



# Population-based screening in the maternal- child health context: evidence-informing policy

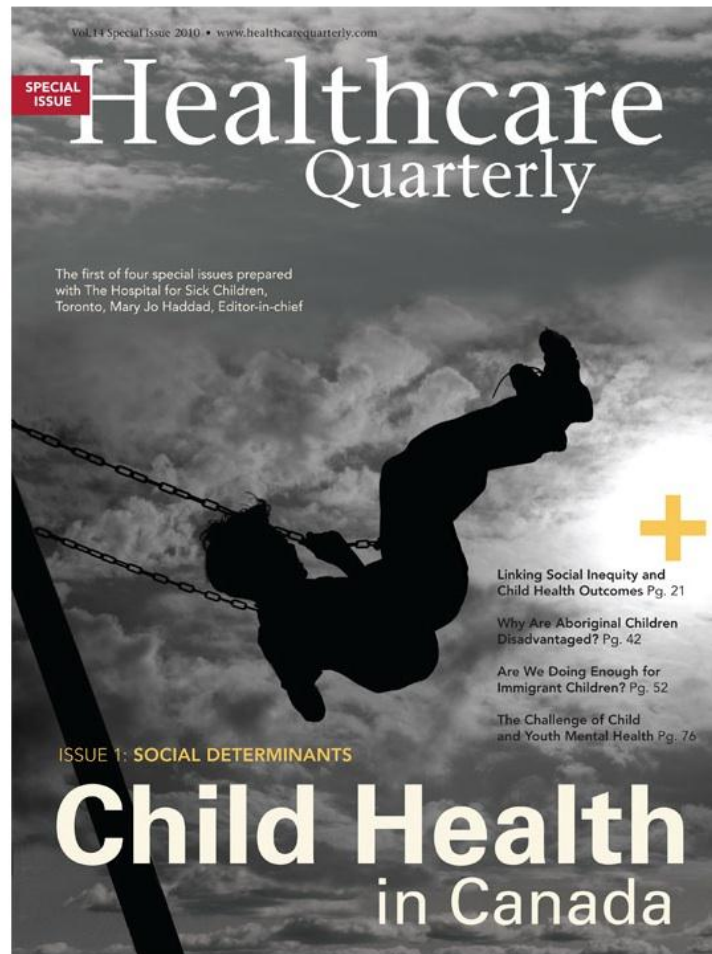
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January 24, 2012

# Outline

1. Population-based screening, a complex intervention in the maternal-child health context
2. Recent policy developments in ON specific to this context
3. Data from 3 newborn screening projects, highlighting some of the complexities
4. Future research directions

# Why focus on children?



# Why focus on children?

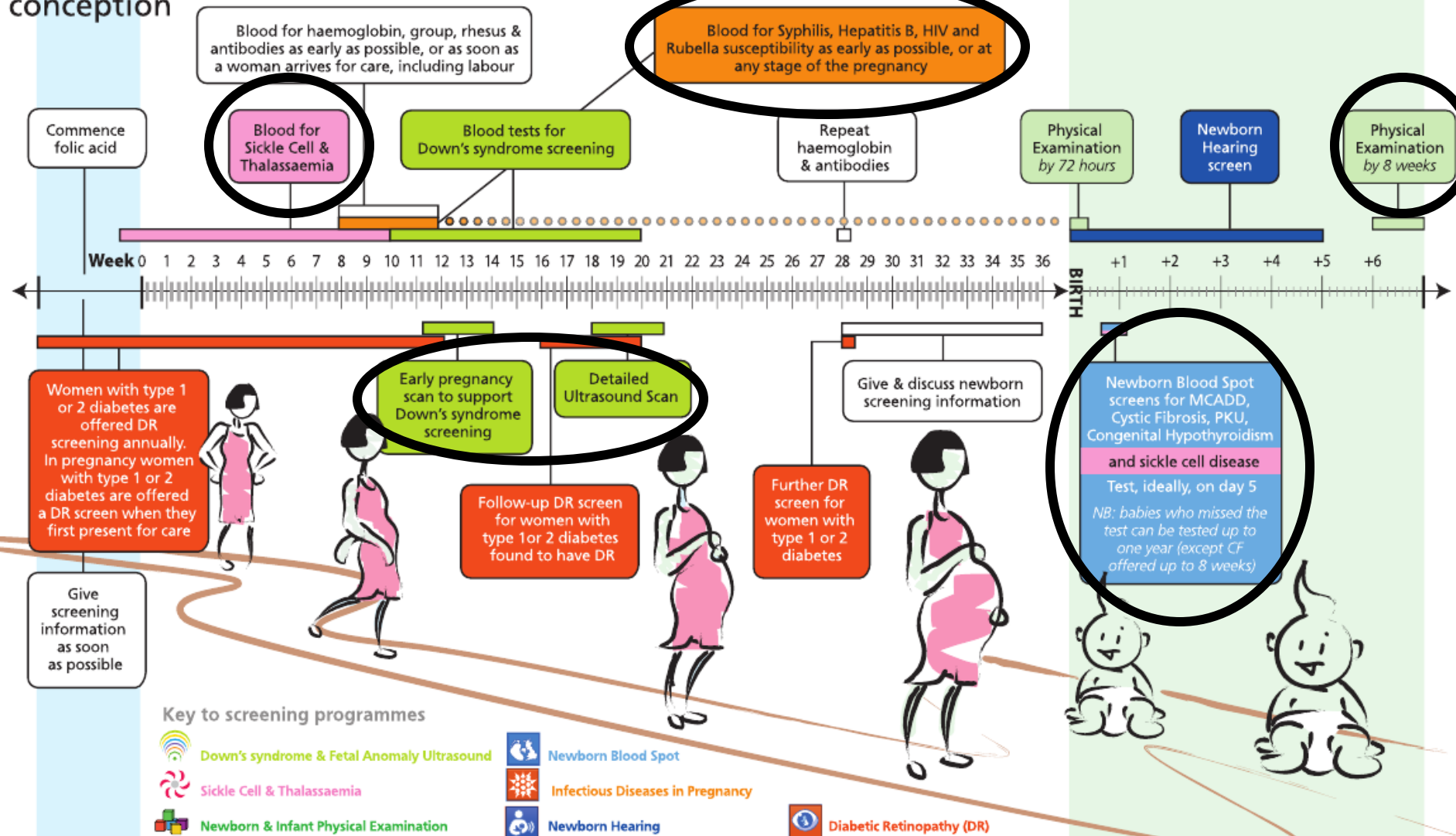
- Prenatal and early childhood influences are antecedents to adult health
- Early intervention in childhood to reduce risk factors, disease morbidity is key to longer term health
- Early intervention requires **high quality early detection strategies**
- Knowledge gaps regarding effective delivery, evaluation, and governance of early detection strategies for children

Women and their families should understand the purpose of all tests before they are taken

## Pre-conception

## Antenatal

## Newborn



# Antenatal and Newborn Screening Timeline – optimum times for testing

## SCREENING

The CCI Conference on Preventive Aspects of Chronic Disease, held in 1951, defined screening as "the presumptive identification of unrecognized disease or defect by the application of tests, examinations, or other procedures which can be applied rapidly. Screening tests sort out apparently well persons who probably have a disease from those who probably do not. A screening test is not intended to be diagnostic. Persons with positive or suspicious findings must be referred to their physicians for diagnosis and necessary treatment."<sup>1</sup> It should be noted that, by definition, unrecognized symptomatic as well as pre-symptomatic disease is included; also, physical examination is considered part of the procedure, so long as it can be classed as rapid. The term "other procedures" may also embrace the use of questionnaires, which are assuming an increasingly important place in screening. Finally, tests may be "diagnostic", though not necessarily so intended; for example, a gynaecological examination could be covered by this definition provided it were rapidly carried out. In general, we have taken the definition to imply a relatively simple (though not necessarily unsophisticated) method of case-finding.



