# Living Rare in Canada

## Rare Diseases: Increasingly Common and Increasingly Rare

November 2025

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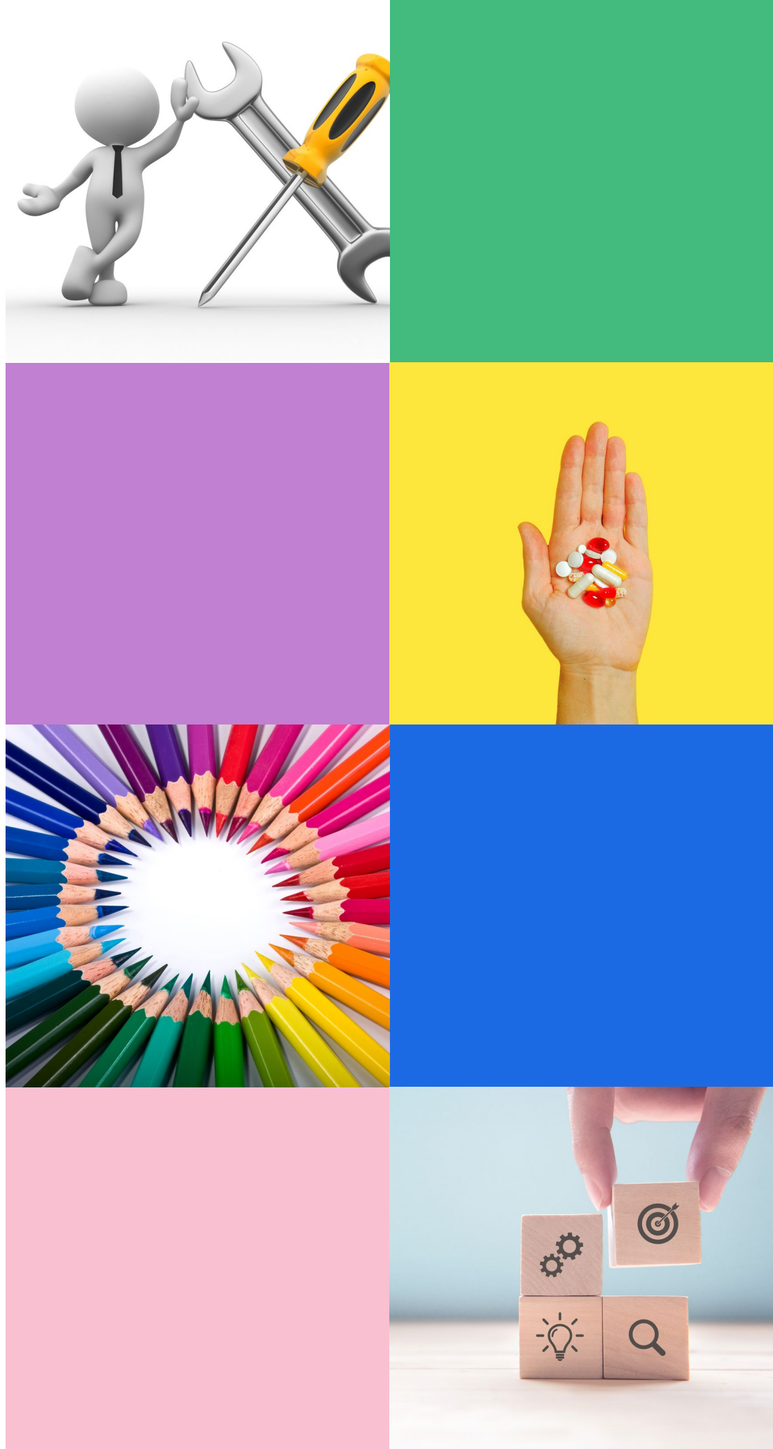
President & CEO
Canadian Organization for Rare Disorders



# What We Will Discuss

- What is a Rare Disease and Why Many Definitions
- Why Rare Diseases are Common and How Common Diseases Are Becoming Rare
- Innovative Drugs for Rare Diseases: Hype vs. Fact
- Strategies for Access: What Canada is Doing and Not
- Are Rare Disease Drugs too Expensive
- Canada’s New Rare Disease Drug Strategy: What is Working and Not
- Rare Diseases Need More Than Drugs

# How USA Orphan Drug Act Defined Rare Diseases

A CONGRESSMAN

A MOTHER

AN ACTOR

A “rare disease or condition” is one that affects fewer than 200,000 people in the U.S., or affects more but there’s no reasonable expectation the development and availability costs would be recovered from U.S. sales

# Rare Diseases in European Union: Orphan Drugs + More

- **Before vs. After EU Orphan Drug Legislation:**
  - Pre-2000: Fragmented R&D, minimal incentives.
  - 2000: Regulation (EC) No 141/2000 introduced orphan designation, centralised EMA approval, 10 years exclusivity.
  - Impact: >3,000 designations; ~260 authorised orphan medicines as of 2025; sustained investment and research growth.
- **Definition & Burden:**
  - EU definition: ≤5 in 10,000 people affected; severe, progressive, life-threatening with no approved therapy
  - Estimated 27–36 million EU citizens living with rare diseases across 6,000–8, 000 conditions.
- **European Reference Networks (ERNs):**
  - Rationale: Connect scarce expertise across borders (‘patients travel virtually’).
  - *Implementation: 24 ERNs, ~1,600+ centres in 27 MS + Norway.*
- **National Plans & Strategies**:
  - As of Nov 2025: Nearly all 27 MS have a plan or are renewing..

# Middle East Rare Disease: Indigenous Genetic Homozygous Consanguinous

## Saudi Arabia:

- SFDA orphan-drug pathway; Vision 2030 Saudi Genome Program.
- Nationwide Newborn Screening (since 2005); expanding rare-disease registries.
- Centers of excellence (KFSH&RC) and genetic-disease research hubs.

## United Arab Emirates:

- MOHAP regulatory reforms for orphan/rare-drug approvals.
- Abu Dhabi Genome & Newborn Screening Program (>800 treatable diseases).
- Emirati Genome Program powering precision medicine.

## Egypt:

- Rare Disease & Orphan Drug policy initiatives; expanding newborn screening.
- Presidential initiative for 19+ genetic disorders (since 2021).
- Building access pathways and reimbursement models for orphan therapies.

## Regional Peers:

- Qatar: Gene Therapy Center & Sidra Medicine’s rare-disease genomics.
- Bahrain: National Genome Program (100k-sample target).
- Oman: Genome & Data Programme; national NBS frameworks.
- *Kuwait: Diplomatic leadership on global rare-disease rights (UN 2025).*

# Asia Pacific Rare Disease: Varied Definitions and Benefits

| Country | Definition | Legal Framework | Benefits |
|---|---|---|---|
| **TW Taiwan** | <1 in 10,000 | Rare Disease and Orphan Drug Act (2000) | *NHIA covers 76 orphan drugs; access to special foods, diagnosis, and medication support* |
| MY **Malaysia** | No official; ~<1 in 4,000 used | None | *Limited treatment access; advocacy-driven support* |
| PH **Philippines** | <1 in 20,000 | Rare Diseases Act (RA 10747, 2016) | *PWD status benefits, Rare Disease Registry, R&D support* |
| ID **Indonesia** | No official; <2,000 cases used | None | *Minimal government support; NGOs fill gap* |
| TH **Thailand** | No official; low incidence & impact | None | *Some UCS coverage; barriers in diagnosis and treatment* |
| VN **Vietnam** | No official; WHO <10 per 10,000 used | None | *Collaborations with NGOs for care and access* |
| **SG Singapore** | <1 in 2,000 | No formal law | *Rare Disease Fund provides co-funded access to high-cost therapies* |
| **HK Hong Kong** | No official | None | *Hospital Authority support for early diagnosis and limited treatment* |
| **IN India** | <1 in 2,500 | National Policy for Rare Diseases (2021) | *Financial support, 3 disease groups, Centres of Excellence* |
| **CN China** | No official; national rare disease list (2018) | None | *Hospital network, pilot reimbursement programs, expanded access* |
| **JP Japan** | <50,000 people | Orphan Drug Law | *Priority review, 10-year exclusivity, subsidies, NHIC reimbursement* |
| **AU Australia** | <5 in 10,000 | TGA Orphan Drug Program | *Fee waivers, priority review, PBS coverage of approved drugs* |
| **CA Canada** | No official; National RD Drug Strategy <1 in 2,000 (2023) | None, Rare Disease Drug Strategy | *$1.5 billion to improve access to drugs, support early diagnosis, data collection* |

