



# Fragile X in British Columbia

Past, present and future

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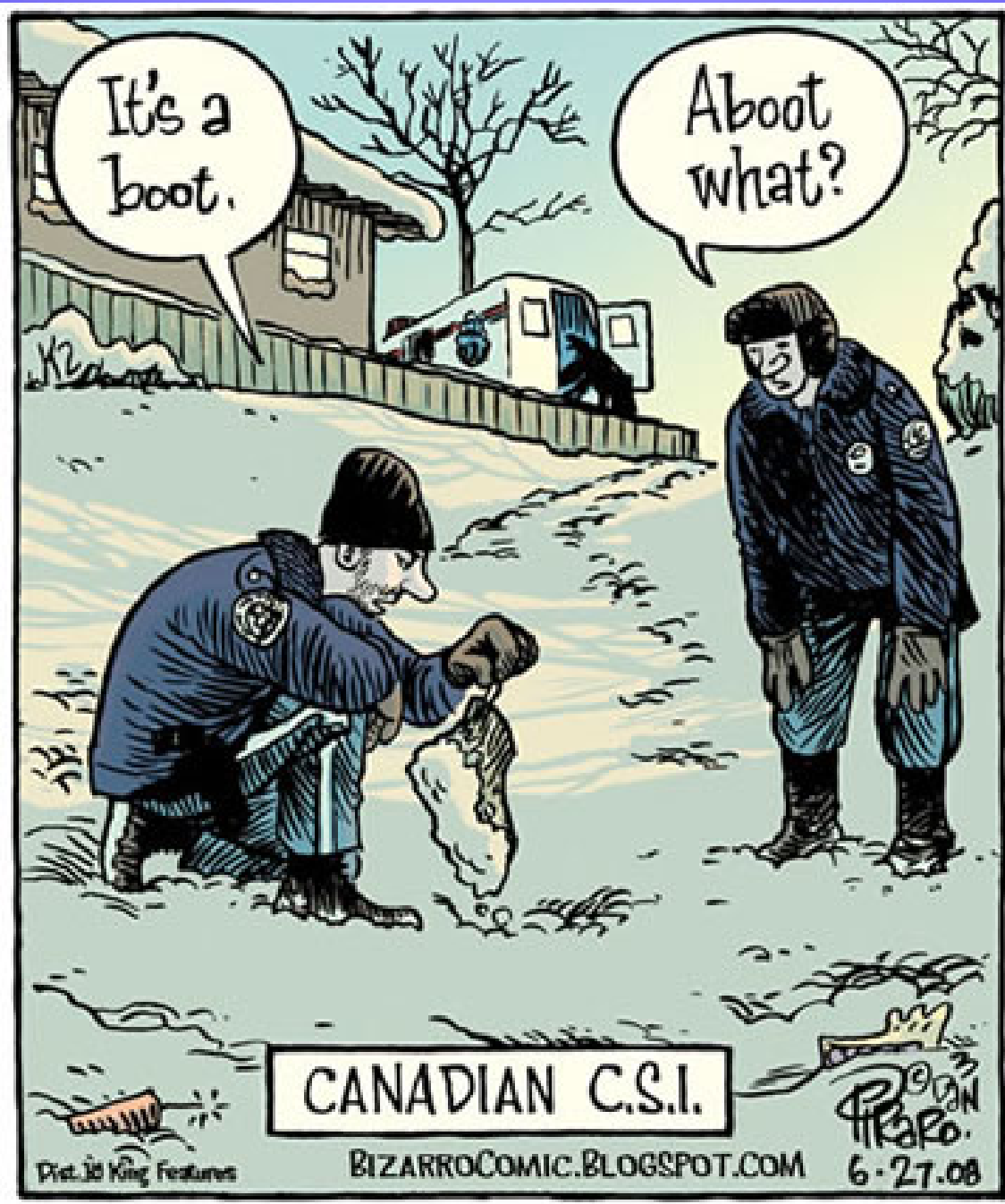
"Matthews ... we're getting another one of those strange 'aw blah es span yol' sounds."