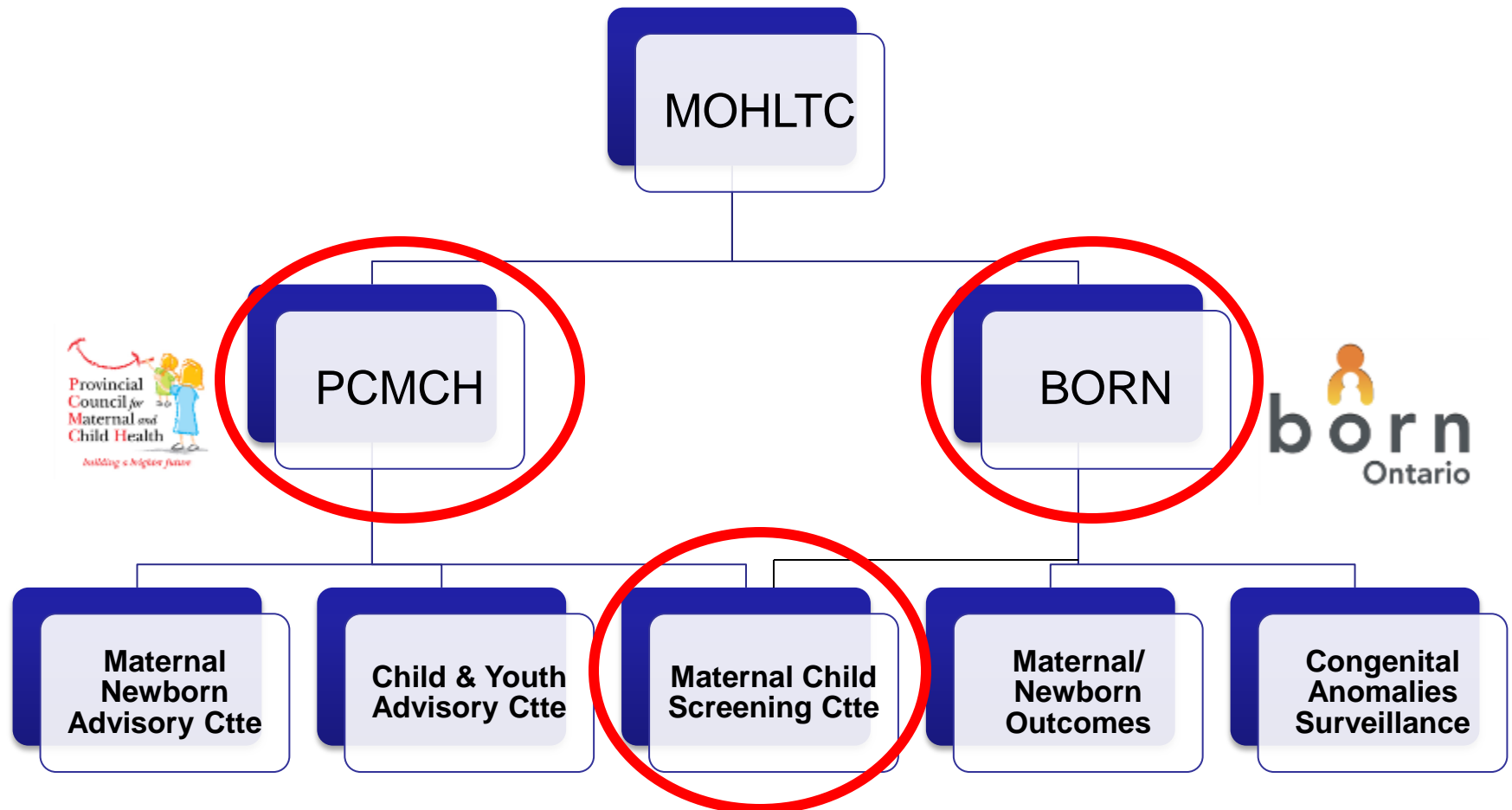
# Traditional Screening Principles

1. Is the disease an important public health problem?
2. Is there an effective treatment for localized disease?
3. Are facilities for further diagnosis and treatment available?
4. Is there an identifiable latent or early symptomatic stage of disease?
5. Is the technique to be used for screening effective?
6. Are the tests acceptable to the population?
7. Is the natural history of the disease known?
8. Is there a strategy for determining which patients should and should not be treated?
9. Is the cost of screening acceptable?
10. Is effective treatment available and does management of cases in the early stages have a favorable impact on prognosis?

# Emerging complexities

- Increasingly sophisticated technologies
  - interpretive challenges
- Broadening scope of 'targets'
  - mild manifestations or risk states
- Health & non-health outcomes
  - index case & family
- Preference-sensitive decision making
- Access challenges
  - Screening itself and implicated services

