

# Overview of Newborn Screening Practices in Canada

Dr. P. Chakraborty, MD, FRCPC, FCCMG  
Metabolic Genetics

*Children's Hospital of Eastern Ontario, Department of Genetics  
University of Ottawa, Departments of Pediatrics and  
Pathology/Laboratory Medicine.*

## References

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## The Canadian Context

- Federation of 10 provinces and 3 federal territories
- Constitutionally defined division of jurisdiction
- Health Care: provincial jurisdiction
- Newborn screening programs generally administered by Public Health departments
- Federal government: no role to date

## History

- Guthrie test for PKU
  - 1963: Prince Edward Island
  - 1963-1970: Remaining provinces
- Congenital hypothyroidism
  - Mid 1970's: All provinces
- Subsequent divergence between provinces

# The Provinces

- Differences
  - Cultural
  - Ethnic mix
  - Language mix
  - Prenatal / carrier screening programs
- Similarities
  - Legal and political culture
  - Single-payer, government run health insurance system

# The Provinces - Screening

Births and birth rate, by province and territory  
(Number of births)

[2000-2001](#)   [2001-2002](#)   [2002-2003](#)   [2003-2004<sup>f</sup>](#)   [2004-2005<sup>p</sup>](#)

|                           | number of births |         |         |         |         |
|---------------------------|------------------|---------|---------|---------|---------|
| Canada                    | 327,107          | 328,155 | 330,523 | 335,701 | 337,856 |
| Newfoundland and Labrador | 4,732            | 4,636   | 4,596   | 4,595   | 4,511   |
| Prince Edward Island      | 1,381            | 1,313   | 1,374   | 1,416   | 1,409   |
| Nova Scotia               | 8,922            | 8,693   | 8,635   | 8,628   | 8,580   |
| New Brunswick             | 7,202            | 6,971   | 7,104   | 7,086   | 7,023   |
| Quebec                    | 71,825           | 72,602  | 72,273  | 74,378  | 75,303  |
| Ontario                   | 127,741          | 128,947 | 129,256 | 131,121 | 131,454 |
| Manitoba                  | 13,939           | 13,746  | 13,765  | 13,985  | 14,111  |
| Saskatchewan              | 12,084           | 11,996  | 11,794  | 12,063  | 12,144  |
| Alberta                   | 37,197           | 37,602  | 39,450  | 40,520  | 41,015  |
| British Columbia          | 40,367           | 39,932  | 40,534  | 40,099  | 40,465  |
| Yukon Territory           | 348              | 344     | 322     | 339     | 345     |
| Northwest Territories     | 656              | 651     | 658     | 706     | 711     |
| Nunavut                   | 713              | 722     | 762     | 765     | 785     |

<http://www40.statcan.ca/l01/cst01/demo04a.htm>

## The Provinces - Screening

- Number of diseases screened
  - Reasons for screening
- Technologies used
- Legal framework and consent
- Governance and the use of advisory committees
- Treatment and follow-up practices.

Number of diseases screened

**TABLE 2  
Newborn screening: Canada**

| Province/Territory        | Diseases   |
|---------------------------|--|
| British Columbia          | PKU, CH, galactosemia, MCADD, hearing deficits*  |
| Alberta                   | PKU, CH, biotinidase, hearing deficits*  |
| Yukon                     | Covered by British Columbia  |
| Northwest Territories     | Western area covered by Alberta, eastern area covered by Manitoba  |
| Nunavut                   | Western area covered by Manitoba, eastern area covered by Quebec   |
| Saskatchewan              | PKU, CH, tandem mass spectrometry (27 diseases), hearing deficits*   |
| Manitoba†                 | PKU, CH, congenital adrenal hyperplasia, galactosemia, galactose-1-phosphate uridyl transferase, biotinidase, Duchenne muscular dystrophy (males), hearing deficits* |
| Ontario                   | PKU, CH, hearing deficits*   |
| Quebec                    | PKU, CH, tyrosinemia, hearing deficits*, urine screen‡   |
| New Brunswick             | PKU, CH, hearing deficits*   |
| Prince Edward Island      | PKU, CH, MCADD   |
| Nova Scotia               | PKU, CH, MCADD (soon to add seven other diseases to the panel), hearing deficits*  |
| Newfoundland and Labrador | PKU, CH, homocystinuria (one region), tyrosinemia  |

