# Reconciling the downsides of screening: Mothers' experiences with false positive newborn screening results for cystic fibrosis

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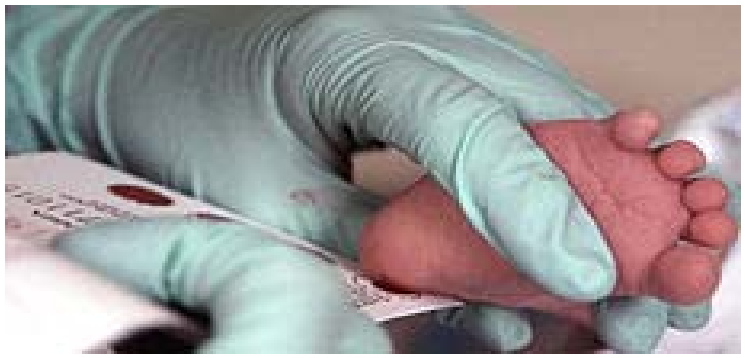
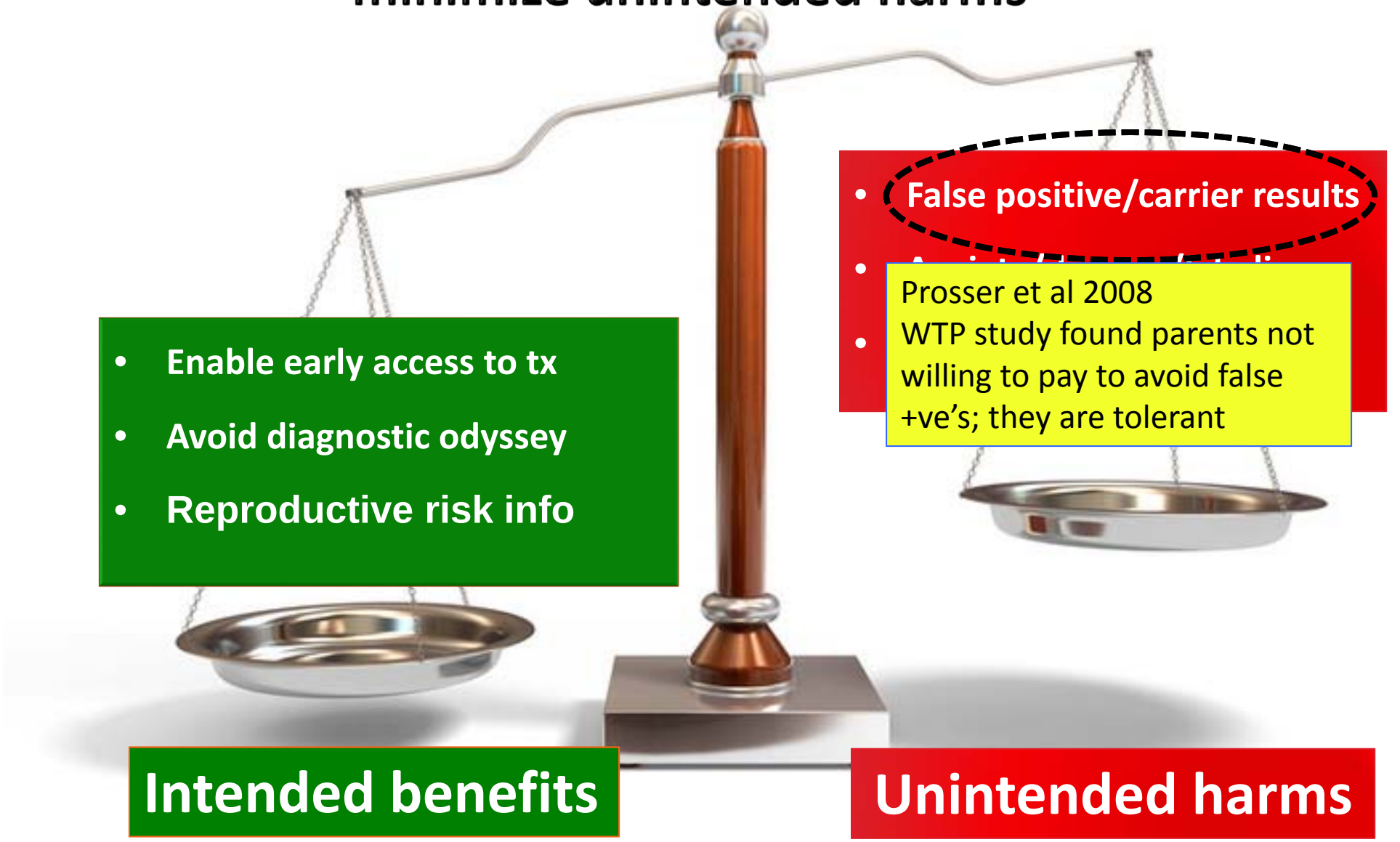
# Newborn Screening (NBS)