# Ontario's Approach

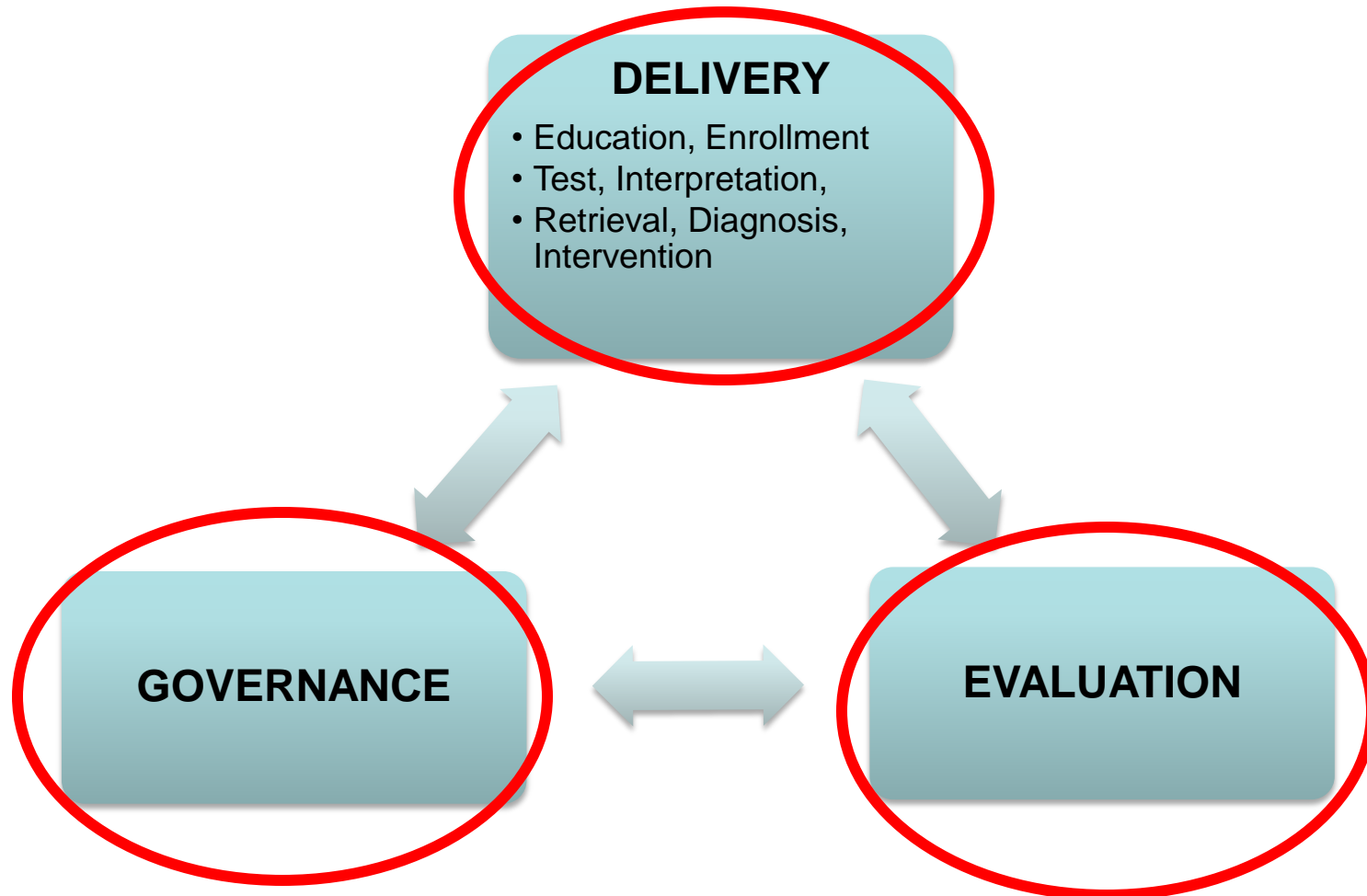


# MCSC Definition of Screening

- **Screening** is the *systematic, population-based application of a test or inquiry to individuals who do not have symptoms of a specific disease or condition in order to identify those who warrant further investigation and/or intervention to achieve better outcomes.*

*Approved: Dec 2011*

# Screening as a System of Care





# Expansion of Newborn Screening

## HISTORICALLY:

- Treatable disorders, prior to onset of irreversible symptoms
  - PKU paradigm
- Scope of screening limited by technology
- 'Public health emergency' model

## NOW:

- Technology enables rapid screening of many conditions concurrently
- Much enthusiasm re: opportunities to intervene early
- But evidence re: potential benefits/harms still emerging
- 'Public health emergency' model persists...

# In Ontario

- NBS expanded in 2006
- Now includes 29 disorders
- Centralized in Ottawa
- Screen positives retrieved by NSO and referred to 1 of 5 treatment centres



# Along with expansion has come

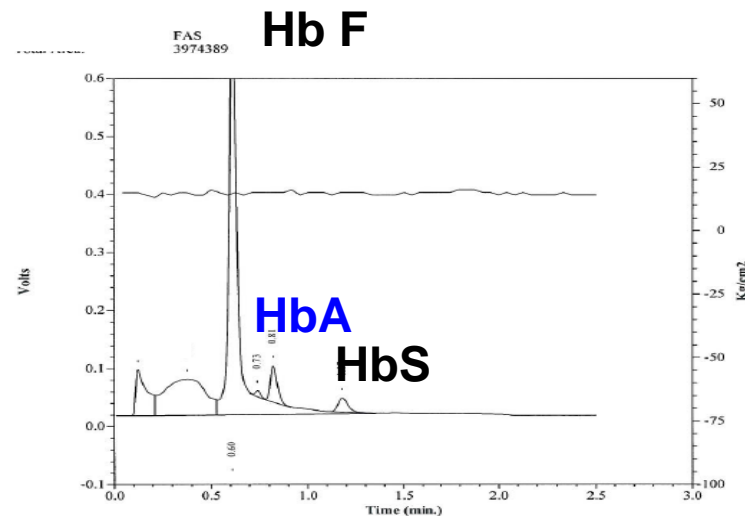
- Larger numbers of affected infants requiring specialist treatment
- Wider range of other screen positive infants, including carriers and infants with uncertain dx
- Broader definition benefit, including information for families to guide life and reproductive planning
- Growing impact on health services, new roles for primary care and specialist providers

**Evidence base to support the effective delivery of expanded NBS – as a system of care - is limited**

# The case of sickle cell disorder

# Newborn screening for SCD

- SCD
- Evidence re: prophylactic antibiotics
- 7350 screen positives, all conditions (NSO: 2006-2011)
  - 441 screen positive for hemoglobinopathies
- 8650 SCD carriers



## What questions did this raise?

- How should we manage screening results that are incidental to the primary goal?
- Especially if they are *typically* considered benign
- How should we reconcile this with long-standing policy that opposes genetic carrier testing in minors?
- What approach is most sensitive to the racialized nature of this disease?

# Ontario's response to this dilemma

- Advisory Ctte on Newborn & Childhood Screening
- Jurisdictional Scan
- Commissioned Research
  - Funded by MOHLTC
  - Multi-disciplinary team
  - Disclosure policy on hold until evidence available

# Research Question

How should sickle cell carrier results, generated incidentally through newborn screening, be managed in Ontario?

# Design/Methods

## Cross-sectional Survey

Health Care Providers

- OB, FP, MW, NUR, PED, GEN, HEM

## Qualitative Interviews

Health Care Providers

- OB, FP, MW, NUR, PED, GEN, HEM

Advocates

Parents

## Focus Groups

New Parents

Parents exposed to SCD

# Key Survey Constructs

- Reasons favouring/opposing carrier disclosure
- General beliefs about NBS, including consent
- Practices & attitudes regarding education pre- and post-NBS
- Perceived barriers re: NBS and SCD care
- Demographics