# Rare Diseases—Increasingly Common & Increasingly Rare

**7,000 to 10,000 Rare Diseases[1]**

- 200 added annually[2]
- 4,000+ identified genetic cause[1]
- 880+ orphan drug approvals for 392 rare diseases[4] benefitting 30 million patients[5]

**Orphadata 72% genetically caused[4]**

- 70% pediatric[1]
- 84.5% point prevalence < 1 in 1,000,000[3]
- 80% population burden = 4.2% most common rare diseases[3]
- Worldwide estimates 263-466 million[3]


1. EURORDIS. What is Rare Disease Available at https://www.eurordis.org/content/what-rare-disease Last accessed January 2022
2. Haendel M et al. How many rare diseases are there? Nat Rev Drug Discov 2020; 19:77-78
3. Wakap SN et al. Estimating cumulative point prevalence of rare diseases: analysis of Orphanet database. Eu J Human Genetics 2020; 28:165-173
4. https://pmc.ncbi.nlm.nih.gov/articles/PMC10290406/?utm_source=chatgpt.com
5. Gabay M. The Orphan Drug Act: An Appropriate Approval Pathway for Treatments of Rare Diseases?. Hosp Pharm. 2019;54(5):283-284. doi:10.1177/0018578719867665

# Intersection of Rare Diseases and Precisely Defined Subsets of Common Conditions

Hyperglycemia (12.2 %), T2 Diabetes (6.2%), Hyperlipidemia (42.2%), Osteoarthritis (10%)

Cancer, Stroke, Dementia, TB, HIV/AIDS, Malaria, Ebola, Sepsis

High Prevalence, Infectious and Chronic Conditions

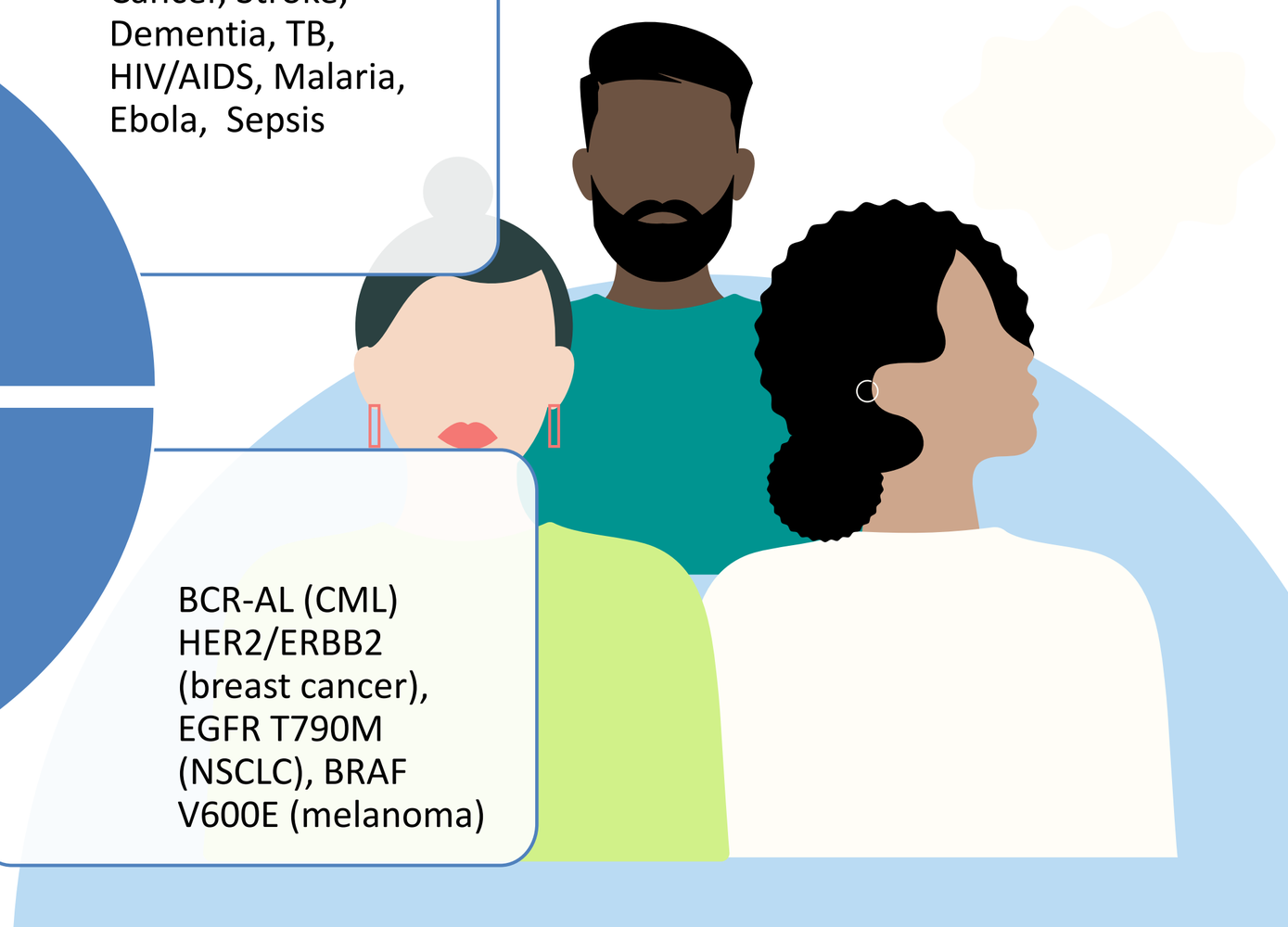
Complex, Multifactorial Causes and Multisystemic Impact

Low Prevalence, Rare and Ultra-Rare Diseases

Clinical Capability to Precisely Diagnose and Treat

Fragile X(1:4k), Marfan (1:5k), Cystic Fibrosis (1:3k), DMD (1:5k), Tays-Sach (1:320k), Progeria (1:4m)

BCR-AL (CML) HER2/ERBB2 (breast cancer), EGFR T790M (NSCLC), BRAF V600E (melanoma)



# Common Diseases: Increasing Rare Genomic Mutations

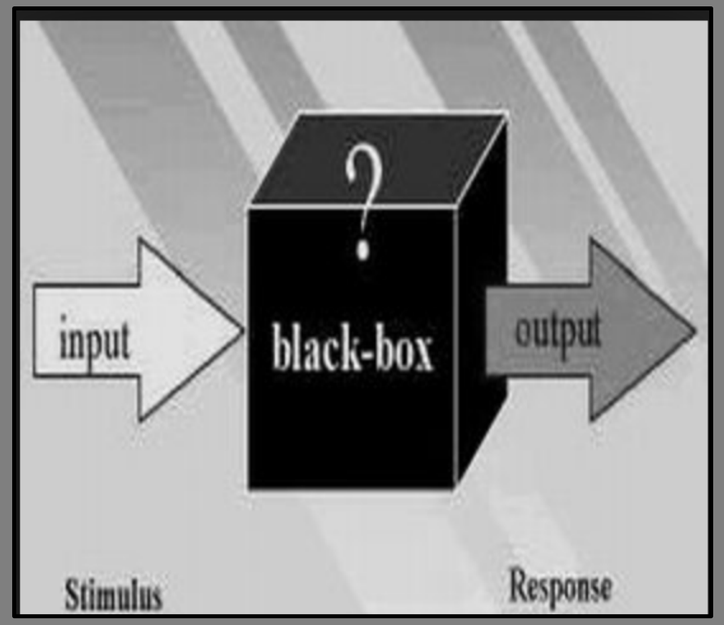
## 10s of 1000s of Genetic/Genomic Mutations for Common Conditions

