Research Square

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Prenatal aneuploidy screening in metropolitan Melbourne: Uptake and sociodemographic variation in cell-free DNA based non-invasive prenatal testing

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Abstract

Objectives: To measure prenatal aneuploidy screening uptake and test choice, including non-invasive prenatal testing (NIPT), and to investigate variation by maternal sociodemographic and clinical characteristics.

Study Design: Retrospective multi-centre cohort study.

Setting, Participants: Women who gave birth at ≥20 weeks' gestation across 11 public hospitals in Melbourne between January 2022 and December 2024.

Main Outcome Measures: Main outcomes were prenatal aneuploidy screening uptake and screening test choice.

Results: Of 102,729 eligible women in this study, prenatal screening data were available for 80,355 (78.2%). Among these, 66,343 (82.6%) had aneuploidy screening, 12,632 (15.7%) declined screening, and 1,380 (1.7%) were not offered screening. Among women who had screening, NIPT was the most common choice (80.6%, n=53,475), followed by combined first trimester screening (16.6%, n=10,991) and second trimester serum screening (2.5%, n=1,634).

Compared with women who accepted screening, those who declined or were not offered screening were more likely to be <25 years of age (adjusted relative risk ratio [aRRR] 3.86, 95% CI:3.75 to 3.96; and aRRR 4.30, 95% CI:4.06 to 4.56, respectively), or smoke during pregnancy (aRRR 1.22, 95% CI:1.18 to 1.27; and aRRR 1.86, 95% CI:1.72 to 2.00, respectively).

Among the 66,343 women who had screening, NIPT was more common among those aged ≥35 years (adjusted incident rate ratio [aIRR] 1.07, 95% CI:1.05 to 1.10), lived in socioeconomically advantaged areas (aIRR 1.09, 95% CI:1.06 to 1.13) and had private obstetric care (aIRR 1.11, 95%CI:1.04 to 1.19).

NIPT uptake was lower among women with parity ≥3 (aIRR 0.73, 95% CI:0.69 to 0.77), and those born outside Australia, particularly Oceania (aIRR 0.73, 95% CI:0.63 to 0.86).

Conclusions: This study provides the first large-scale assessment of prenatal screening utilisation by women birthing in Australian public hospitals. NIPT was used by 80% of women undergoing screening. Clustering of sociodemographic factors among women who declined screening, or used non-NIPT forms of prenatal screening, may indicate inequities in access to, or uptake of, screening. These findings highlight the need to investigate potential structural barriers, including direct patient costs, and to consider policy options that support more equitable access to evidence-based prenatal screening.

PLAIN LANGUAGE SUMMARY

The known:

Non-invasive prenatal testing (NIPT) is a recommended screening option for trisomies 21, 18 and 13. Despite superior accuracy, the lack of Medicare rebate means it remains the most expensive (~AUD$500) option for prenatal screening in Australia.

The new:

NIPT was used by 80% of women who had screening and gave birth at 11 Melbourne public hospitals. Lower uptake occurred among women who were socioeconomically disadvantaged, born overseas, multiparous, or younger than 30 years.

The implications:

Differences in NIPT uptake may indicate inequities in access to prenatal screening. They highlight the need to examine barriers, including out-of-pocket costs, and consider policy options to support more equitable antenatal care.

INTRODUCTION

Prenatal screening for fetal trisomy 21, trisomy 18 and trisomy 13 is recommended for all pregnant women in Australia because these conditions are associated with substantial perinatal morbidity and mortality.[1] Until 15 years ago, our prenatal screening tests relied on combinations of maternal age, ultrasound and serum markers to assess the probability of fetal aneuploidy. In 2012, non-invasive prenatal testing (NIPT) based on sequencing of cell-free DNA in maternal blood became commercially available in Australia.[2] NIPT is now established as the most accurate screening test for trisomies 21, 18, and 13. For trisomy 21, reported sensitivity is 99% with a false positive rate of < 0.1%, compared with 87% sensitivity and a 5% false positive rate for combined first trimester screening (CFTS), and 75% sensitivity with an 8% false positive rate for second trimester serum screening (STSS).[3–5]

The Living Evidence for Australian Pregnancy and Postnatal Care (LEAPP) guidelines[6] and the Royal Australian and New Zealand College of Obstetricians and Gynaecologists (RANZCOG)[1] recognise the high accuracy of NIPT, with RANZCOG recommending it be offered to all pregnant women interested in screening. These recommendations have supported the incorporation of NIPT into Australian clinical practice on a self-funded basis.