# Learning Objectives

1. To learn about the population of British Columbia, and the prevalence of fragile X syndrome.
2. To learn about current delivery of care, and barriers, for families with fragile X syndrome in BC.
3. To learn about the Canadian Pediatric Surveillance Program.

# Outline

1. Introduction to British Columbia
2. Fragile X syndrome diagnoses in BC
3. Current clinical care for people with FXS in BC
4. Canadian Pediatric Surveillance Program
5. Future state



It's a boot.

About what?

CANADIAN C.S.I.

Prod. 30 King Features

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P. Bizarro.  
6-27-08

# British Columbia



# British Columbia

- Population 4.5 million
- 40,000-45,000 births per year
- Ethnic demographic:
  - 10% Chinese
  - 6.4% South Asian
  - 6% other Asian
  - 4.8% Aboriginal

# Children's health care in BC

- The provincial health authority oversees provincial programs many of which are distributed throughout the province geographically eg. autism assessment, cancer care, cystic fibrosis, etc.
- Family practitioners are the 'medical home' for most children
- General pediatricians perform consultations, and provide primary care for children with complex or chronic medical conditions





UBC Department of Medical Genetics,  
BC Children's Hospital, Vancouver



Vancouver Island Medical Genetics,  
Victoria General Hospital











# What the U.S. thinks about Canada?



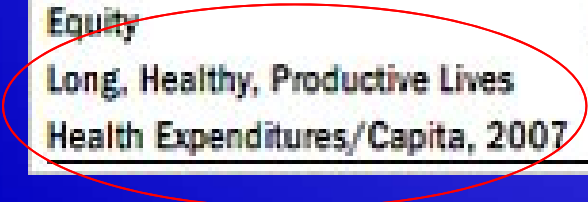
# The Commonwealth ranking 2010

Exhibit ES-1. Overall Ranking

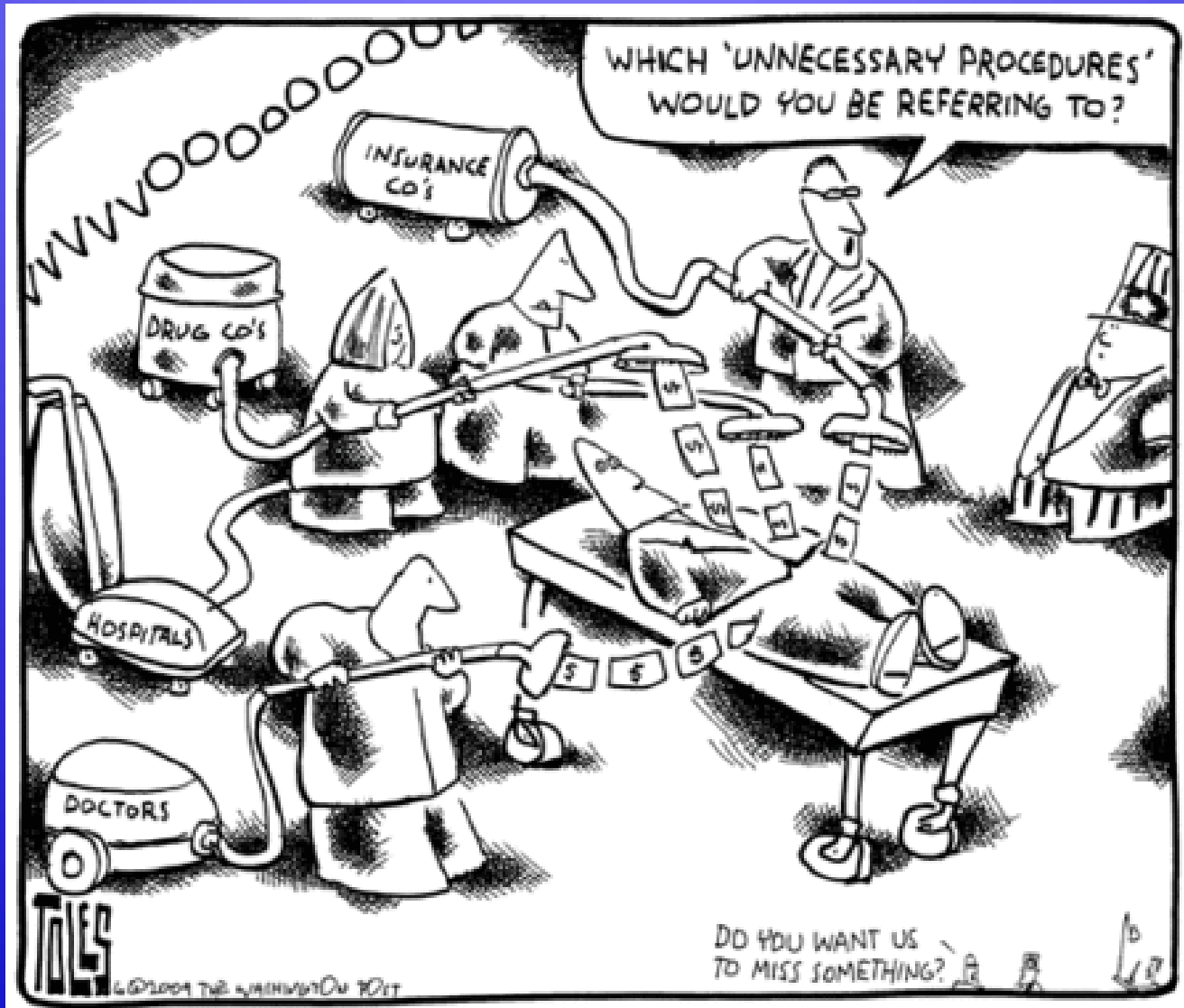
Country Rankings	
	1.00-2.33
	2.34-4.66
	4.67-7.00



	AUS	CAN	GER	NETH	NZ	UK	US
<b>OVERALL RANKING (2010)</b>	3	6	4	1	5	2	7
<b>Quality Care</b>	4	7	5	2	1	3	6
Effective Care	2	7	6	3	5	1	4
Safe Care	6	5	3	1	4	2	7
Coordinated Care	4	5	7	2	1	3	6
Patient-Centered Care	2	5	3	6	1	7	4
<b>Access</b>	6.5	5	3	1	4	2	6.5
Cost-Related Problem	6	3.5	3.5	2	5	1	7
Timeliness of Care	6	7	2	1	3	4	5
<b>Efficiency</b>	2	6		3	4	1	7
<b>Equity</b>	4	5	3	1	6	2	7
<b>Long, Healthy, Productive Lives</b>	1	2	3	4	5	6	7
<b>Health Expenditures/Capita, 2007</b>	\$3,357	\$3,895	\$3,588	\$3,837*	\$2,454	\$2,992	\$7,290



# What Canada thinks about the U.S.



# Molecular diagnoses in BC

- One molecular genetics lab performs fragile X testing for the province.
- On average, approximately 1000 samples are submitted annually.
- Records from a 9 year span were reviewed to ascertain number of full mutation diagnoses, and demographic characteristics.



