of clinical trials (for example, Principal Investigator).

The main sources included in this update:

1. (Review decisions - Drug and Health Products Portal (hpfb-dgpsa.ca)
2. Drug and Health Product Submissions Under Review (SUR): New drug submissions under review - Canada.ca
3. Search Orphan Drug Designations and Approvals (fda.gov)
4. Home | ClinicalTrials.gov
5. https://recherchesantequebec.ca/

Note:

Reducing the number of sources allows for optimized updates, which supports the transition to automation.

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Stage #2: Search

To move away from a heavily manual search process, while adhering to the original search criteria, the methodology below has been followed by KPMG when evaluating new therapies for inclusion since the end of August 2023.

01 Databases

Identify a database (Canadian, American and European)

Discover and evaluate all available clinical trial databases with advanced search capabilities to align with key selection criteria

Clinicaltrials.gov	Orpha.net	Trialsearch.who.int
Base de données internationale	Base de données internationale	Base de données internationale
Fonction de recherche avancée	Fonction de recherche avancée	Fonction de recherche avancée
Focus sur les maladies rares	Focus sur les maladies rares	Focus sur les maladies rares

02 Detailed search

Refine search criteria to collect all desired trials

Search the WHO International Clinical Trials Registry Platform for Orphan Drugs and Genetic Therapies.

03 Additional filters

Apply additional filters

Apply additional filters to identify specific industry partners, and trials with a start date greater than September 2023.

04 Integrate into the tool

Manually adding new therapy data to the tool

Manually integrate all data into the tool and collect missing information from public sources.

05 Quality control & updating

Quality control by Montréal InVivo and updating of the tool

Review of the data by the Montreal InVivo Steering Committee and addition of 32 clinical trials identified and confirmed by the Committee, with public sources.

Search parameters used:

Main search:
- Recruitment Status: "All"
- Countries: "Canada, United States of America, European Union, United Kingdom, France, Germany, Italy, Spain, Belgium"
- Dates: "01/09/2023 - 04/06/2024"
- Phases: "Phase 2, Phase 3“
- Rare Disease/Orphan Drug: YES

Note : Two additional searches were carried out to identify other gene and RNA-based therapies.

1
- Recruitment Status: "All"
- Countries: NONE
- Dates: "01/09/2023 - 04/06/2024"
- Phases: "Phase 1, Phase 2, Phase 3"
- Genome editing: YES

2
- Intervention: "mRNA OR RNA OR Messenger RNA"
- Recruitment Status: "All"
- Countries: NONE
- Dates: "01/09/2023 - 04/06/2024"
- Phases: "Phase 1, Phase 2, Phase 3"

Quality Control

- For all versions provided by service providers, the steering committee of the Montreal InVivo Rare Diseases Committee conducted rigorous and impartial quality control. This included the following steps:
  - Validation of a sample of treatments.
    - If the information in the report was correct, the quality control was considered positive and the project moved on to the next steps to finalize the report.
    - If the information was found to be erroneous or incomplete, all the data were reviewed by the relevant stakeholders (e.g., Montreal InVivo office, patient groups, companies) and corrected. In these cases, the original information was retained for documentation purposes.

Market Intelligence

- For all versions provided by service providers, the steering committee of the Montreal InVivo Rare Diseases Committee oversaw the addition of relevant and public market information to enhance its tool. Specifically, these additions included:
  - Patient Associations (completed by: Muscular Dystrophy Canada, Regroupement québécois des maladies orphelines, CATALIS Québec)
  - Registries (completed by: Regroupement québécois des maladies orphelines)

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