

## WHAT

Measure the performance and impact of Ontario's prenatal screening system for aneuploidy after the incorporation of cfDNA testing, also known as NIPT (which measures placental DNA circulating in the blood of the pregnant person).

## WHERE

In Ontario, pregnant individuals are offered publicly funded multiple marker screening (which includes measuring hormones from the blood, and other information). Publicly funded cfDNA screening is available to those with an increased chance of having a pregnancy with certain aneuploidies, like trisomy 21 (Down syndrome) or trisomy 18.

## WHO

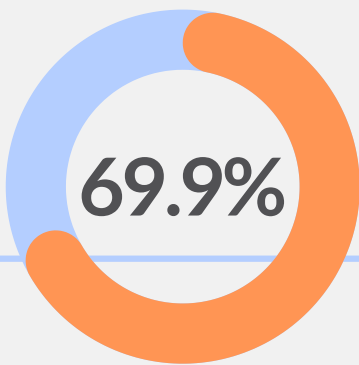
Data from 373,682 singleton pregnancies with a due date between Sept 1, 2016 and March 31, 2019, that were offered publicly funded prenatal screening.

## WHY

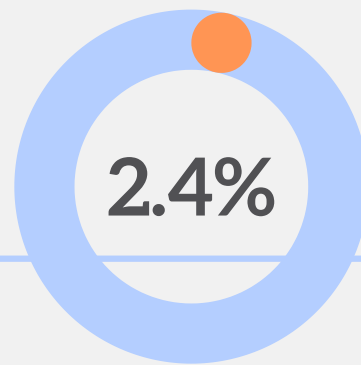
Evaluating real-world prenatal screening system performance is important for making improvements to the system and supporting decision making for pregnant individuals.

# Performance of Prenatal Screening for Aneuploidy in Ontario

A **population-based** study examining routinely collected data from **BORN Ontario**, the province's perinatal registry.



Prenatal screening is a choice. The offer of prenatal screening was accepted for 69.9% of all singleton pregnancies



2.4% of screened pregnancies had invasive prenatal diagnostic testing, a 2-fold decrease since cfDNA screening was introduced

### OVERALL SENSITIVITY

trisomy 21 | 89.9%

trisomy 18 | 80.5%

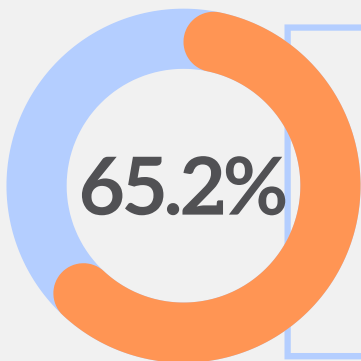
Sensitivity = proportion of pregnancies with trisomy 21 / trisomy 18 that received positive screening results

### OVERALL SPECIFICITY

trisomy 21 | 98.8%

trisomy 18 | 99.9%

Specificity = proportion of pregnancies without trisomy 21 / trisomy 18 that received negative screening results



65.2% of pregnancies with a screen-positive cfDNA result had invasive prenatal diagnostic testing. Pregnant people continue to make individual decisions about prenatal testing

## Conclusion

This study demonstrates that pregnant Ontarians have access to a robust, high-quality prenatal screening system that reduces invasive prenatal diagnostic testing and supports informed decision making.