# Results

- 93 full mutation results
  - 93/9000 samples = approx 1% detection rate
- Children under age 18 yrs: n=50
- Adults 18 +: n=43
  - 8 men with no reported family history, average age 47 yrs (23-59 yrs)
  - 4 women with no reported family history + intellectual disability indicated

# Numbers of full mutations

Year	N	Males	Females	Prenatal dx
2002	12	55	7	0
2003	17	11	6	0
2004	15	8	7	1
2005	9	4	5	2
2006	9	7	2	0
2007	12	8	4	0
2008	11	4	7	0
2009	4	3	1	0
2010	4	4	0	0
TOTAL	93	54	39	3

# Children only

Year	Boys (all)	Boys (no f/h)	Girls (all)	Girls (no f/h)
2002	3	3	4	1
2003	6	5	2	0
2004	7	5	2	0
2005	4	2	0	0
2006	6	4	1	0
2007	5	3	0	0
2008	3	2	1	0
2009	1	1	1	0
2010	4	3	0	0
TOTAL	39	28	11	1

# Average age of new diagnoses in children

- In probands (no family history reported):
- N=28 boys
- Age range 9 months-16 yrs
- Mode 2 yrs (n=10)
- Median 3 yrs
- Mean age at diagnosis: 4 years

# Prevalence of fragile X syndrome

Boys <18 yrs n=39.

9 years of testing:

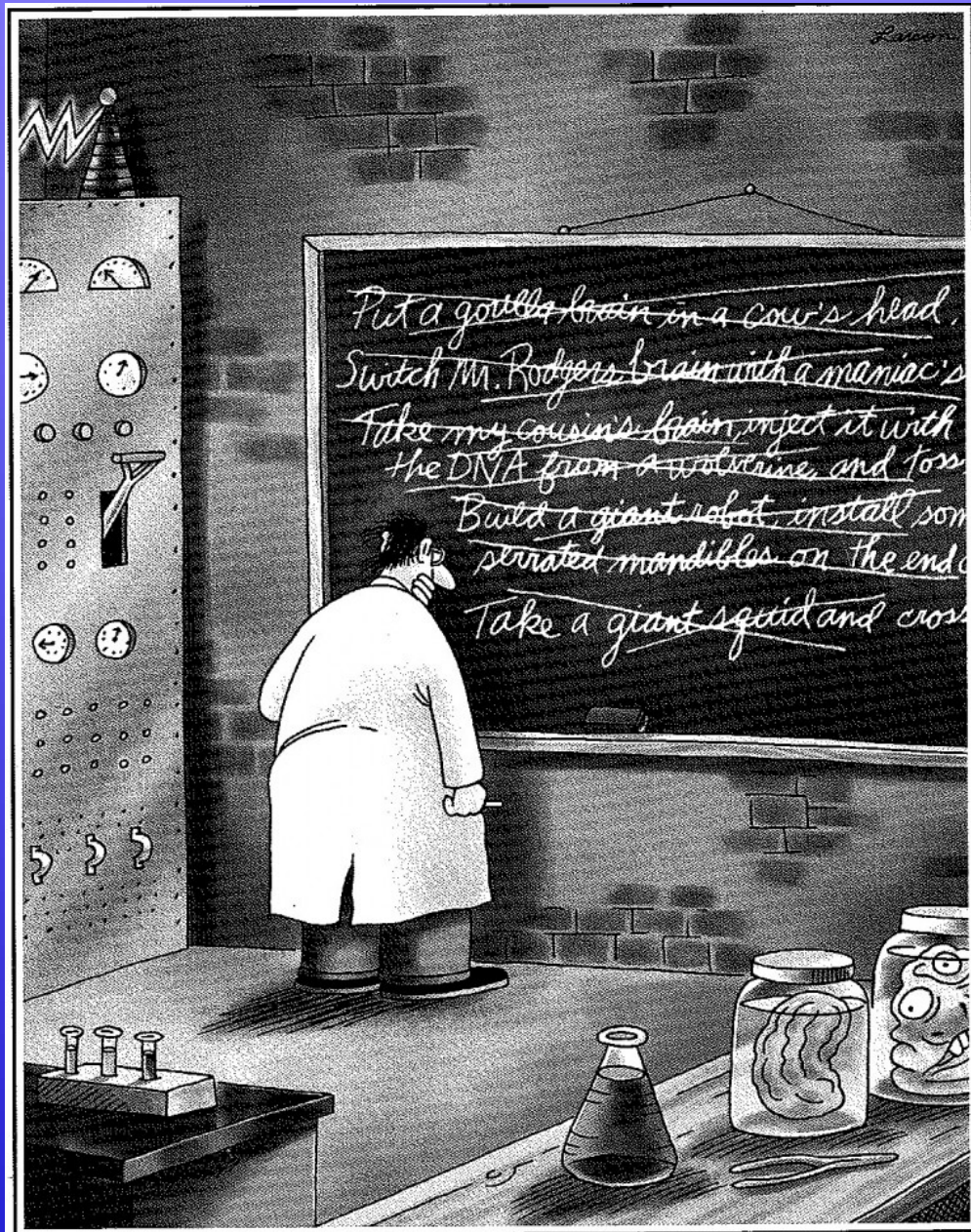
- In 9 years *preceding* testing period, total births in B.C.: 396,817