- 100,000+ genome mutations across various types of cancers[1]
- 5,000+ genetic variants for CAD, FH, CM, and arrhythmias[2]
- 1,000+ genetic variants  for type 1 and type 2 diabetes[3]
- 1,000+ genetic mutations in neurodegenerative conditions[4]
  - 20 genetic mutations linked to Alzheimer’s; 200+ influence
  - 90 mutations linked to Parkinson’s; 15 influence

1. The Cancer Genome Atlas (TCGA) and the International Cancer Genome Consortium (ICGC)
2. ClinVar, OMIM8
3. GWAS;  dbSNP and ClinVarGabay M.
4. AlzGene and PDGene

Promise of Innovative Transformative Therapies

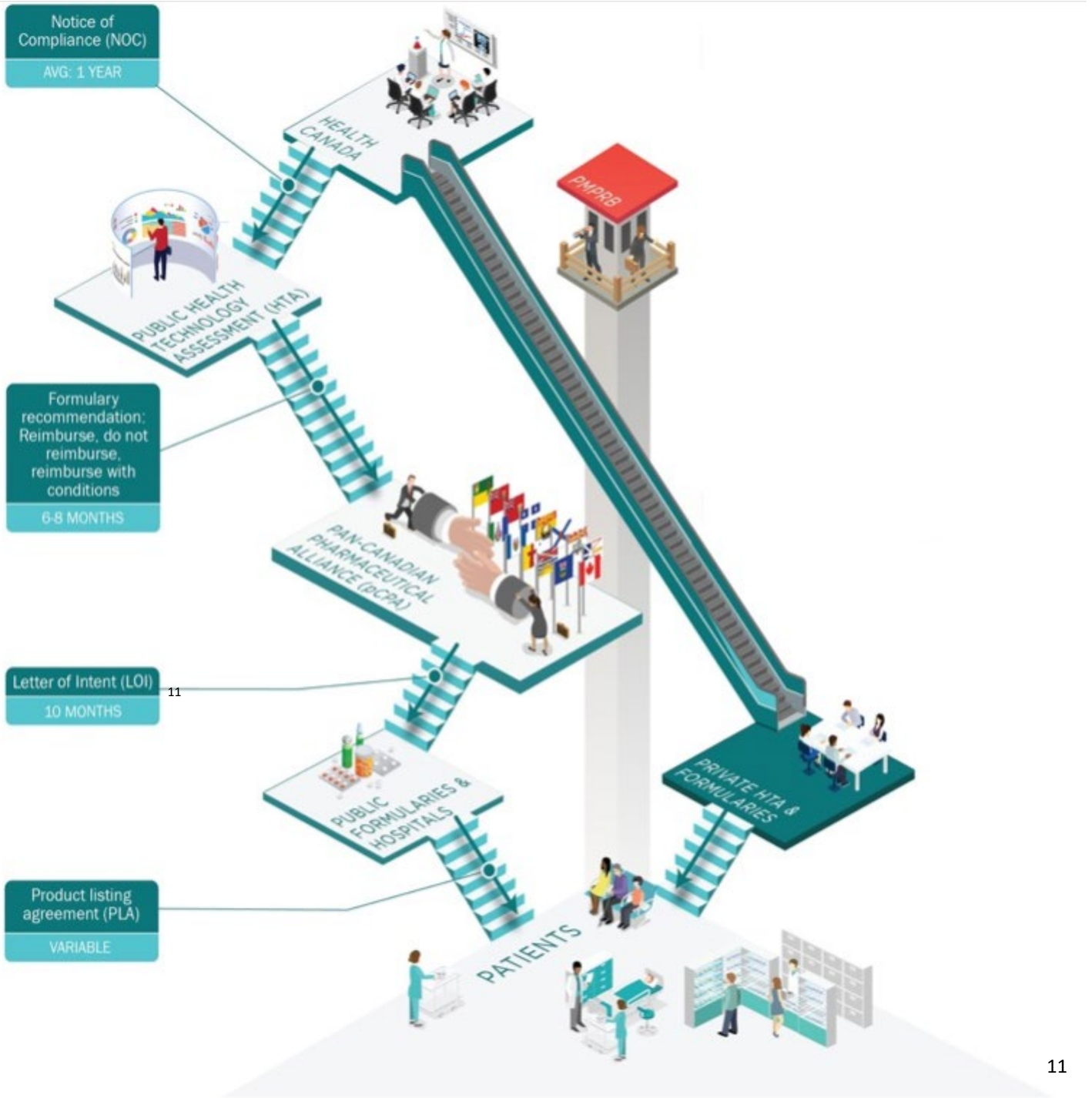
Reality of Access to Breakthrough Medicines



# How medicines are reviewed / funded

- **Four major steps** for <u>government</u> review and funding decisions

- **2/3rds of Canadians have coverage under private benefits programs** – which provide faster coverage for more medicines



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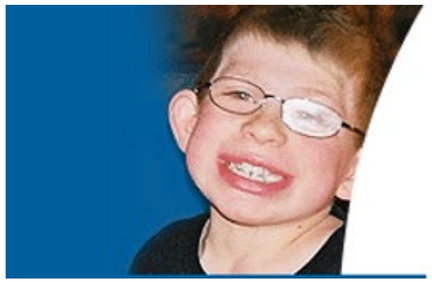
# CDR Denies Fabry & MPS-1 Drugs

Fabry and MPS patients protest at ?FPT Health Ministers' Annual Meeting; demand access to life-saving drugs

canadian**fabry**association
l'association canadienne de **fabry**



MPS
*The Canadian Society for Mucopolysaccharide & Related Diseases Inc.*

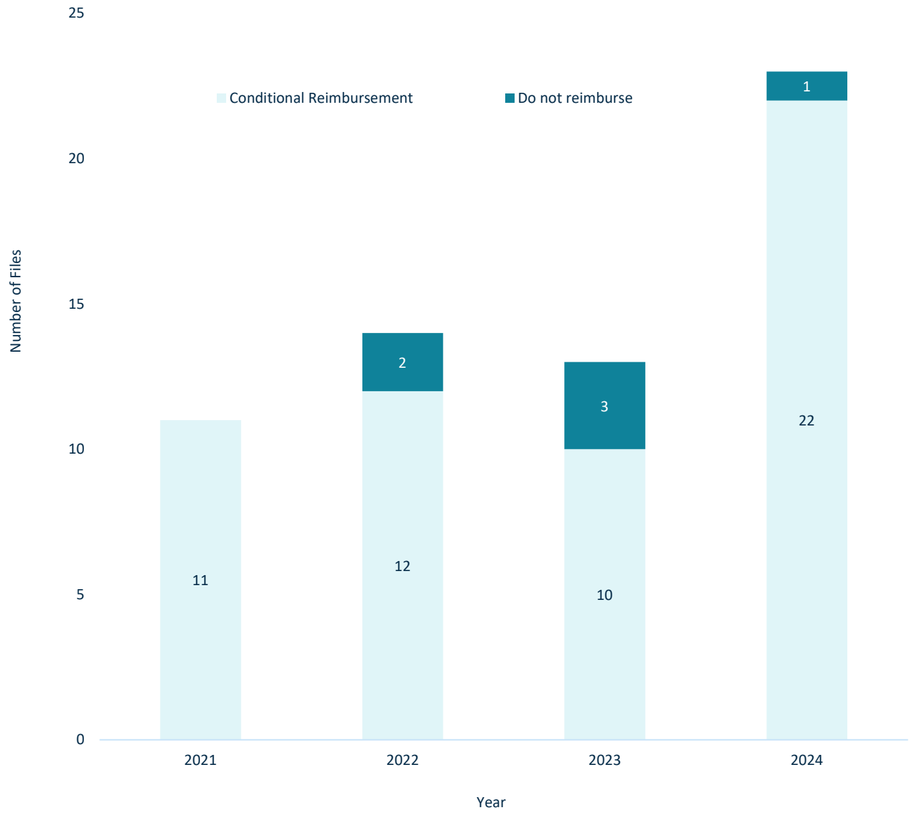
Solution: Fabry's Disease and MPS-I, time-limited research program meeting treatment guidelines" on a risk sharing basis.