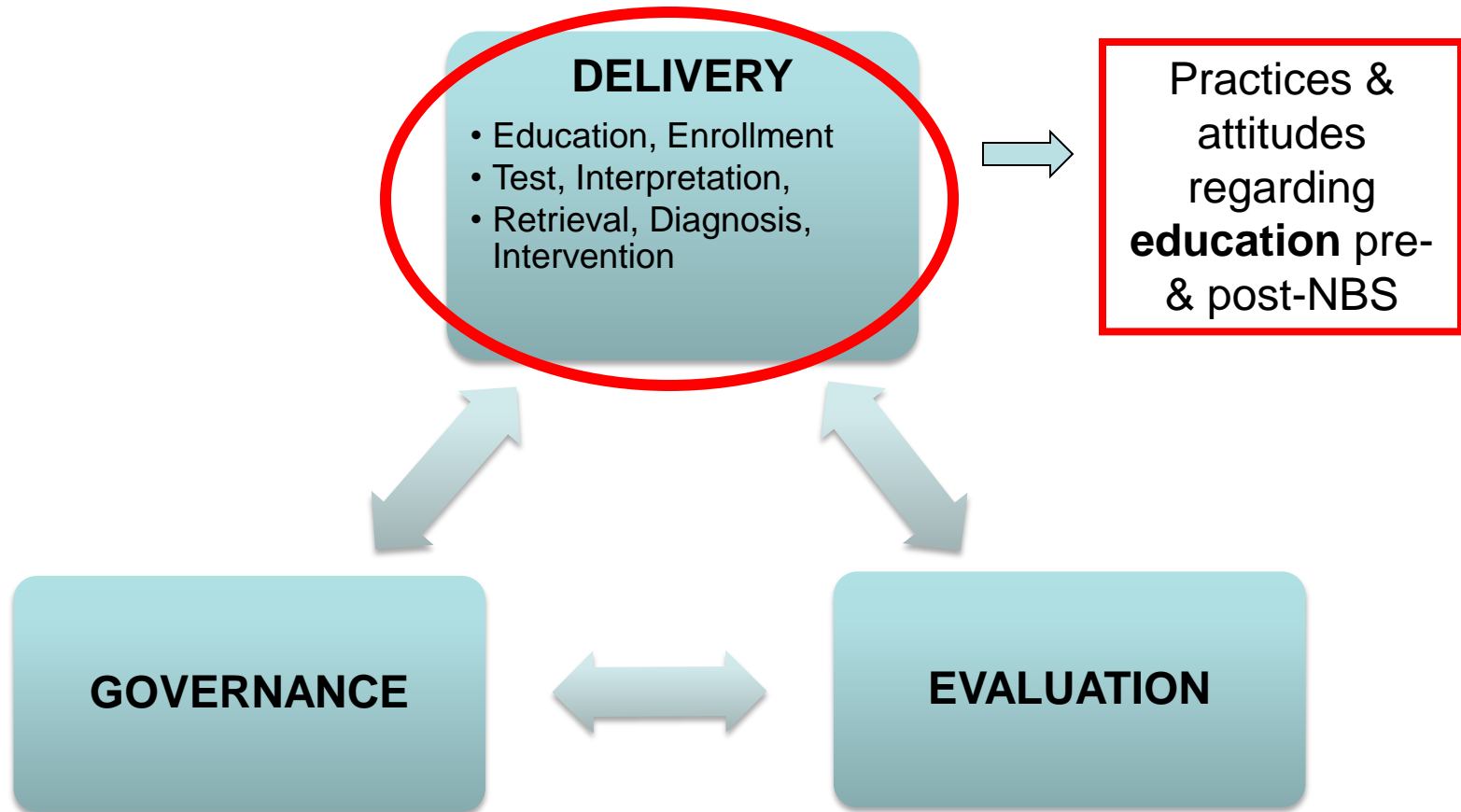
# Survey Response Rate

Provider Group	Completed (%)
Obstetricians (OB)	212 (54.1)
Midwives (MW)	250 (76.7)
Nurses (NUR)	489 (72.6)
Family Physicians (FP)	296 (50.6)
Pediatricians (PED)	273 (63.0)
Genetics Professionals (GEN)	53 (68.0)
Hematologists (HEM)	42 (51.8)
<b>TOTAL</b>	<b>1615 (62.9)</b>

# Survey Demographics

Demographic	Total N (%)
<b>Practice Setting</b>	
Academic	404 (25.3)
Non-academic	1196 (74.7)
<b>Geographic Location</b>	
Metro/Central City	674 (41.7)
Other	941 (58.3)
<b>Years in Practice</b>	
0-10	688 (45.3)
11+	883 (54.7)
<b>Gender</b>	
Female	1171 (73.1%)
Male	430 (26.9%)

# Screening as a System of Care



## The majority of physicians rarely/never educate parents pre-NBS.

Educate pre-NBS	OB (%) N=167	MW (%) N=244	FP (%) N=195	PED (%) N=155	NUR (%) N=456	X <sup>2</sup>
Consistently/ Usually	15.6	98.4	16.9	9.0	46.5	<.0001
Sometimes	18.0	0.8	18.0	25.8	25.4	<.0001
Rarely/Never	66.5	0.8	65.1	65.2	28.1	<.0001

**<1/2 of OBs perceive this to be their responsibility. FPs do, but face barriers.**

Belief/Barrier	OB (%) N=167	MW (%) N=244	FP (%) N=195	PED (%) N=155	NUR (%) N=456	X <sup>2</sup>
Responsibility to inform	41.3	97.2	72.3	56.3	78.7	<.0001
Insufficient time	65.2	17.3	65.6	58.6	37.9	<.0001
Insufficient compensation	72.6	25.1	58.3	48.6	18.6	<.0001
Not confident re: NBS	51.9	19.8	65.0	14.3	39.1	<.0001

## Perceived responsibility and cognitive barriers have strongest association with education.

Variable	Adjusted OR (95% CI) – Model 1
Provider Group	
OB	1.00
FP	--
PED	0.56 (0.33–0.94)
NURS	3.08 (1.91–4.98)
Belief/Barrier	
Feel responsible	2.92 (2.08–4.08)
Lack Confidence	0.33 (0.24–0.46)
Demographic	
Non-urban	1.494 (1.100–2.028)

## PEDs are most likely to educate in detail, FPs report doing this much less.

Educate post-NBS	FP (%) N=249	PED (%) N=211	MW (%) N=250	TOTAL N=710
Brochures (C/U)	16.3	26.5	34.2	25.7
General Discussion (C/U)	29.9	56.2	48.1	44.4
Detailed Discussion (C/U)	24.8	68.6	28.5	39.1

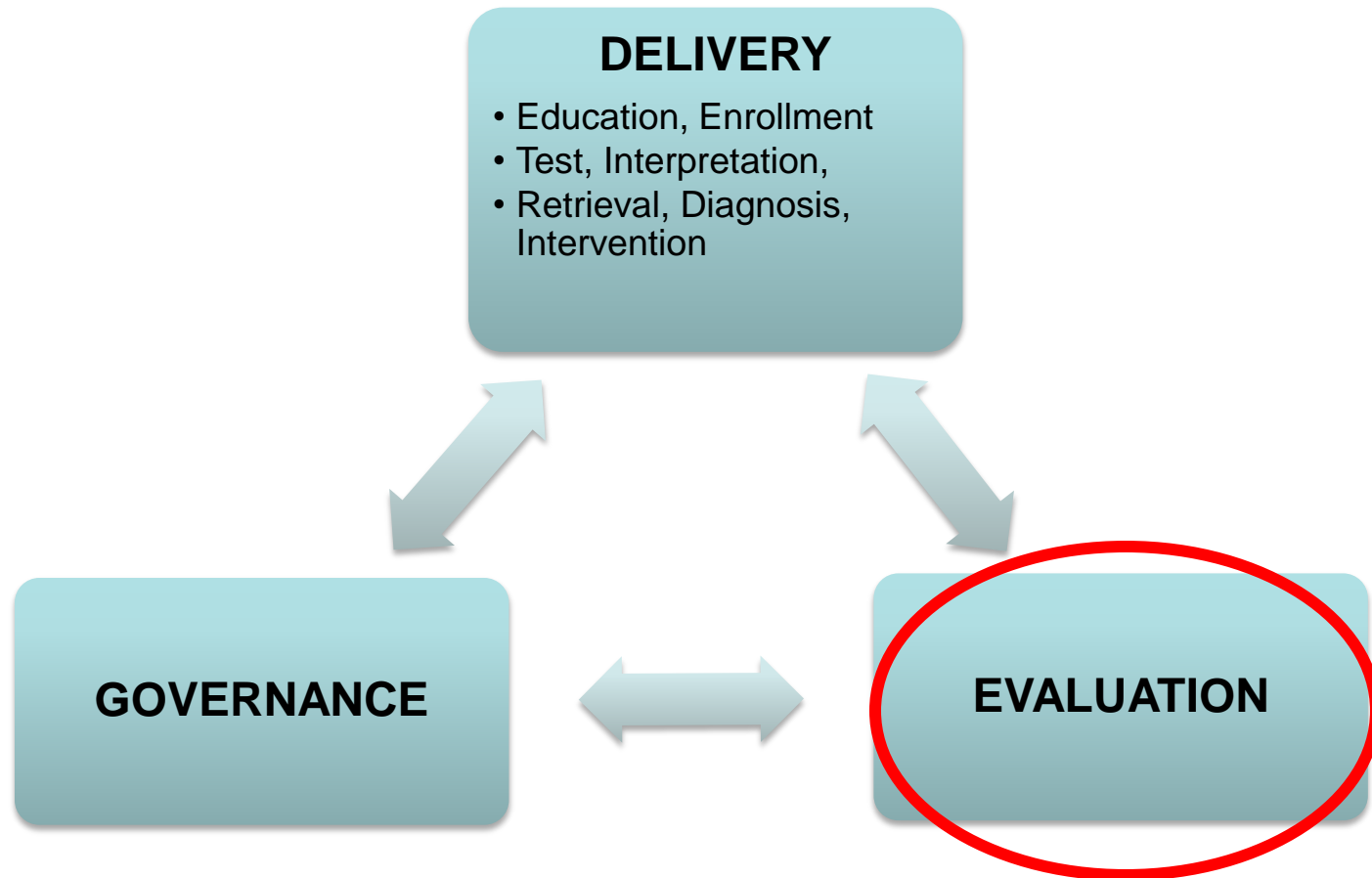
## Majorities feel responsible to educate post NBS. Practice/belief discrepancy most striking for FPs.