*\*Not yet universal (5% to 95%) and methods vary; †Manitoba also has targeted screening. The Oji-Crees are screened for glutaric acidemia type 1 and the Hutterites are screened for carnitine palmitoyl transferase 1 deficiency; ‡Unique program of urine-impregnated filter paper testing at 21 days of age. (see text) for amino acids and organic acids (4). CH Congenital hypothyroid; PKU Phenylketonuria; MCADD Medium-chain acyl-CoA dehydrogenase deficiency*

|                  | PKU | CH | CAH | Gal | Tyr | Biot | MCAD | HB | Hear | MS/MS |
|------------------|-----|----|-----|-----|-----|------|------|----|------|-------|
| British Columbia | ✓   | ✓  |     | ✓   |     |      | ✓    |    | ✓    | *     |
| Alberta          | ✓   | ✓  |     |     |     | ✓    |      |    | ✓    | *     |
| Saskatchewan     | ✓   | ✓  |     |     |     |      | ✓    |    | ✓    | ✓     |
| Manitoba         | ✓   | ✓  | ✓   | ✓   |     | ✓    |      |    | ✓    | *     |
| Ontario          | ✓   | ✓  | ✓   | ✓   | ✓   | ✓    | ✓    | ✓  | ✓    | *     |
| Quebec           | ✓   | ✓  |     |     | ✓   |      |      |    | ✓    |       |
| New Brunswick    | ✓   | ✓  |     |     |     |      |      |    | ✓    |       |
| P.E.I.           | ✓   | ✓  |     |     |     |      | ✓    |    | ✓    | *     |
| Nova Scotia      | ✓   | ✓  |     |     |     |      | ✓    |    | ✓    | *     |
| Newfoundland     | ✓   | ✓  |     |     | ✓   |      |      |    |      |       |

## Additional tests

- Manitoba:
  - CPT1 deficiency (Hutterites)
  - Glutaric Aciduria Type 1 (Oji-Cree)
  - Duchenne Muscular Dystrophy
- Quebec
  - Urine screening for amino and organic acid diseases (1971)

## Reasons for screening

- **Intervention**
- Reproductive counselling
- Enumeration and surveillance

# Technologies

## MS/MS technology

- Currently in use
  - British Columbia
  - Saskatchewan
  - Nova Scotia (PEI)
- In deployment
  - Alberta
  - Manitoba
- Announced
  - Ontario

## MS/MS technology

- Allows rapid, simultaneous measurement of a large number of metabolites
  - Acylcarnitines – organic acid / fatty acid diseases
  - Amino acids – amino acid diseases
- Not all metabolites must be measured
  - Can selectively screen for certain diseases
- But, some metabolites can give information about several diseases
  - May be difficult to screen for a single chosen disease

## MS/MS technology

- Currently in use
  - British Columbia                      MCAD, PKU
  - Saskatchewan                              Acylcarnitines / Amino acids
  - Nova Scotia (PEI)                          MCAD, PKU
- In deployment
  - Alberta    PKU + ?
  - Manitoba                                        ?
- Announced
  - Ontario    ACMG core condition panel



## Choice of technologies

- Congenital hypothyroidism
  - TSH vs. T4
  - Primary vs. secondary
- Galactosemia
  - Enzyme activity vs. Galactose / Gal-1-P
  - Sample handling vs. feeding status
- PKU
  - MS/MS vs. Guthrie / spectrophotometric
  - Phenylalanine and tyrosine vs. phenylalanine