Approx 50% of births are males: 198,409

$39/198,409 = \underline{\mathbf{1 \text{ in } 5000}}$

- Same prevalence as in Atlanta (Coffee et al., 2009)
- Higher than seen in Nova Scotia (Beresford et al., 2000)



The curse of mad scientist's block



# Canadian Pediatric Surveillance Project (CPSP)

- Canada-wide federally funded protocol to obtain data on various childhood conditions
- Each condition on the list is the subject of a specific study
- Pediatricians across the country provide monthly reports on conditions on the list
- Anonymized data collected from questionnaires sent to reporting pediatricians.

# CPSP FXS Study

Funded by the Public Health  
Agency of Canada

May 2012 through April 2014

Primary objective:

To ascertain the prevalence of  
Fragile X Syndrome in Canada



# Study Objectives

## Secondary:

- To describe the demographics, regional and ethnic variations of FXS in Canada
- To describe the clinical features, age at diagnosis and co-occurring conditions (eg autistic spectrum disorder) of FXS in Canada
- To determine the access and availability of medical services for FXS in Canada
- To compare Canadian data with other published prevalence estimates

# Study Objectives

## Secondary

- To improve professional awareness of fragile X syndrome and encourage universal implementation of professional guidelines recommending diagnostic testing in children presenting with developmental delay.
- To assess the potential impact of population screening for FXS.

# The Future

- Clinical trial(s)-near future (1 yr)
- Provincial health authority responsibility for a province-wide Fragile X clinic-nearish future (2 yrs)
- Population screening-waaayyy in the future?(prob ten years)

# Overcoming barriers to care

Education and awareness

Travel funding for families

Outreach clinics

Email and telephone consultations

Telehealth! Telehealth! Telehealth!







# Acknowledgements

Dr. Tanya Nelson, PhD, Molecular Geneticist

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Jocelyn Carter, MSc, Genetic Counsellor