Pilot for EXPENSIVE DRUGS FOR RARE DISORDERS?

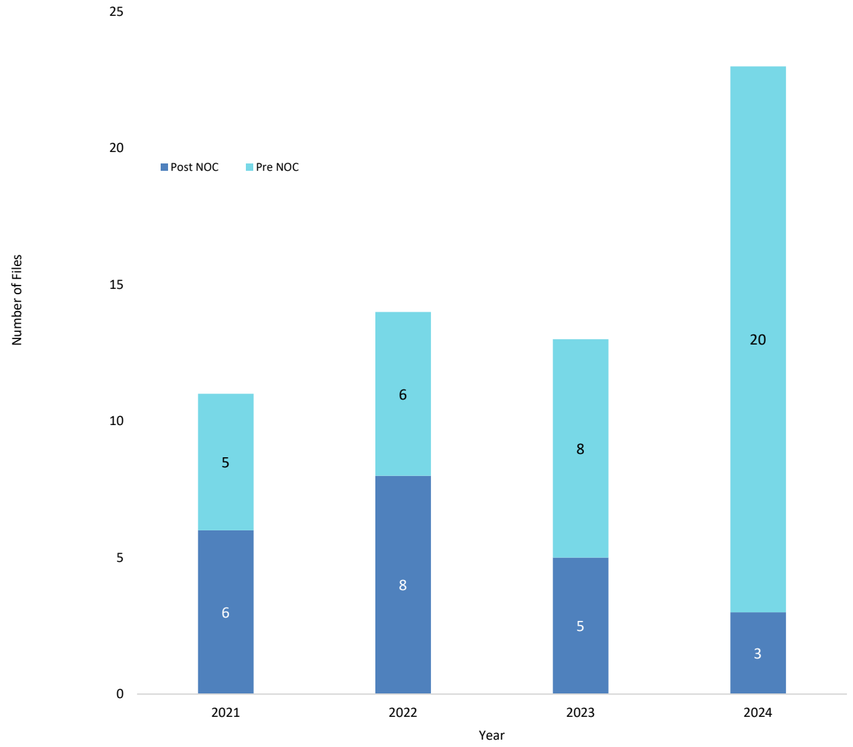
# GOOD NEWS: Last 4 years: 90% of 61 rare diseases (DRDs) reviewed by CDA received positive recommendations & 64% were submitted pre-NOC

Pre-2018, less than 70% of DRDs received positive CDA-AMC recommendations.

## Number of DRD Files Reviewed By CDA-AMC




## Percentage of DRD Files submitted to CDA-AMC pre- or post-NOC




Note: This analysis is based on the methodology used for MORSE's CRaFT DRD Sub-report; oncology and blood products are not included.

# CDR “Yes” but Not at This Price = Delayed Access

## Girl, 12, pleads for province to fund expensive drug for cystic fibrosis

**Canada AM: Young girl makes a plea to premier**

Beth Vanstone, Madi's mother says the Ontario government is putting a price on people's lives by charging such expensive fees for the drug.

**CTV Toronto: Girl goes to premier with questions**

Colin D'Mello meets a 12-year-old girl who's taking on the government: Why won't Ontario cover the costs of a pricey cystic fibrosis drug?

**CTV Barrie: Girl asks premier for OHIP coverage**

Beeton girl asks Ont. premier for OHIP coverage for expensive drug that could eliminates cystic fibrosis symptoms. Katherine Ward reports.