## Turnaround time

- Required turnaround time for expanded screening
  - Time between detection and onset of disease
- Nova Scotia – weekly run
- Ontario – 6 day work week for <72 h TAT
- Number of samples received
  - NS: 30-35 / day
  - Ontario: 400-600 /day

## Other technologies

- Manitoba targeted screening
  - Mutation analysis
- Quebec urine screening / Alberta amino acids
  - Thin layer chromatography
- Quebec tyrosinemia
  - Succinylacetone measured if tyrosine elevated

## Legal framework and Consent

## Legal Mandate

- Two provinces mandate newborn screening:
  - Saskatchewan - *Hospital Standards Regulations, 1980*
    - phenylketonuria and congenital hypothyroidism
  - Quebec - *Health and Social Services Act, R.S.Q. c. S-4.2*
    - “[a] public or private institution under agreement that operates a hospital centre shall make a by-law respecting the fixing of screening examinations required at the time certain users are admitted or registered, in accordance with the standards made under paragraph a of section 15 of the *Medical Act* (R.S.Q., c. M-9).”
- Need for legal mandate in single payer systems?

## Consent

- Not clearly delineated in any Canadian jurisdiction
- Written consent not required
- Informed dissent
  - may or may not require specific reason
  - E.g. religious, personal belief
- No province has consent process for secondary use of blood dots at time of sampling

## Consent

- Saskatchewan
  - Mandated by law, but parents can dissent by signing waiver.
- Quebec
  - Consent included in mother's hospital admission consent. Dissent allowed.
- New Brunswick
  - Considering consent form.
- Ontario
  - Not excluded from Consent to Treatment Act, but no written consent form required. Dissent allowed.

## Governance and Advisory Committees

## Governance

- Programs in all provinces are governed by Public Health department of Ministries of Health / Social Services
- Ontario program is being moved from Public Health
- The newborn screening facilities are housed in a variety of settings
  - Hospital labs
  - Public Health labs

## Advisory Committees

- All provinces have an advisory committee except Quebec
- 5 / 9 committees have official mandate
- Composition of committees variable
  - Public health
  - Metabolic geneticists
  - Ministry officials

## Other Decision Making Processes

- Professional organizations
  - Garrod Association
  - Canadian Pediatric Society
- Parent organizations
  - Save Babies Through Screening Canada
- Arms-length agencies
  - CCOHTA
- Government
  - Medical Advisory Secretariat (Ontario)

## Treatment and Follow-up

## Follow-up

- All provinces except Ontario have centralized computer system for documenting follow-up and quality assurance
- Follow-up and treatment may involve interprovincial cooperation
  - PEI Nova Scotia
  - Saskatchewan Manitoba
- British Columbia
  - Screening lab in the central diagnosis and treatment hospital
- Ontario
  - Screening lab being moved to CHEO
  - Five regional diagnosis and treatment centres
- Quebec
  - Two screening labs (CHUL and CHUS)
  - Four regional diagnosis and treatment centres

## Treatment

- Provincial health insurance programs cover access to specialist care
- Treatment costs are variably and often only partially covered
  - synthetic formula > medication > medical food
- Most provinces use the National Food Distribution Centre (NFDC) in Montreal
- Treatment for adults is covered in six provinces
  - Ontario: coverage for women with PKU contemplating pregnancy

## Summary

- Practices and policies in Canada are quite varied from province to province
- Often lagging behind those of other industrialized nations
- Need for institution of processes to:
  - ensure up to date screening panels and practices
  - clarification of the legal status of screening practices and consent requirements
  - modernization of policies for education, treatment and follow-up

## Future considerations

- Confederal (e.g. CCOHTA) or Federal (e.g. PHAC) process to set guidelines
  - Along model of HRSA / ACMG panel
  - Precedence: national vaccination strategy
- Consider broadening criteria for screening
- Increased regional cooperation
  - Especially for provinces with smaller population to improve turnaround time and access to specialist care.