- Newborn blood spot screening is a longstanding population screening program, in place since 1960s
- "Heel prick test" at 2-3d of age
- Targets serious treatable disorders not clinically evident in newborn period, enables early intervention
- But large #'s false positive results are generated
  - Majority are carrier results – unaffected but repro risk associated
- In Ontario, NBS expanded in 2006, now includes 29 disorders
  - Coordinated from single lab (Newborn Screening Ontario, NSO)
  - ~140,000 babies screened/yr
  - ~1400 screen positive (~1%)
  - ~140 have disease (~0.1%)



# Enduring challenge: maximize intended benefits & minimize unintended harms



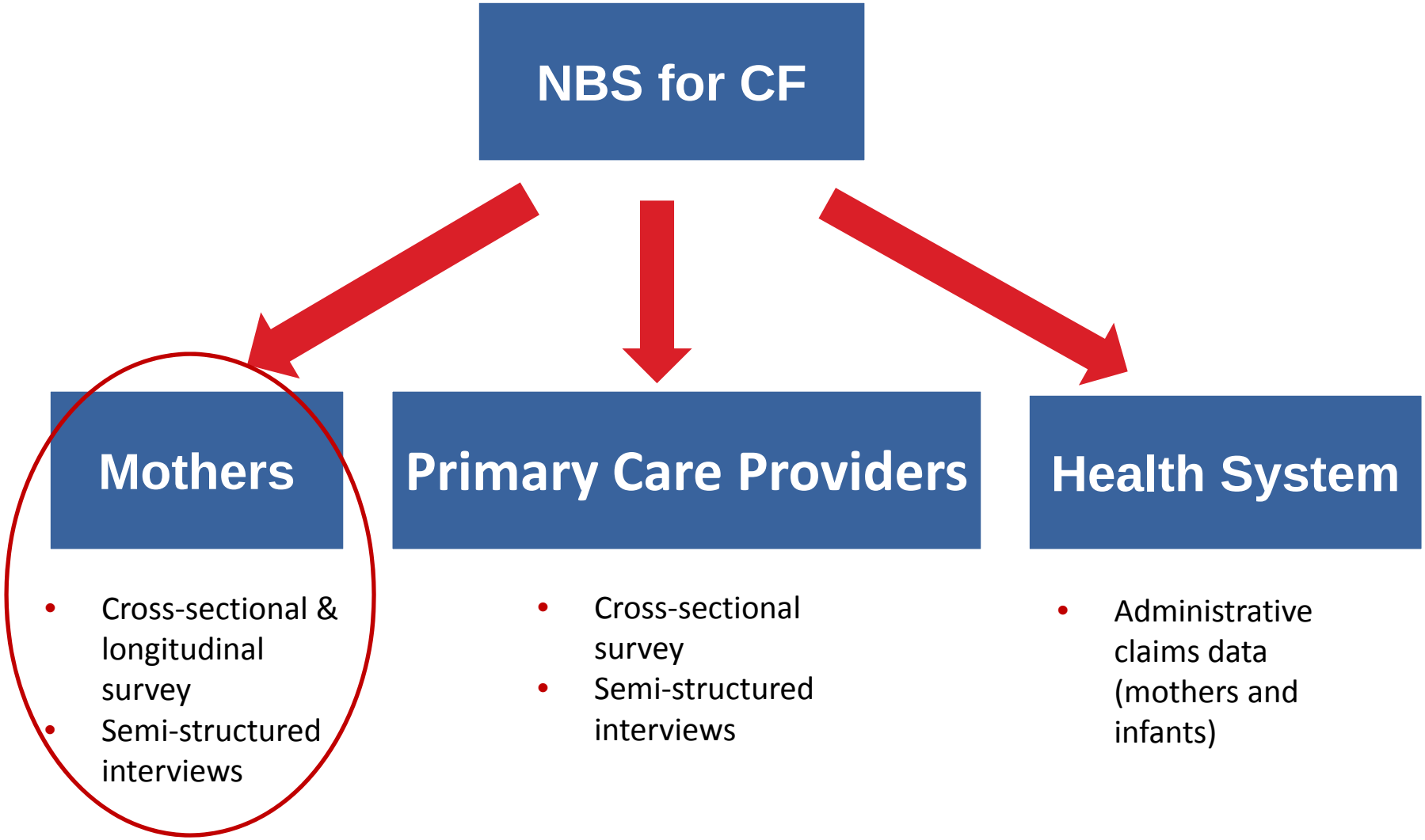
# Cystic fibrosis (CF) as a paradigm

- CF added to NBS panel in Ontario in 2008; also in BC, AB, MB, NS since then
- CF is an inherited condition that leads to poor lung function, respiratory infection, and inadequate nutrition
- Evidence that identifying CF in newborns and intervening early improves nutritional status, height and weight gain
- In Ontario, there are about 400 screen positive cases identified each year
  - Over 80% are false positives (mostly carriers)

# Research Question

**What are mothers' experiences with false positive newborn screening results for cystic fibrosis?**

# Study Design: Longitudinal mixed-methods cohort study



# Methods

- Cross-sectional & longitudinal survey
  - Surveyed mothers of infants with false positive results within 3 months of birth
  - Data collection:
    - Time 1 (T1): April 2011 – December 2012