Belief/Barrier	FP (%)	PED (%)	MW (%)	TOTAL
Total n	249	211	250	710
Feel responsible to care	73.7	92.4	76.5	80.1
Should provide				
Brochures	66.4	74.0	77.3	72.7
General disc	75.9	86.0	85.5	82.3
Detailed disc	24.8	68.6	28.5	39.2
Insufficient				
Time	45.1	36.0	19.2	33.1
Compensation	56.0	41.2	35.6	44.4
Training	76.9	38.0	68.8	62.9

# Key Messages: Education

- There are provider-level challenges to educating parents about expanded NBS
- Targeted capacity building is warranted

# Screening as a System of Care



# The case of cystic fibrosis

# Newborn screening for CF

- CF
- Evidence re: improved nutritional status, height & weight gain
- ~400 screen positive cases/yr in ON
  - 8% uncertain diagnosis
  - 82% false positive (majority = carriers)
  - 10% true positive
- SickKids NBS CF Consultation Clinic

# Research Question

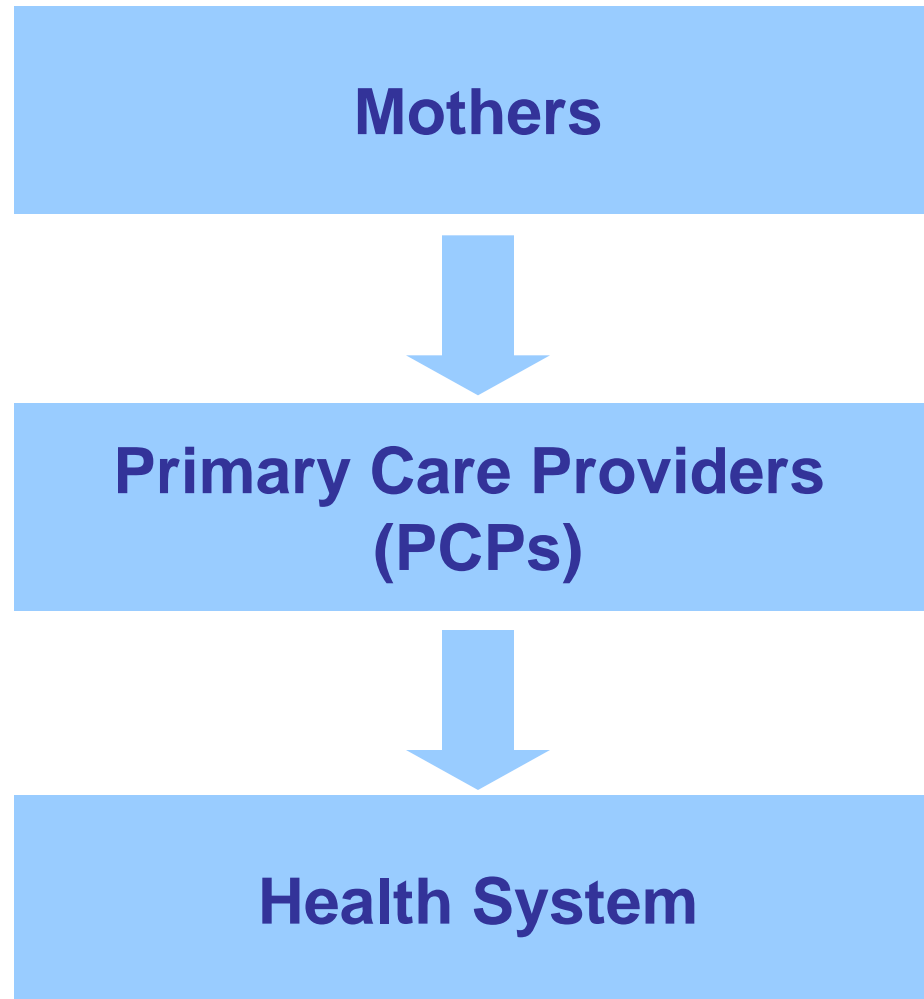
What is the impact of CF newborn screening on:

- Mothers of CF screen positive infants
- Primary care providers
- Health service utilization by CF screen positive infants and their mothers

# Anticipated Outcomes

- A longitudinal examination of family experience and health service utilization for the ***uncertain*** category of screen positive results
- A rigorous test of the hypothesis that false +ve results can lead to over-medicalization
- An understanding of maternal factors that might identify mothers at risk of negative psychological response
- A comprehensive analysis of the primary care role in expanded NBS

# Study Design



# Methods

## Cross-sectional Survey

### Mothers

- Knowledge, attitudes, psych response
- Longitudinal

### PCPs

- Barriers, facilitators re: notifying & caring for screen +ve infants

## Qualitative Interviews

### Mothers

- Meaning of results, NBS process, personal story
- Longitudinal

### PCPs

- Perceived role in NBS
- Personal experiences

## Admin Claims Data (1yr)

### Mothers

### Infants

- Utilization of primary care, emergency and inpatient hospital services



# An Early Look: Mothers



**Mothers' Perspectives  
on Newborn Screening**

# An Early Look: Mothers

- Survey response rates
  - Screen Positive: 55%
  - Screen Negative: 40%
- 8 Qualitative interviews complete

## CF knowledge is higher among mothers of false +ve infants.

	False Positive N=43		Screen Negative N=95		P value*
	Mean	SD	Mean	SD	
Score	6.95	1.63	3.32	2.75	<0.0001

\*Independent Samples t-test

# Anxiety scores are relatively low and not different between groups.

	False Positive N=41		Screen Negative N=91		P value*
	Mean	SD	Mean	SD	
Score	31.66	10.66	33.87	10.13	0.152