Despite its superior accuracy and professional endorsement, NIPT is not subsidised by public or private health insurance in Australia. In contrast, CFTS is partially subsidised and STSS fully subsidised by government.[7] NIPT typically costs AUD$400–500 out of pocket, which may influence test choice and has raised persistent concerns about equity of access.[8–10] Indirect evidence of variation has been observed, with women from socioeconomically advantaged areas more likely to undergo NIPT-indicated diagnostic testing than those from disadvantaged areas.[11] Given professional recommendations, such variation is unlikely to be explained by clinical need or patient preference alone,.[12, 13]

Previous applications to the Australian Medical Services Advisory Committee (MSAC) for public funding of NIPT have been unsuccessful, citing cost-effectiveness concerns and the lack of contemporary population-based data.[14] The most recent state-wide Victorian data from 2015 reported that 84% of pregnant women underwent prenatal screening, of whom 73% had CFTS, 20% had NIPT, 5% had STSS and 2% had a combination of tests.[15] More recent national survey data from 2023 suggest that NIPT uptake has now overtaken CFTS, with most clinicians offering NIPT as the first-line screening test.[9] This trend is supported by Medicare data showing a 40% reduction in claims for CFTS between 2013-2021.[16]

Australia lacks systematic, population-based data on contemporary prenatal screening utilisation. Such data are needed to monitor prenatal screening practices and to inform clinical and policy decisions.[17] The aims of this study were to assess (i) overall uptake of prenatal screening, (ii) the proportions of women utilising NIPT, CFTS, or STSS, and (iii) associations between prenatal screening choice and maternal sociodemographic and clinical factors across public maternity hospitals in metropolitan Melbourne.

METHODS

Study design

This was a secondary analysis of the Collaborative Maternity and Newborn Dashboard for the COVID-19 pandemic (CoMaND) research dataset. The CoMaND project was a multi-centre retrospective cohort study that monitored the impact of COVID-19 pandemic on perinatal and obstetric outcomes across 12 public hospitals in metropolitan Melbourne.[18] The CoMaND dataset includes perinatal data on over 300,000 births at ≥ 20 weeks' gestation from 12 public maternity services in metropolitan Melbourne from January 2018 to December 2024.[19] Data on pregnancy outcomes < 20 weeks' gestation, including miscarriage and termination of pregnancy, were not available.

In Australia, prenatal screening for fetal chromosome conditions is usually discussed and arranged in primary care, most commonly by general practitioners, prior to referral for hospital-based maternity care. Hospitals may also arrange prenatal screening if it has not previously been offered, or if a new clinical finding raises concern for a chromosome condition (e.g. following an ultrasound finding). On 1 July 2021, public hospitals in metropolitan Melbourne began routinely collecting prenatal screening data within their maternity data collection. The fixed-response fields for aneuploidy screening captured screening status ("accepted", "declined", "not offered"), screening type (NIPT, CFTS, STSS,) and screening result (low chance, high chance, failed, not performed, result unknown). These fields were typically completed at the first hospital antenatal clinic visit by the reviewing clinician, based on the maternal history and investigation results provided by the referring clinician, but could be updated throughout the pregnancy if required. As all data was extracted after the birth outcome, the dataset reflected the most up-to-date screening status for each pregnancy. Information on the specific chromosomal condition suspected on prenatal screening (e.g. a high chance result for trisomy 21) and results of confirmatory

prenatal or postnatal diagnostic testing were not available. However, data on congenital anomalies (including genetic and chromosomal conditions) recorded in the maternity record were included in the CoMaND dataset.