# Timelines for Rare Drug Funding Still Very Long : Almost 2 years After HC Approval

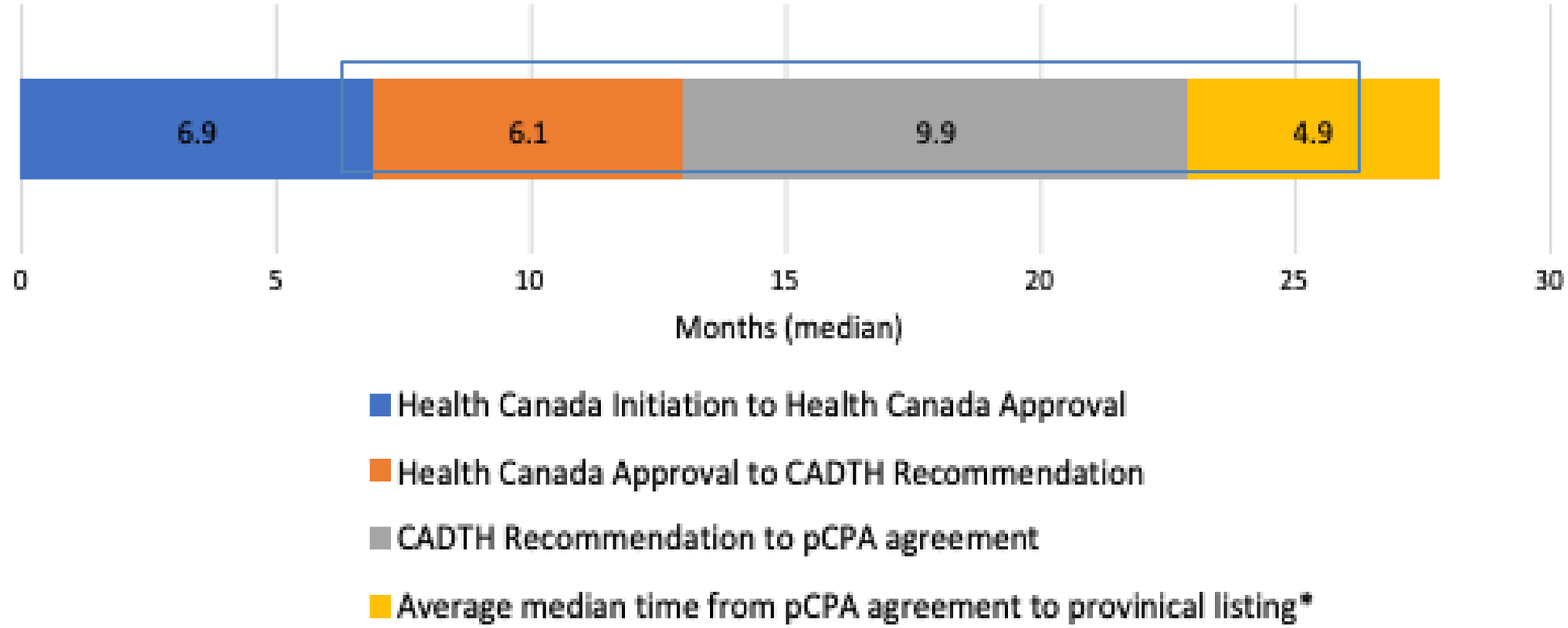
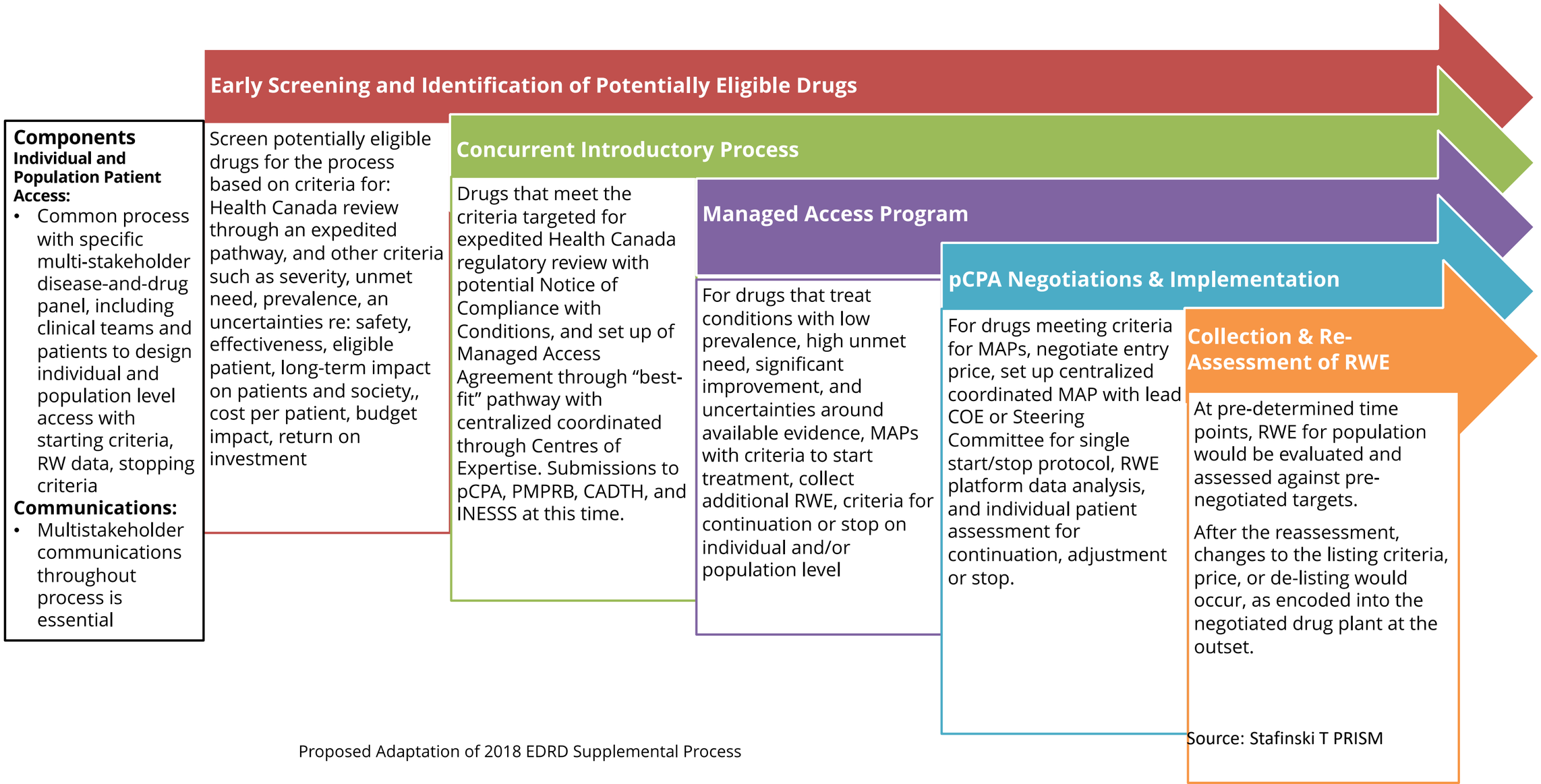



**Fig. 8** Median duration between steps in the Canadian drug access pathway. *PEI was excluded from the average provincial median timeline calculation because the timeline for PEI was only based on 2 drugs being reimbursed

Source: Ward *et al. Orphanet Journal of Rare Diseases (2022) 17:113:* https://doi.org/10.1186/s13023-022-02260-6

# Proposed Alternative Pathway Addressing Uncertainty




Proposed Adaptation of 2018 EDRD Supplemental Process

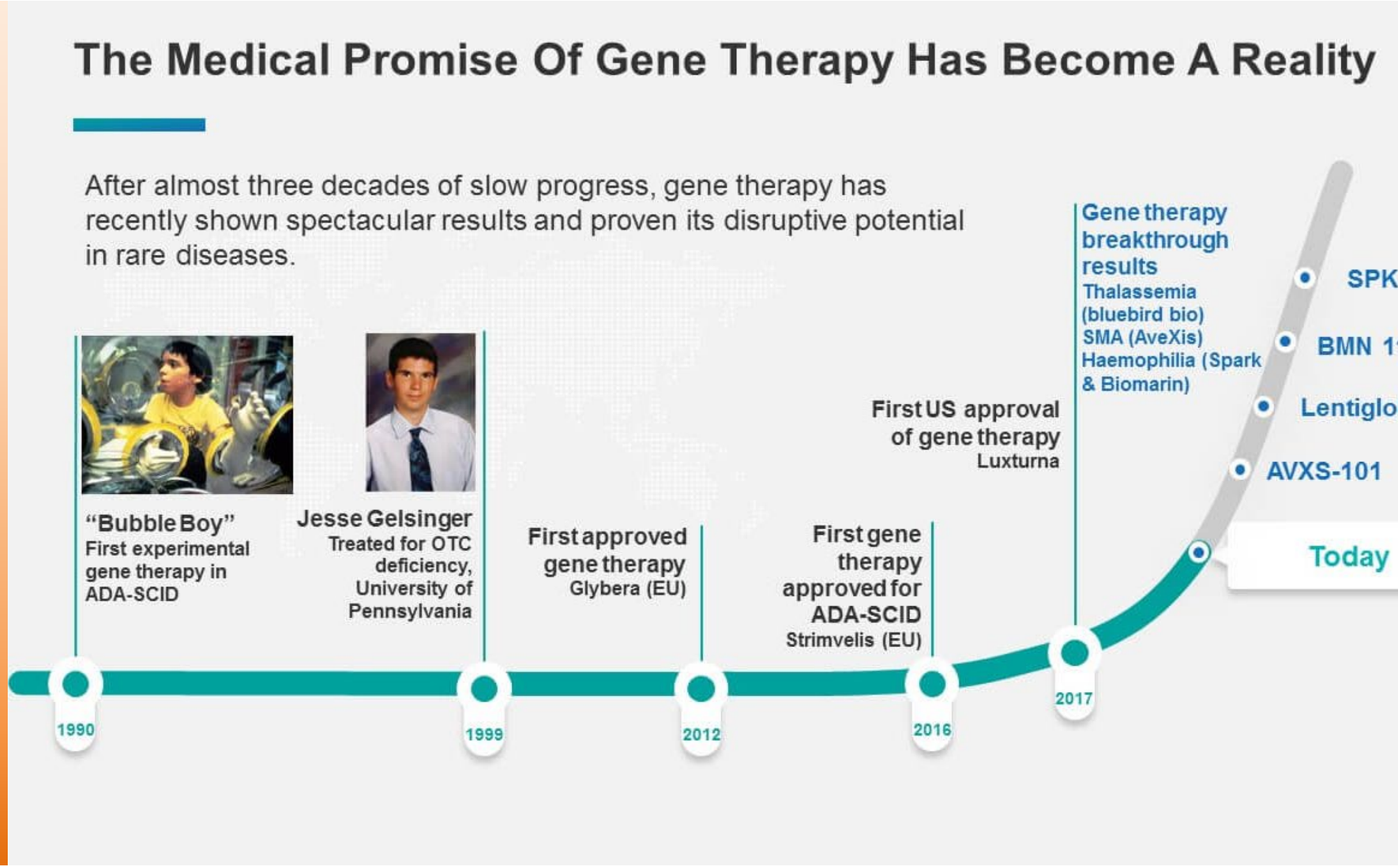
# RWE-Based Decision Options/ Managed Access Pathways

| Country | Types of RWE based assessments |
|---|---|
| France | • Financial-based reimbursement schemes<br>• Conditional approval with request for data collection and re-assessment |
| Germany | • Performance and financial-based reimbursement schemes<br>• Assessment of real-world benefits of all orphan drugs 12 months post-market authorization using disease-based registries |
| Italy | • Performance and financial-based reimbursement schemes<br>• AIFA Monitoring Registries |
| Spain | • Financial-based reimbursement schemes<br>• Catalan Managed Access Programme |
| United Kingdom | • Performance-based reimbursement schemes (managed access agreements)<br>• Financial outcomes based reimbursement schemes |