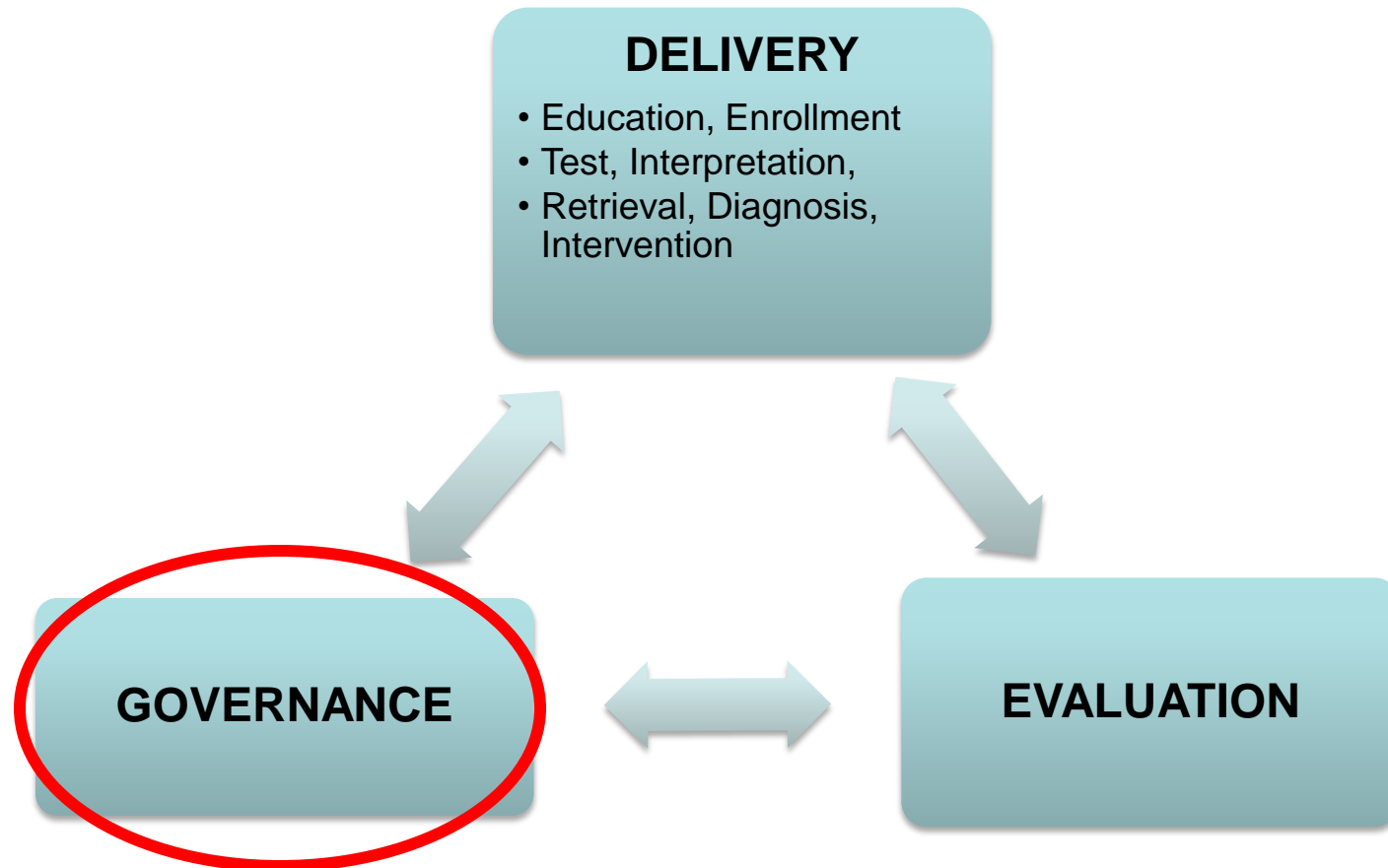
\*Independent Samples t-test

## Child vulnerability scores are low and not different between groups. Unexpected trend?

	False Positive N=42		Screen Negative N=95		P value*
	Mean	SD	Mean	SD	
Total	4.36	3.09	5.47	4.58	0.152

\*Independent Samples t-test

# Screening as a System of Care



# Public Values

# A call for public engagement

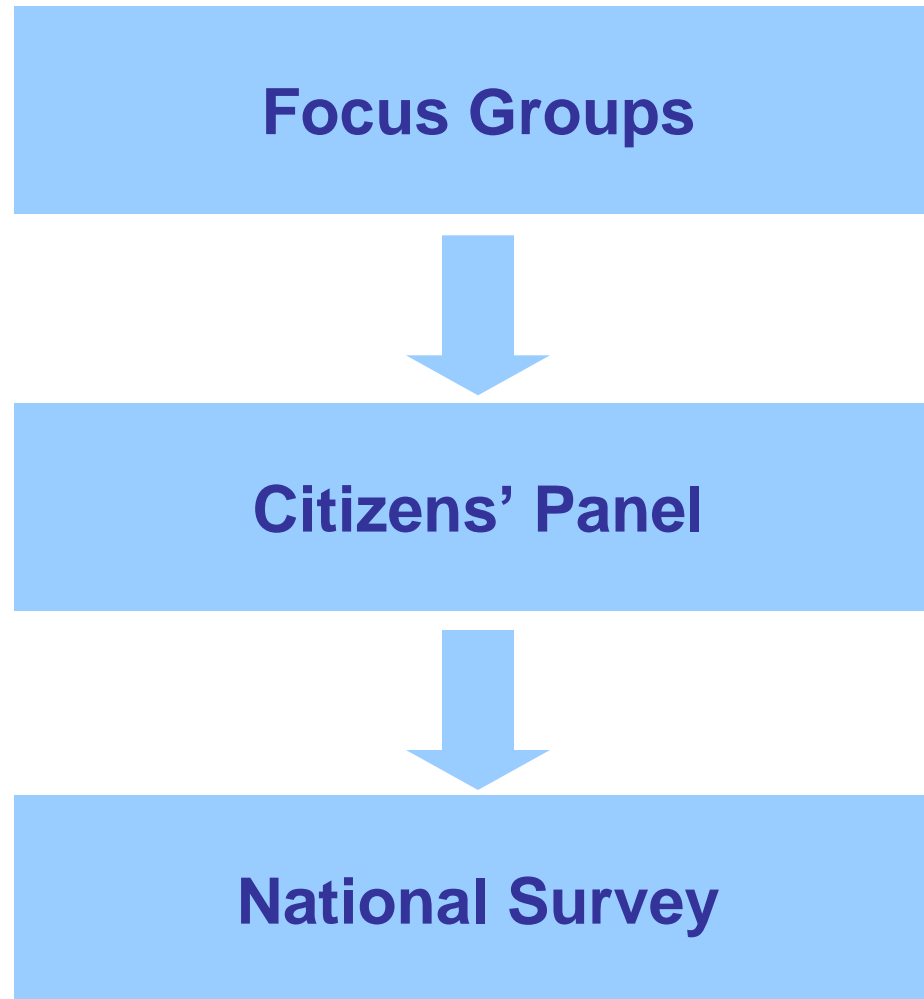
- Debate about goals of NBS & broadening notion of benefit invokes longstanding questions of consent
- Available lit reflects views of parents & providers and is unresolved
- Public values key to diffusion/uptake of new technology
- Public values largely unexplored in NBS context

# Research Questions

What are the public's values towards:

1. The types of **conditions** that should be screened (e.g., treatable or not)
2. Whether parents should provide **consent** to have their infant screened

# Study Design



# Methods

## Focus Groups

- Eight 3-hr focus groups (n=60)
- Educational components:
  - Disorders to screen for, consent

### Mixed Methods:

#### Qualitative

- Deliberative discussion
- Analyzed using principles of qualitative description

#### Quantitative

- Pre- & post-questionnaires
- Analyzed using descriptive & repeated measures statistics