Although data collection on prenatal screening was not initially mandatory, these fields were completed for approximately 70% of births between 1 July 2021 and 31 December 2024.[19] From the 1 July 2024, prenatal screening data collection became mandatory in the Victorian Perinatal Data Collection, which improved data completeness.[20] For this study, we used a subset of CoMaND data from 11 of 12 participating hospitals from 1 January 2022 to 31 December 2024. We excluded a hospital (representing 8.2% of births during the study period, Fig. 1) as prenatal screening data was not available. Collectively, these 11 hospitals accounted for approximately 70.9% of hospital births in metropolitan Melbourne during the study period (See Supplementary Table 1 for details).

Prenatal aneuploidy screening

(i) CFTS incorporates maternal age, fetal crown-rump length, nuchal translucency measurement (with or without nasal bone assessment), and maternal serum biomarkers (pregnancy-associated plasma protein-A and free beta-human chorionic gonadotropin (beta-hCG)) to estimate the chance of trisomies 21, 18 and 13.[1] Maternal serum biomarkers are collected between 9 + 0 to 13 + 6 weeks gestation, with ultrasound assessment performed between 11 + 0 to 13 + 6 weeks. (ii) STSS incorporates maternal age and maternal serum biomarkers (alpha-fetoprotein, beta-hCG, unconjugated estriol and inhibin A), measured at 14 to 20 weeks' gestation, to estimate the chance of trisomies 21 and 18.[1] (iii) NIPT analyses maternal cell-free DNA from 10 weeks' gestation and depending on the platform, can screen for a range of chromosomal conditions.[1]

Data analysis

Statistical analysis was conducted using StataSE version 19.5.[21] Descriptive statistics were used to summarise maternal, hospital, and prenatal screening characteristics. Categorical variables were reported as counts and proportions with 95% confidence intervals (CI), and group differences were assessed using chi-square test.

Prenatal screening uptake was examined as a categorical variable (accepted, declined, not offered). Among women with recorded screening, screening type (NIPT, CFTS, STSS, test type not specified) was described by hospital and maternal clinical characteristics.

Associations between maternal clinical and sociodemographic factors and prenatal screening status (screening accepted, declined, not offered) were estimated using multinomial logistic regression, with results reported as relative risk ratios (RRR) and 95% confidence intervals.

Associations between maternal clinical and sociodemographic factors and prenatal screening type (NIPT vs all other screening types) were estimated using multivariable Poisson regression, with results reported as incidence rate ratios (IRR) and 95% confidence intervals.

We adjusted for covariates previously associated with prenatal screening uptake or known to influence prenatal screening test choice, as well as markers of healthcare engagement. This included maternal age, body mass index (kg/m^2), parity (births ≥ 20 weeks' gestation), maternal region of birth (coded from country of birth using Australian Bureau of Statistics regions)[22], area level socioeconomic status based on postcode (Australian Bureau of Statistics, Index of Relative Socioeconomic Advantage and Disadvantage (IRSAD))[23], smoking status during pregnancy, gestation at first antenatal visit, vaccination status (pertussis and COVID-19), hospital site, and model of care (private obstetric care within public hospitals vs. all other models of care).

We used multiple imputation by chained equations to account for missing data in prenatal screening status and covariates (including hospital name, maternal age, BMI, maternal region of birth, socioeconomic status, and COVID-19 and pertussis vaccination status), generating six imputed datasets under a missing-at-random assumption.[24] Regression analyses were performed using the pooled estimates from imputed datasets, with complete-case analyses conducted as a sensitivity analysis.

An arbitrary 5% difference in IRR and RRR was considered potentially clinically important.

Ethics approval

Ethics approval

was obtained from X Human Research Ethics Committee (Ref no. 2023-027, 21 November 2023).