&

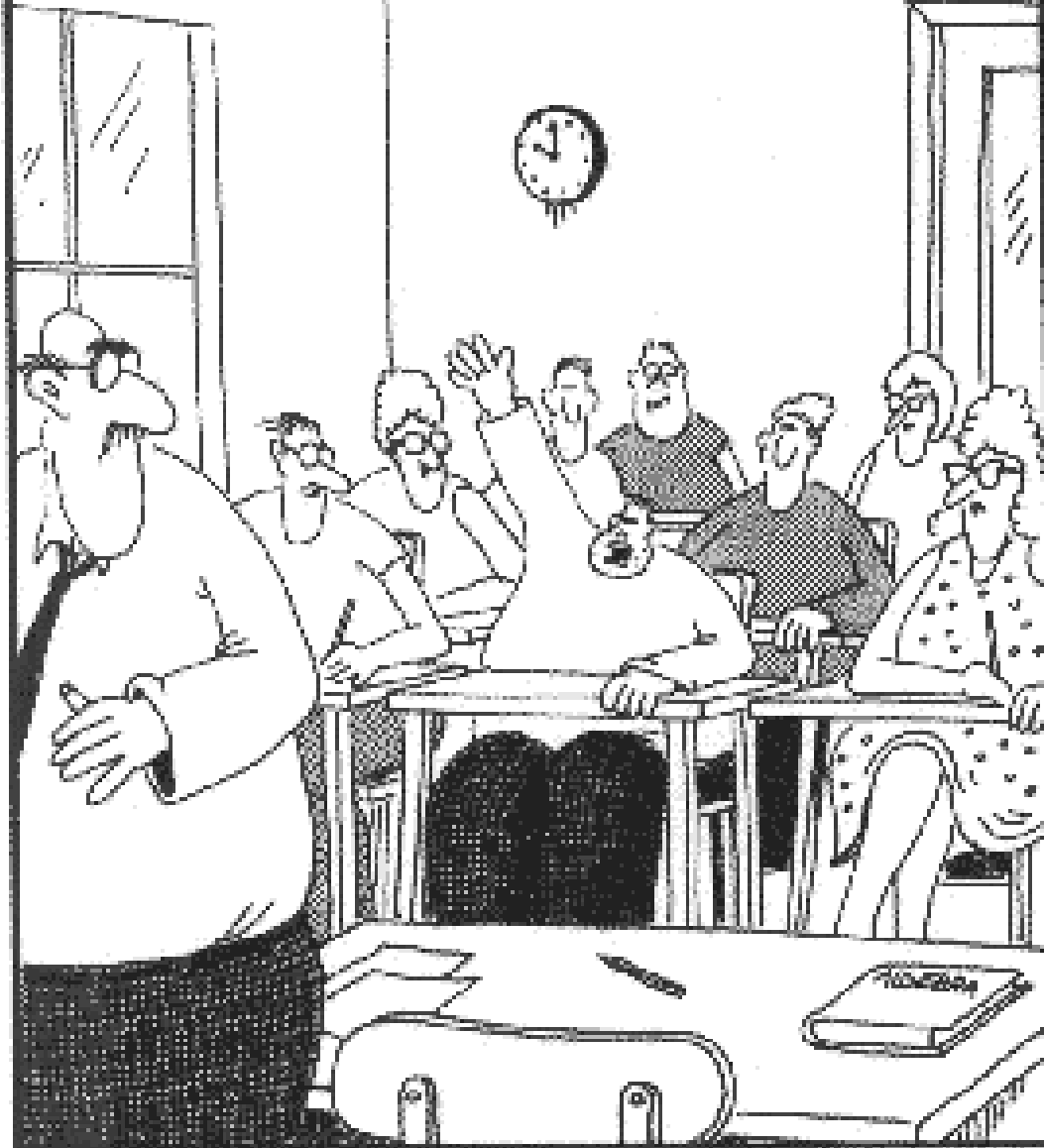
my esteemed mentors:

Dr. Jeannie Visootsak

Dr. Stephanie Sherman

1986

Looney



"Mr. Osborne, may I be excused? My brain is full."

# Top Ten Things About Canada

10. Our national bird is tastier than yours! Actually the Canada Goose is neither our National bird nor edible nor legal to kill unfortunately.
9. Our \$1 bills were turned into gold (coloured) coins!
8. Our national flag is a leaf and two bars — things you can find in any Canadian town.
7. Nearly 2% of the population are registered hockey players, ten times the rate in most other hockey playing countries!
6. Our annual military budget is less than the cost of Kim Kardashian's wedding.
5. You can get poutine, pierogies and sushi at the food courts!
4. We make good comedians: Martin Short, Jim Carrey, Mike Myers, to name a few! (The cold keeps the jokes fresh?)
3. We have a Queen, which is kind of like when your parents go away and leave your Grandma in charge.
2. We popularized a ski hat (the 'toque') as a fashion statement.
1. Ketchup-flavoured potato chips!