Source: Stafinski T PRISM

# GENE THERAPIES: CURES AT ANY COST?

- US FDA estimates: 10-20 new gene therapies by next year
- This doesn't include:
  - Therapies that don't involve genes or cells e.g. enzyme replacement therapy
  - Therapies for hard-to-treat or rare cancers



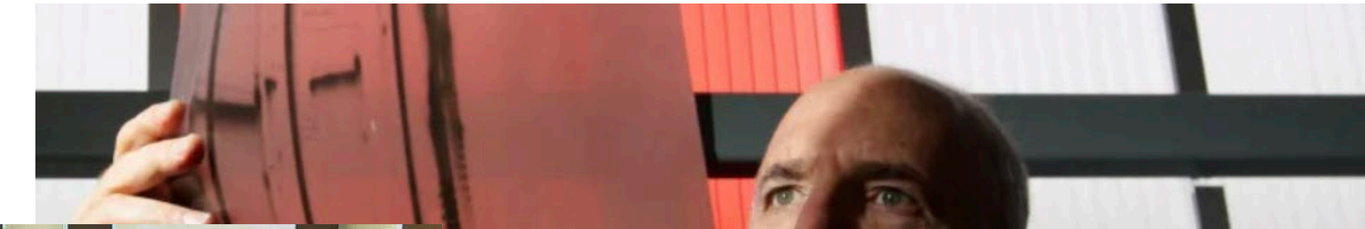
# Can other gene therapy developers avoid Glybera's fate?

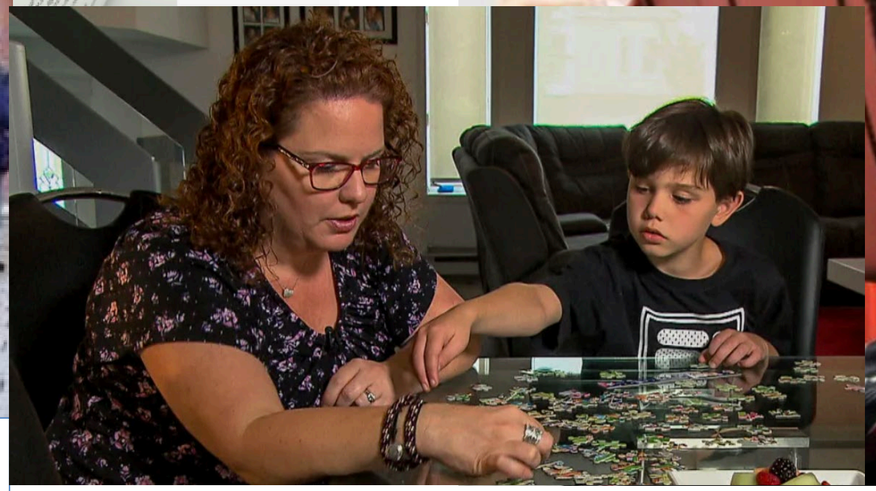
By **Tracy Staton** • May 4, 2016 10:55am

Cell & Gene Therapy | gene therapy | Chiesi | GSK


"Last" manufactured dose of gene therapy Glybera was administered to 38-year-old German Postman diagnosed with lipoprotein lipase deficiency" who suffered severe attacks of pancreatitis. 3-years later, he remains symptom-free.

# Canadian breakthrough that became the world's most expensive drug, then vanished, gets second chance







Dr. Michael Hayden, director of the Centre for Molecular Medicine and Therapeutics at the Child & Family Research Institute in Vancouver, will be leading the Canadian scientific team as they re-engineer Glybera. (Darryl Dyck/The Canadian Press)

# Life for Lucy - Conquering SMA

TEAM FUNDRAISER

Scott Van Doormaal and 3 others are organizing this fundraiser.

Created May 10, 2020 | Medical, Illness & Healing

**LUCY IS GETTING ZOLGENSMA!**

**Thank you for all the love & support. We are so grateful to each and every one of you.**

**$2,471,100** raised

Share

The organizer has currently disabled new donations to this fundraiser.

Anonymous
**$25** · 1 mo

Anonymous
**$200** · 1 mo

Minette Laffer
**$40** · 1 mo

See all

# Race against time for family of toddler with ultra-rare disease, whose only chance at life costs $3 million

*The Toronto family started a GoFundMe campaign, raising almost $700,000 since it was posted at the start of May but it still isn't enough to save their child*

**Bobby Hristova**
Oct 02, 2019 • Last Updated 11 months ago • 4 minute read

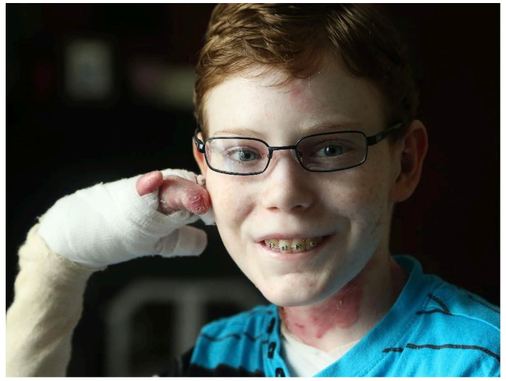
# 3-year-old Spinal Muscular Atrophy Twins: No Newborn Screening, Misdiagnosis, Lack Timely Specialist Care, Delayed Treatment, Too Old for Gene Therapy

TEAM FUNDRAISER

April Chan and 2 others are organizing this fundraiser on behalf of Kimsaung Sov.

# Ottawa's 'butterfly boy,' Jonathan Pitre, dies at age 17

2000 - 2018



He had one of the most painful diseases known to medicine, epidermolysis bullosa (EB), but was defiantly happy. He couldn't scratch without tearing his skin, but dreamed of playing hockey. Ostracized by other children, he thrived in the adult world: poised, thoughtful and genuine. In 2016, Jonathan received experimental stem cell transplant but suffered many infections.