# Demographics

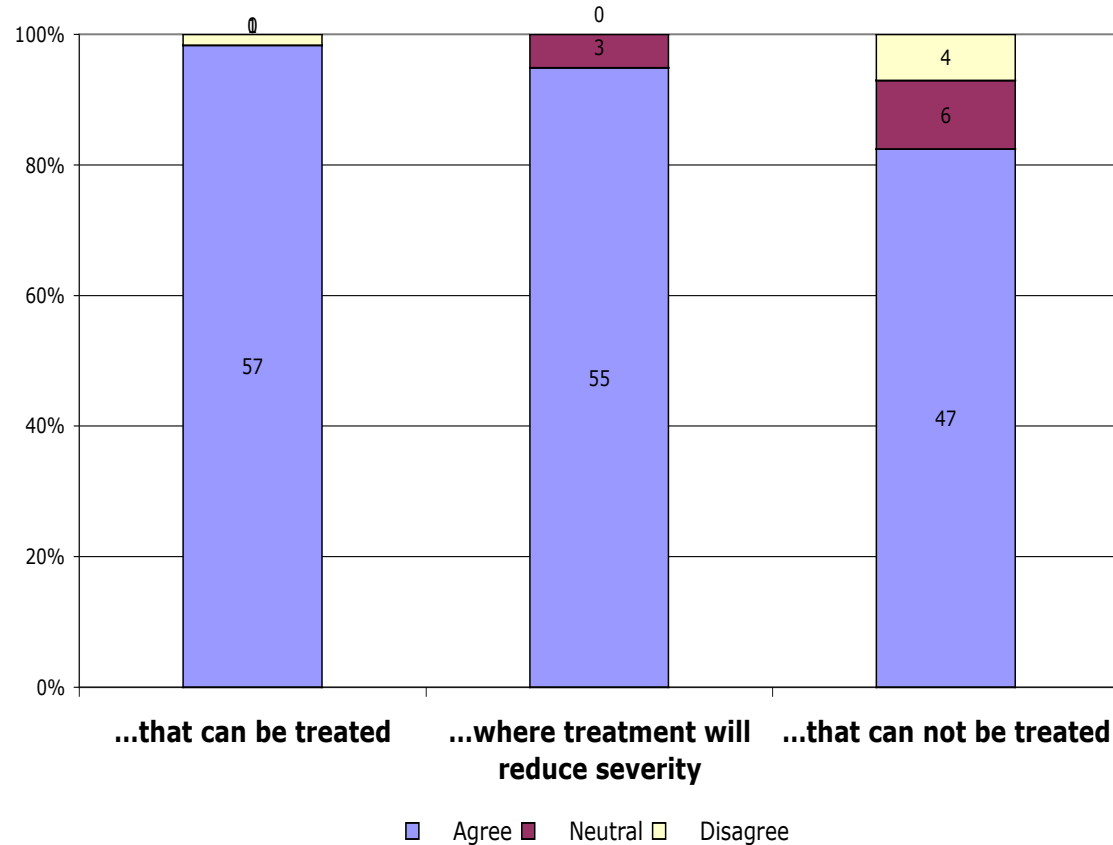
Focus Group Participants (Total N=60)		
	n	%
Female	36	60%
18-29 yrs	16	27%
30-49 yrs	26	43%
50+ yrs	18	30%
Married/common law	24	40%
Some college & above	52	87%
>1 Child	26	43%

Hayeems et al (2012) Pediatrics, under review

**What types of conditions  
should be screened?**

# Majority supports NBS for treatable & untreatable conditions

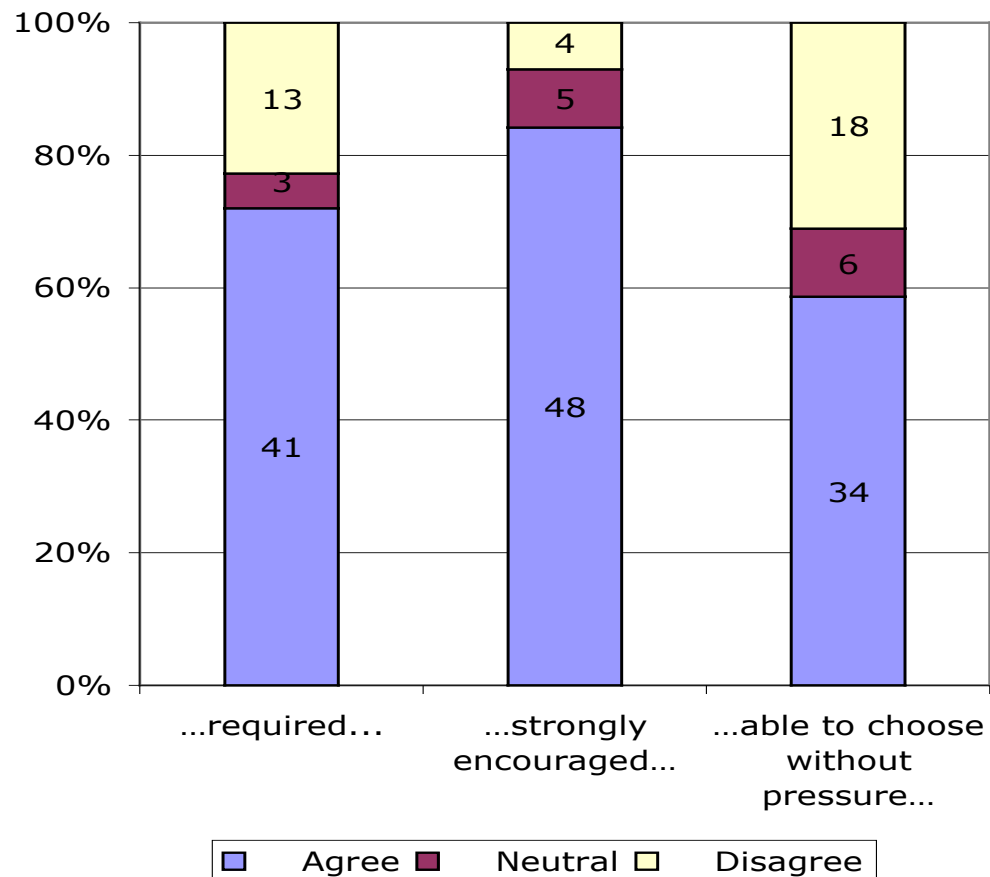
Q: "I think NBS should try to find babies with serious disorders... "



**Should parents be given more  
discretion in NBS?**

# Majority prefers routinized forms of screening

Q: "I think parents should be..."



# From the discussions:

## On anxiety [false positive]:

**Navigating Harms:**  
 “I think in my mind that it’s worth it for those nine families to have that experience to avoid this one family having whatever the experience of not catching this might be like.”

