

# Driving Advances In Rare Disease Diagnosis, Care And Treatment



**FRAGILE**

FRAGILE X RESEARCH  
FOUNDATION OF CANADA

## FRAGILE X SYNDROME

**1 in 4000 males**

**1 in 8000 females**

#1 known genetic cause of autism in males

Unique inheritance pattern

### PREMUTATION CARRIERS:

**1 in 150 - 300 women are carriers**

**1 in 400 - 850 men are carriers**

**Karen Kelm**, PRESIDENT

karen@fragilexcanada.ca

## SUPPORTING IMMEDIATE NEEDS OF INDIVIDUALS

## IDENTIFYING THE EXPERTISE OF PARENTS

- Redefining care pathways that operationalize the WHO's ICF Framework of health
- Removing the load families and care providers bear

## BUILDING PARTNERSHIPS AND COLLABORATIONS

- Families
- Researchers and clinicians
- Healthcare
  - Physician Learning Program, Office of Lifelong Learning - University of Alberta
- Medical schools
  - Sharing the family journey – University of Calgary
  - Curriculum development – McMaster University
- Public Policy

# IMPROVING CARE IN FRAGILE X SYNDROME AND BEYOND

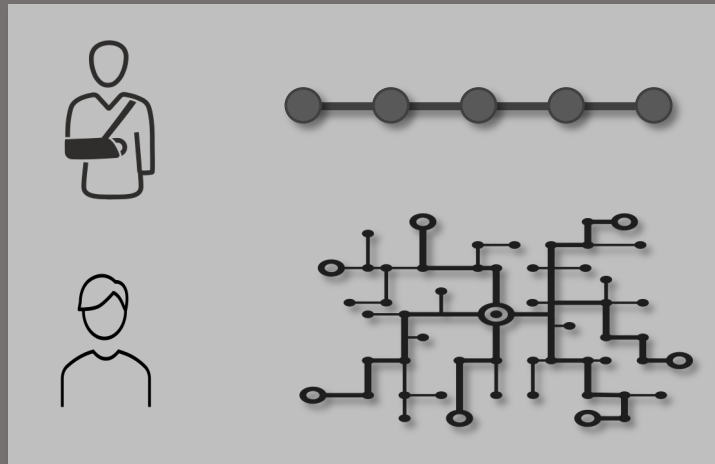
## Mental models

How someone experiences and understands the world around them.



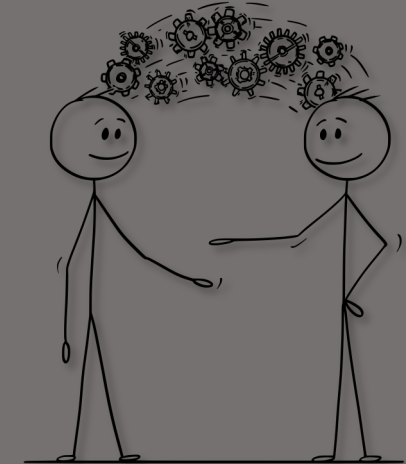
## Health literacy

“the ability to access, understand, evaluate, and communicate information in a way to promote, maintain and improve health in a variety of settings across the life-course.”



## Knowledge transfer

Sharing knowledge between individuals.



**Kelm, K., & Bolduc, F. V. (2021).** *How Knowledge Mapping Can Bridge the Communication Gap Between Caregivers and Health Professionals Supporting Individuals With Complex Medical Needs: A Study in Fragile X Syndrome.* *Frontiers in psychiatry*, 12, 731011. <https://doi.org/10.3389/fpsy.2021.731011>

Rootman, I. *A Vision for a Health Literate Canada: Report of the Expert Panel on Health Literacy.* (2008).

Sturgiss, E., Luig, T., Campbell-Scherer, D. L., Lewanczuk, R. & Green, L. A. Using Concept Maps to compare obesity knowledge between policy makers and primary care researchers in Canada. (2019).

Fowler, J. *The Power Of Mental Models: How To Make Intelligent Decisions, Gain A Mental Edge And Increase Productivity.* (2019).

## FRAGILE X SYNDROME

**1 in 4000 males**

**1 in 8000 females**

#1 known genetic cause of autism in males

Unique inheritance pattern

### PREMUTATION CARRIERS:

**1 in 150 - 300 women are carriers**

**1 in 400 - 850 men are carriers**

Acknowledge the challenges associated with Fragile X syndrome but also draw upon the strengths and assets of individuals and all of those involved on the care team.



...AND BEYOND

**WE NEED TO HUMANIZE CARE  
SEE INDIVIDUALS FOR WHO THEY ARE  
– NOT FOR WHO THEY ARE NOT**

**FRAGILE**  
FRAGILE X RESEARCH  
FOUNDATION OF CANADA