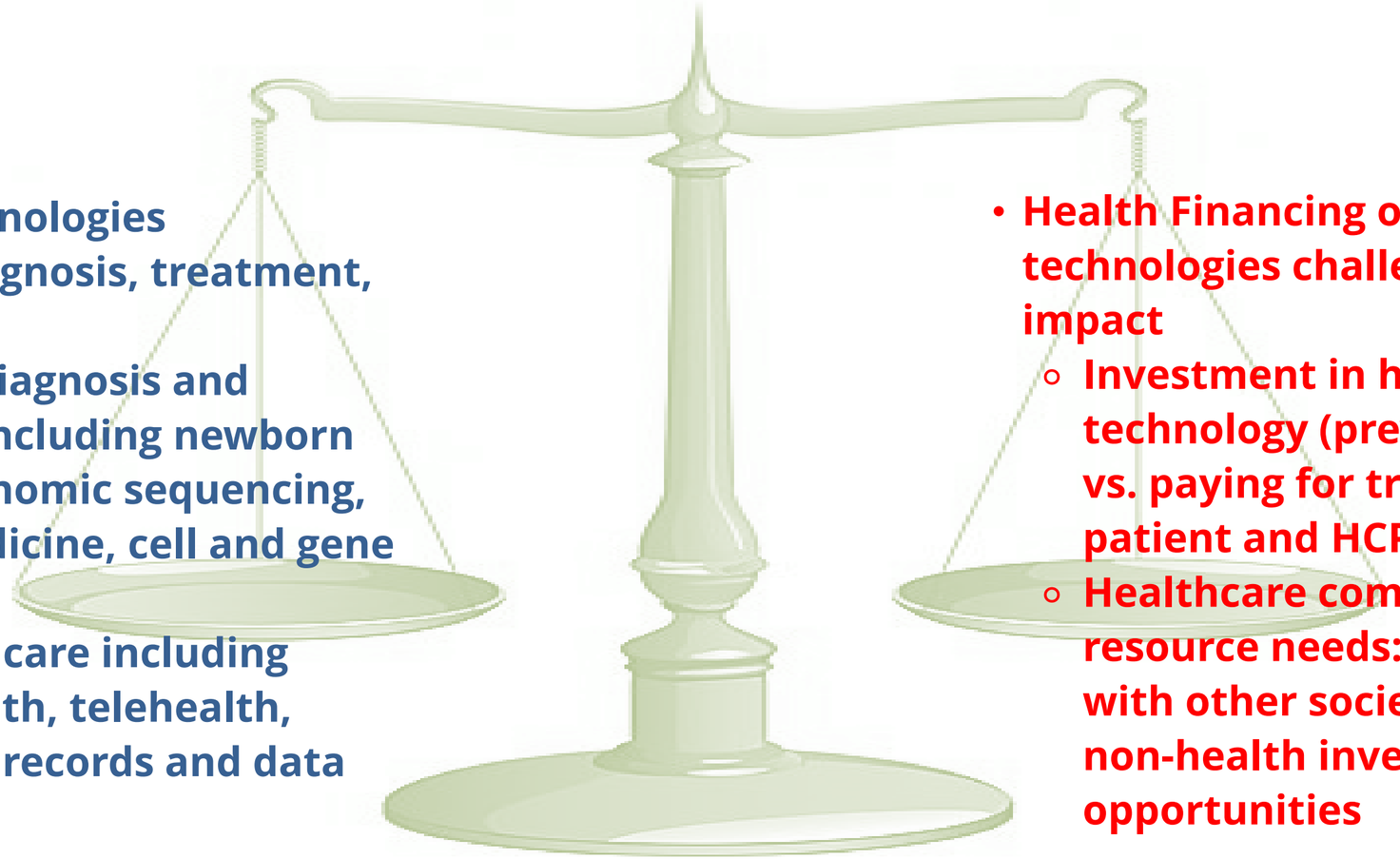
May 2023

## First-of-its-kind gene therapy approved for healing wounds in "butterfly children"

Children born with a rare disease that makes their skin as fragile as a butterfly's wings, leaving them vulnerable to frequent wounding and constant pain, will finally have access to a new treatment that aims to fix the genetic cause of the condition.

The therapy, known as Vyjuvek and made by Krystal Biotech, was approved by the FDA on Friday afternoon for treating patients six months or older with dystrophic epidermolysis bullosa, or DEB. The drug is a long-awaited solution for many families afflicted by the hellish condition, for which there are no approved treatments in the US.

# New Technology or Balanced Budgets?
# Or Can We Have It All



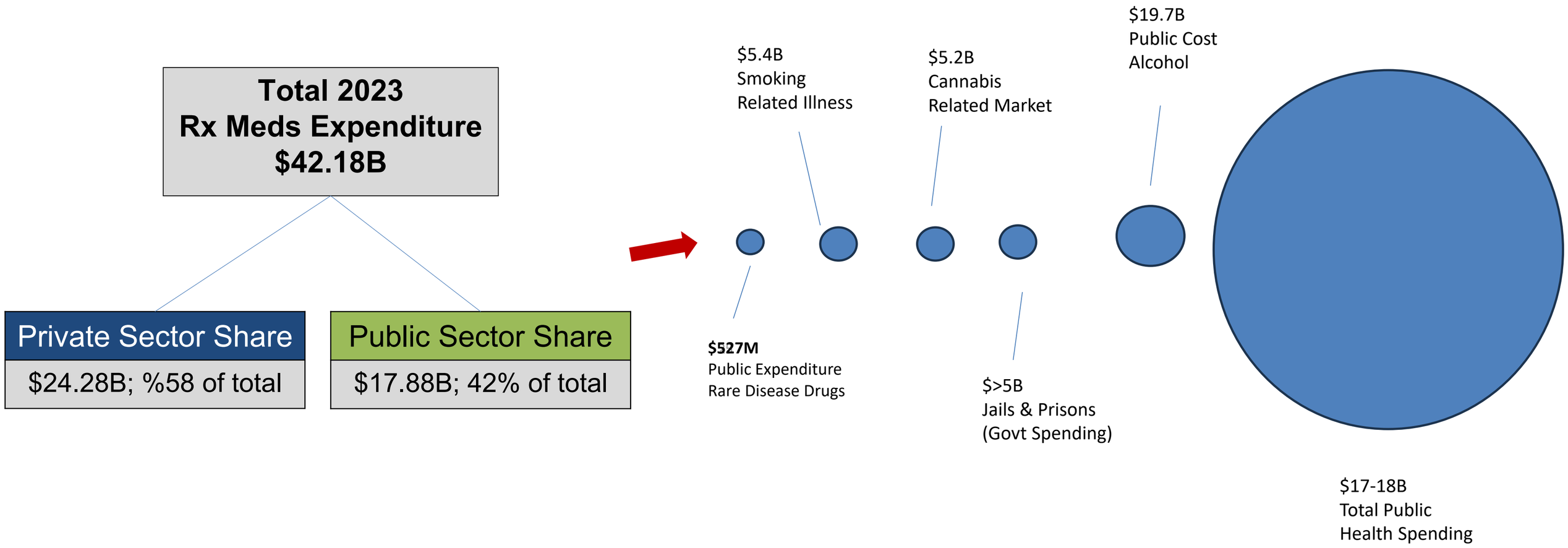
- **Innovative Technologies accelerating diagnosis, treatment, and prevention**
  - **Gene-based diagnosis and treatments, including newborn screening, genomic sequencing, precision medicine, cell and gene therapies**
- **Digital-enhanced care including wearables, e-health, telehealth, electronic health records and data registries**

- **Health Financing of innovative technologies challenged by budget impact**
  - **Investment in health fueled by technology (prevention, well-being) vs. paying for treatment driven by patient and HCP needs**
  - **Healthcare competing with other resource needs: among diseases, with other societal priorities, with non-health investment opportunities**

# Financial Impact on Health Budgets: Fears/Hype vs. Reality/Evidence

- Hype: Durable cell and gene therapies potentially transform patient lives, but payers fear unsustainable costs arising from the more than 1000 therapies in development pipeline.
- Model-based Reality: Est. 63.5 cumulative US product indication approvals through 2030, with 93 000 patients treated in 2030 generating US$24.4 billion list price product revenues (costs) not including ancillary medical costs or cost offsets.
  - Cancer therapies receive US$16.9B (69%) of 2030 revenues,
  - Gene therapies for all other conditions receiving US$7.5B (31%).
  - Average revenue (cost) per patient treated by a durable gene therapy (non-oncology) is US$149 000.
  - 88% of patients, average cost: 44 700 to 50 700: musculoskeletal conditions, such as joint osteoarthritis (US$50 000) and wet AMD, with comparatively low price of $100 000,

# Who pays for meds and how much for rare…




Source: (2019) Forte et al, *The Current and Future Cost of Orphan Drugs in Canada* (Poster).