    - Time 2 (T2): April 2012 – January 2014
  - Items included: demographics, **NBS experience**, carrier status and cascade testing, knowledge of CF, **attitudes towards NBS**, psychosocial measures
  - Survey included a question requesting consent to contact for qualitative interview
  - Analyzed using descriptive statistics

# Methods

- Semi-structured interviews
  - Participants who agreed contacted by telephone and interview arranged
  - In person or by telephone, depending on participant preference and logistics
  - Data collection
    - T1: October 2011 – March 2013
    - T2: November 2012-December 2013 (analysis underway)
  - Explored participants' **NBS experience** (notification, confirmatory testing, role of primary care provider in the process), carrier status and cascade testing, knowledge of CF, **attitudes towards NBS**
  - Analyzed using qualitative description

# Quantitative results

Completed surveys from mothers:

- T1: 134 of 245 (55%)
- T2: 95 of 214 (44%)

# Survey Sample Characteristics

| | Time 1<br>n (%)<br>(N=134) | Time 2<br>n (%)<br>(N=95) |
|---|---|---|
| **Marital Status** | | |
| Married/common law | 123 (91.8) | 91 (95.8) |
| Other | 11 (8.2) | 4 (4.2) |
| **Education level** | | |
| High school or less | 18 (13.4) | 11 (11.7) |
| College | 54 (40.3) | 37 (39.4) |
| Undergrad or higher | 62 (46.2) | 46 (48.9) |
| **Income** | | |
| <$80,000 | 52 (40.3) | 28 (31.1) |
| >$80,000 | 77 (59.7) | 62 (68.9) |
| **City Size** | | |
| 100,000+ | 93 (69.9) | 61 (65.6) |
| <100,000 | 40 (30.1) | 32 (34.4) |