[screening] should be mandatory

## On unwanted information:

“I don’t really see it as one way or another [then], I see this one [DMD] knowing parents should be allowed to have four years ahead that they may want to enjoy the baby without technically healthy. So, like, personally, I’d rather not know and just, like, love my child for those [years]”

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# Key Messages

- Citizens values are consistent with wider notions of benefit:
  - Strong support for broad scope of screening
  - Treatment was a primary rationale (welfare of the *child*)
  - Information is perceived as an additional, even sufficient, benefit
  - High tolerance for harms
- Despite enthusiasm, we assert caution around unlimited uptake of infant screening
- Further public engagement warranted

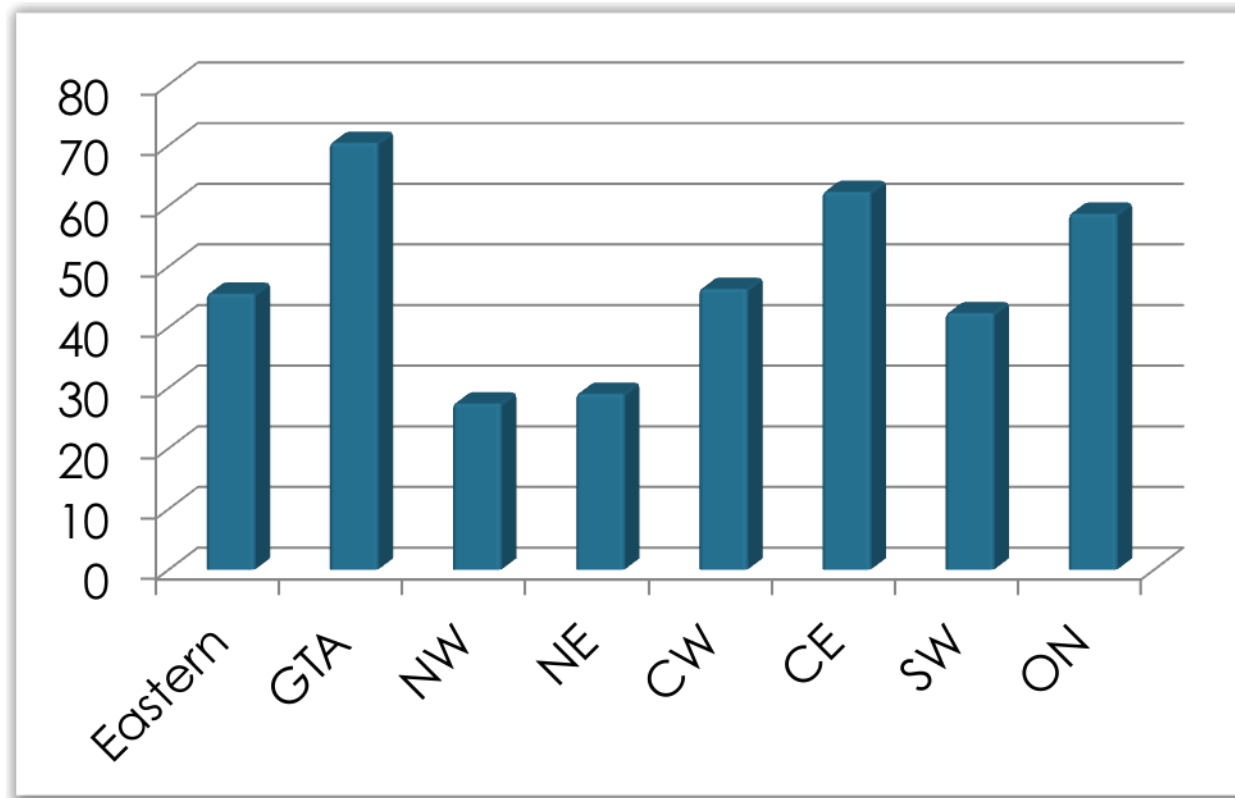
# In Summary

- Expanded NBS is complex and value-laden
- Challenges persist re: ensuring effective delivery of expanded NBS
- Much remains to be learned about evaluating and governing this (and other) screening systems
- Current data and governance infrastructure favourable

# Future Directions

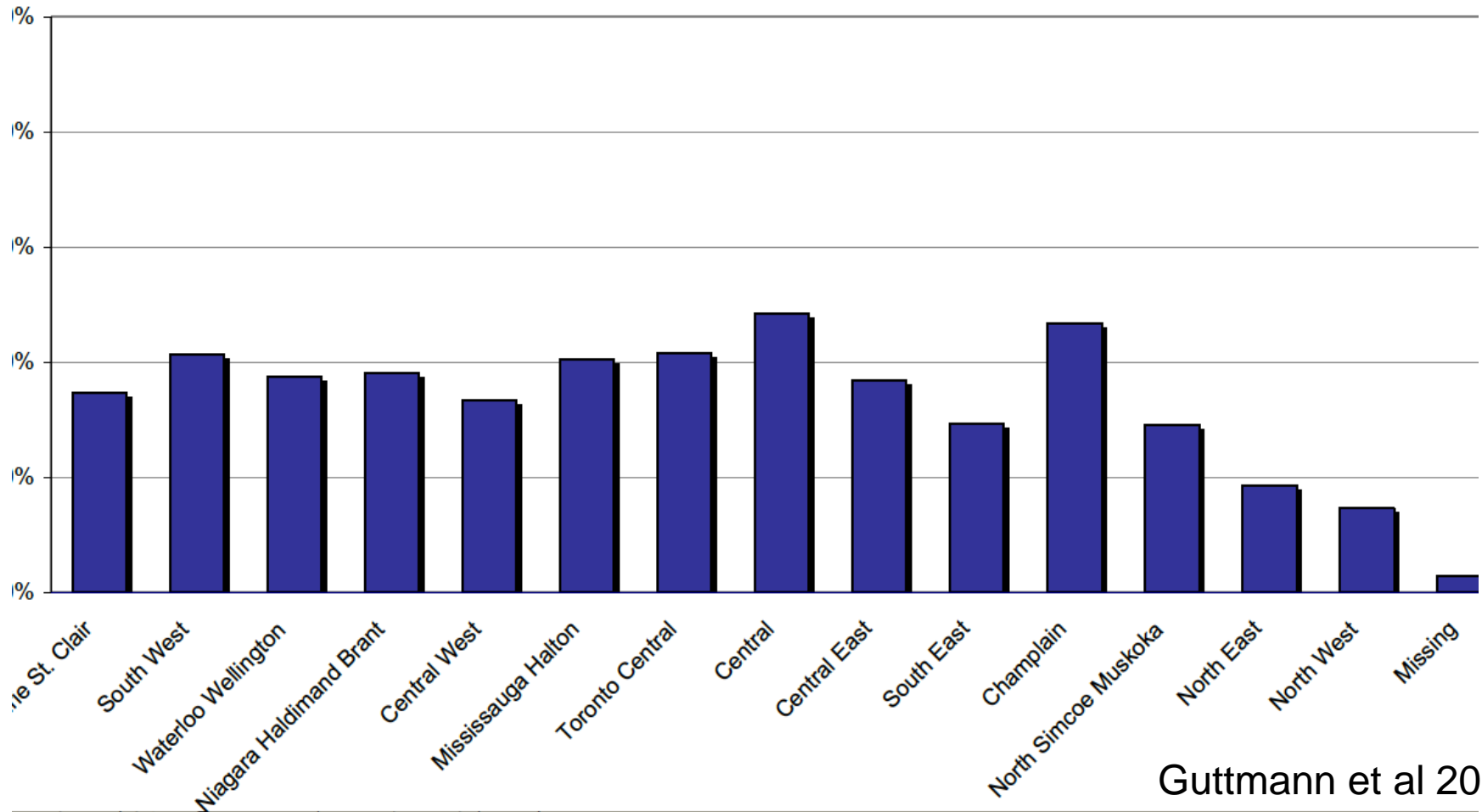


## Proportion of pregnant women receiving prenatal screening varies by LHIN (Ontario 2008).



**Goal: 100% offer of screening test**

## Proportion of children receiving enhanced well baby screening is low and varies by LHIN (Ontario, 2010).



Guttman et al 2011

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Thank you

**QUESTIONS/COMMENTS**