# National Drugs for Rare Diseases

Government of Canada | Gouvernement du Canada

**Government of Canada improves access to affordable and effective drugs for rare diseases**


new three-year, $1.5-billion national rare disease treatment strategy announced in March 2023

*National Strategy for Drugs for Rare Diseases*

- The goal of the first three-year phase is to increase access to, and affordability of, effective drugs for rare diseases, which will contribute to improving the health of patients across Canada
- Lessons learned will be incorporated into future phases, staying aligned with the government's broader pharmaceutical agenda

**Objectives:**

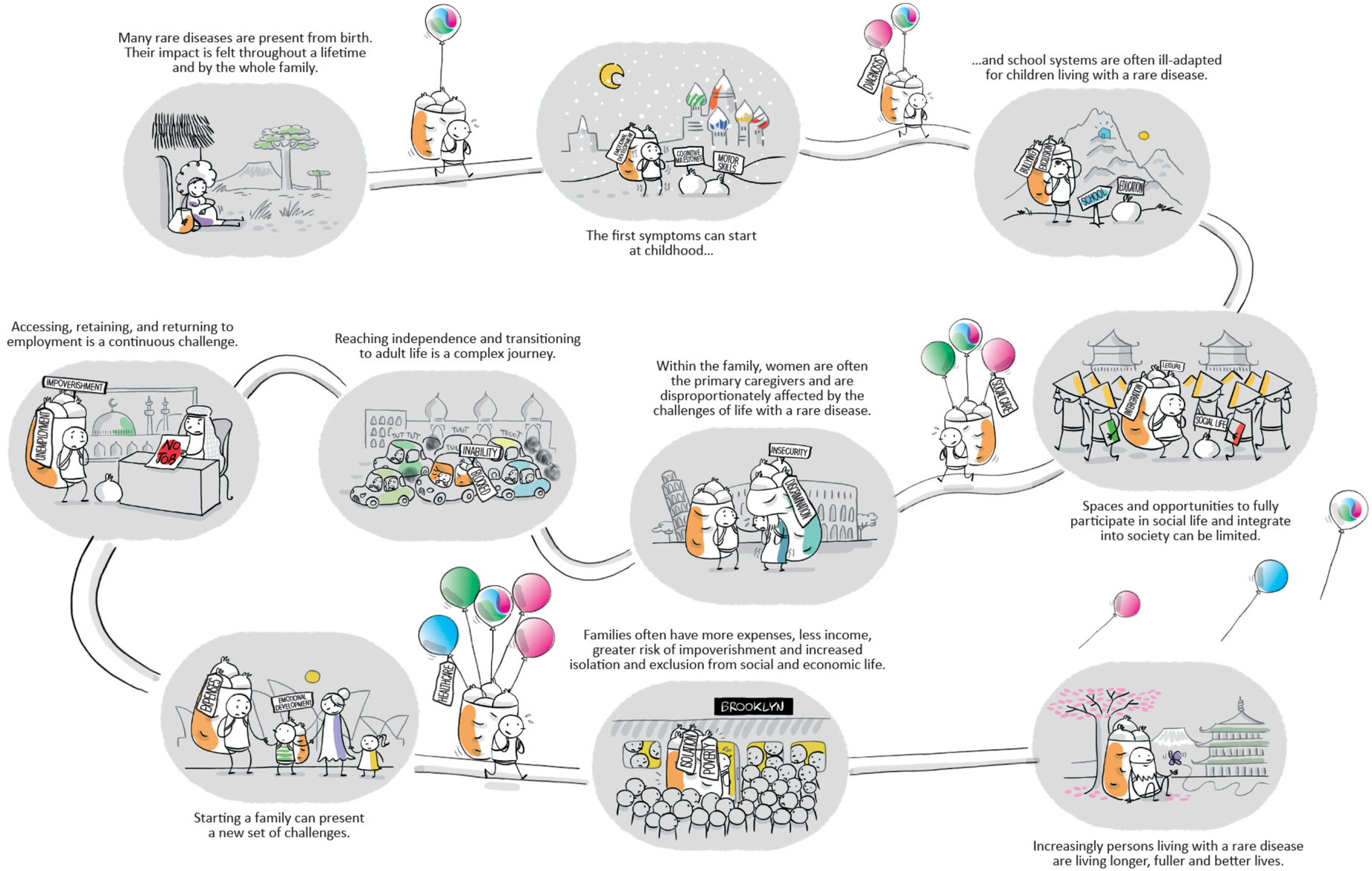
# Provinces/Territories Receive $1.4 B for Rare Drugs

| Province/Territory | Date Signed | 3-Year Budget | Coverage |
|---|---|---|---|
| British Columbia | July 23, 2024 | $194M | *Poteligeo, Oxlumo, Eplinky, Welireg, Yescarta, Koselugo, Sohonos, Imcivree* |
| Newfoundland & Labrador | Nov 15, 2024 | $22M | *Poteligeo, Oxlumo Eplinky, Welireg, Yescarta* |
| Alberta | Dec 5, 2024 | $162M | *Poteligeo, Oxlumo, Eplinky, Welireg, Yescarta, Koselugo, Sohonos* |
| Saskatchewan | Jan 10, 2025 | $40M | *Poteligeo, Oxlumo, Eplinky, Welireg, Yescarta, Koselugo, Sohonos* |
| New Brunswick | Jan 15, 2025 | $32M | *Poteligeo, Oxlumo, Eplinky, Welireg* |
| Ontario | Jan 24, 2025 | $535M | *Poteligeo, Oxlumo, Eplinky, Welireg, Yescarta, Koselugo, Sohonos* |
| Manitoba | Feb 27, 2025 | $48M | *Poteligeo, Oxlumo, Eplinky, Welireg, Yescarta* |
| Prince Edward Island | Mar 7, 2025 | $3.43M | *Poteligeo, Eplinky, Koselugo* |
| Northwest Territories | Mar 13, 2025 | $7.8M | *Poteligeo* |
| Nunavut | Mar 13, 2025 | $8.5M | *PoteligeoEplinky, Welireg, Yescarta* |
| Yukon | Mar 13, 2025 | $8.5M | *Yescarta* |
| Nova Scotia | Mar 20, 2025 | $39M | *Poteligeo, Oxlumo,, Eplinky, Welireg, Yescarta, Koselugo* |
| Quebec | Mar 21, 2025 | $305M | Program focus |

Rare Disease Journey: 4+ years to diagnosis; misdiagnosis = wrong treatment; few and distant specialists; delayed drug access; lack integrated supportive services

# A JOURNEY THROUGH LIFE WITH A RARE DISEASE

RARE DISEASES INTERNATIONAL



# UN Declaration on Rare Disease:

## Address Social, Economic, Educational, Work, Mental Health  Needs to Leave No One Behind

Inclusion & Integration is difficult and/or impossible in mainstream educational systems.[1]

The whole family experiences increased impoverishment due to more expenses related to care.[1]

Women experience gender inequality as persons with a rare disease and as mothers who often become the primary caregiver.[2]

Young adults experience difficulties with every step of independent living, from finding, keeping or returning to a decent work.[3]

There is a lack of medical expertise and a need for public awareness of rare disease diseases.[1]

COVID-19 has aggravated inequalities and social exclusion.[4]


RDI owned photograph


1. Report from Rare Disease Day Policy Event at the United Nations. Second High Level Event for the NGO Committee for Rare Diseases. Published May 2019
2. Report for the inauguration of the NGO Committee for Rare Disease. Published February 2017
3. Patient testimonials to the RDI. Email January 2022
4. RDI Statement on COVID-19 response and recovery. Published July 2020

# CORD Proposes Canada's Rare Disease Strategy: 5 Key Pillars

- Improving early detection and prevention
- Providing timely, equitable and evidenceinformed care
- Enhancing community support
- Providing sustainable access to promising therapies
- Promoting innovative research

# Canadians Still Advocating for Comprehensive Rare Disease Strategy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