# Mothers' Attitudes – T1 & T2

| | T1 - False Positive (n,%) | | T2 – False Positive (n,%) | |
|---|---|---|---|---|
| **Feel better informed knowing NBS result** | | | | |
| Strongly agree | 93 (69.9%) | **127 (95.5%)** | 64 (67.4%) | **85 (89.5%)** |
| Agree | 34 (25.6%) | | 21 (22.1%) | |
| Neutral | 4 (3.0%) | | 8 (8.4%) | |
| Disagree | 1 (0.8%) | 2 (1.5%) | 1 (1.1%) | 2 (2.1%) |
| Strongly disagree | 1 (0.8%) | | 1 (1.1%) | |
| **Feel relieved about NBS result** | | | | |
| Strongly agree | 92 (69.2%) | **120 (90.2%)** | 66 (69.5%) | **82 (86.3%)** |
| Agree | 28 (21.1%) | | 16 (16.8%) | |
| Neutral | 11 (8.3%) | | 8 (8.4%) | |
| Disagree | 1 (0.8%) | 2 (1.5%) | 2 (2.1%) | 5 (5.3%) |
| Strongly disagree | 1 (0.8%) | | 3 (3.2%) | |

- Majorities agreed to **feeling better informed** knowing result
- Majorities agreed to **feeling relieved** about NBS result

# Mothers' Attitudes – T1 & T2

| | T1 - False Positive (n,%) | | T2 – False Positive (n,%) | |
|---|---|---|---|---|
| **Learning about false positive result is disadvantage** | | | | |
| Strongly agree | 14 (10.4%) | **43 (32.1%)** | 13 (13.7%) | **38 (40.0%)** |
| Agree | 29 (21.6%) | | 25 (26.3%) | |
| Neutral | 24 (17.9%) | | 21 (22.1%) | |
| Disagree | 45 (33.6%) | 67 (50.0%) | 21 (22.1%) | 36 (37.9%) |
| Strongly disagree | 22 (16.4%) | | 15 (15.8%) | |
| **Learning about carrier result is disadvantage** | | | | |
| Strongly agree | 1 (0.7%) | **6 (4.5%)** | 2 (2.1%) | **3 (3.2%)** |
| Agree | 5 (3.7%) | | 1 (1.1%) | |
| Neutral | 10 (7.5%) | | 8 (8.4%) | |
| Disagree | 53 (39.6%) | 118 (88.1%) | 33 (34.7%) | 84 (88.4%) |
| Strongly disagree | 65 (48.5%) | | 51 (53.7%) | |

- Minorities agreed that learning about false +ve was a **disadvantage of screening**
- Minorities agreed that learning about carrier result was a **disadvantage of screening**

# Qualitative results

Completed interviews with participants:

- T1: 31
- T2: 35 (analysis underway)

# Distress

- News is unexpected
- Wait time between initial phone call and sweat test
- Logistics (distance to-from treatment centre)

# What makes it tolerable?

## Benefit for me

- Reproductive benefit
- Learning experience
- Clean bill of health

## Benefit for others

- Ethics of care
- Value of early disease diagnosis

## System of care

- Quick notification and retrieval of results
- Supportive treatment centre team
- Trust in the system

## Downplay of burden

- Weighing and balancing the pros and cons of experience

# Mothers report false +ve experience to be highly distressing

## The news is unexpected

*"Well, I guess it kind of blew me out of the water because we signed the form in the hospital, didn't really think much about it. Like, we kind of knew that they did it, but didn't know anything really about it, so forgot about it. So,* ***when the call came it knocked me on my butt****."* [050)

## Waiting for sweat test appointment is hard

*"That 24 hours was wicked.* ***It was the most awful 24 hours I think I've ever endured****. I just couldn't stop crying, I could hardly even look at [infant], I was holding [infant] and I just cried, I literally cried the whole night…" [234]*

## Logistics are challenging (travel distance to-from tx centre)

- *"Well, I mean, it was* ***quite crazy driving there, we live all the way up north*** *so we barely made it there on time, even though we left plenty of time, it took over 3 hours…so the drive down was stressful just because you're worried about everything…we got down there just in time." [173]*

## However, tolerance for distress was expressed in 4 ways:

- Identifying benefit for self
- Identifying benefit for others
- Appreciating the system of care
- Downplaying the burden in retrospect

# Tolerance for distress: Benefit for self

## Reproductive information

*"**I think it's important that we know [infant] is a carrier, like for future children** because I would never want my kid to have CF or any disease that we could try to prevent…My husband and I got tested, so we know that one of us is a carrier" [246]*

## Learning experience

*"I think the experience was very humbling…I mean, it sort of, in an odd way, I think parents get so freaked out about things that can happen to your children, and it actually just took some of that worrying away a little bit. **I just felt like, I don't know…better equipped…you know, a little bit stronger, a little bit tougher**, just I worry less…I worry a bit less now." [182]*

# Tolerance for distress: Benefit for others

## Ethics of care

*"**It's better to catch a kid than have him fall through the cracks…if it means that we get a false positive, I prefer that than a little kid going without detection.** You know, maybe that little kid that got caught as a result of a big catchment, then maybe…you know, maybe they will live for longer or lived longer…whatever it may be" [234]*

# Tolerance for distress: System of care

## Quick notification and retrieval of results

*"So thank goodness we didn't have to wait, like, a week for an appointment…**we were just so relieved to get it done right away**…" [187]*

## Treatment centre team help to mitigate stress

*"Even though it was a really stressful day, and even before we got the test results back, **it was a very positive and supportive environment**, yeah, like I was quite amazed at the level of attention and care we received" [182]*

## Trust in the system

*"I'm **glad to know that there's a program in place for those that need it**. The whole process was very smooth…we got the information we needed…so, even if the outcome had been … cystic fibrosis, **I'm confident that we would have been given the care we needed and I guess the process would have been in place to kind of move that forward for us**…" [035]*

# Tolerance for distress: Downplaying, in retrospect

## Weighing and balancing the pros and cons of experience

*"So, it's just having to go through all that worry on that one day.* ***It was only a day for us, which wasn't a big deal in the grand scheme of things****…" [090]*

*"At the time, it was a stressful thing, but I don't know…****I chalk it up to experience****. Having that week of stress kind of put us in the shoes of other parents who are dealing with kids who have a chronic illness. So, I think it's…****I appreciate that perspective despite how stressful it was****…" [170]*

# Discussion

- We identify significant tolerance for false positive NBS results
  - Not because there is no distress – distress *is* experienced
  - But because this distress is experienced as tolerable

- Distress is made tolerable in several ways:
  - Finding benefit for self
    - Importantly, not only 'reproductive benefit'
    - Also benefit of the leaning experience and of a 'clean bill of health' for infant
  - Belief in importance of benefit for others
  - *Systems of care* that help to mitigate burdens
  - Rationalization of distress as 'not that bad'

# Conclusion

- False positive results are a frequent and concerning downside of population screening programs such as newborn screening
  - We know that distress occurs (anxiety, depression) but it is limited and not sustained in the context of newborn screening
  - “Reproductive information” has been emphasized as a key benefit for families who receive false positive results
- Our study identifies significant tolerance for false positive NBS results
  - Points to importance of investing in a system of care when population screening proposals are mounted
  - Points to screening as a valued collective good
  - Challenges the presumption that reproductive risk information is the key benefit or sole source of solace in this context

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# Thank you

**Questions?**