RESULTS

Over the 3-year study period from 2022 to 2024, 102,729 women gave birth at ≥ 20 weeks' gestation across 11 hospitals (Fig. 1**)**.

Missing vs. recorded prenatal screening data

Prenatal screening data were recorded for 80,355 (78.2%) of births, with missing data for 22,374 births (21.8%) (**Supplementary Table 2**). Women with missing screening data differed from those with recorded data across several sociodemographic and clinical characteristics. Missing data were more common among women who were younger (< 25 years), with higher BMI (≥ 40kg/m^2), or living in more socioeconomically disadvantaged areas. Higher proportions of missing data were also observed among women born in Oceania (excluding Australia and New Zealand) or North Africa and the Middle East,

women who smoked during pregnancy, multiparous women, those attending their first antenatal visit after 12 weeks' gestation, and women who were unvaccinated for COVID-19 or pertussis.

Prenatal screening uptake

Of 80,355 births with completed records, 66,343 (82.6%) accepted prenatal screening, 12,632 (15.7%) declined screening, and 1,380 (1.7%) had not been offered screening (**Table 1**).

In adjusted multinomial models, several maternal sociodemographic and clinical characteristics were associated with declining or not being offered prenatal screening (for full estimates see **Table 1**). Compared with women who accepted screening, those who declined or were not offered screening were more likely to be younger (< 25 years), born in Oceania (excluding Australia and New Zealand) or Sub-Saharan Africa, live in socioeconomically disadvantaged areas, smoke during pregnancy, be multiparous, receive public obstetric care, or be unvaccinated for COVID-19 or pertussis.

For some covariates, associations differed by screening status. For example, compared with Australian-born women, women born in North-East Asia were less likely to decline screening (aRRR 0.58, 95% CI:0.53–0.63) but more likely to have screening recorded as not offered rather than accepted (aRRR 1.54, 95% CI:1.33–1.80). Similarly, women who attended their first antenatal appointment after 12 weeks' gestation were more likely to have screening recorded as not offered rather than accepted (aRRR 2.18, 95% CI:2.09–2.28), compared with those who attended by 12 weeks' gestation, whereas this association was not observed for those who declined screening (**Table 1**).

Prenatal screening type

Among the 66,343 women who had prenatal screening, NIPT was the most common type (80.6%), followed by CFTS (16.6%) and STSS (2.5%). Screening type was unspecified for 0.3% of births (Fig. 1).

Factors associated with NIPT uptake

Factors associated with higher NIPT uptake included advanced maternal age (≥ 35 years; aIRR 1.07, 95% CI 1.05–1.10), living in a socioeconomically advantaged area (IRSAD quintile 5, aIRR 1.09, 95% CI 1.06–1.13), and receiving private care (aIRR 1.11, 95% CI 1.04–1.19) (**Table 2**).

Factors associated with lower NIPT uptake included multiparity (≥ 3; aIRR 0.73, 95%CI 0.69–0.77) and maternal country of birth outside Australia. The strongest negative associations were observed for women born in Oceania (excluding Australia and New Zealand) (aIRR 0.73, 95% CI 0.63–0.86) and Sub-Saharan Africa (aIRR 0.77, 95% CI 0.71–0.84) (**Table 2**).

DISCUSSION

This study provides the first large-scale contemporary analysis of prenatal screening uptake and test choice in Australian public hospitals since the last statewide Victorian report in 2015.[15] Overall prenatal screening uptake remained high, while NIPT has become the predominant screening method. Both screening uptake and screening test choice varied according to maternal sociodemographic and clinical characteristics. Together, these findings suggest that prenatal screening use among women giving birth in Melbourne public maternity hospitals is shaped not only by individual preferences, cultural values, and attitudes towards testing, but also by system-level, financial, and access factors.

While overall prenatal screening uptake was high, variation by sociodemographic characteristics was evident. Some variation is expected because screening is voluntary and reproductive decisions are influenced by personal values, beliefs, and cultural norms.[26] Lower screening uptake was observed among women younger than 25 years, women born in Oceania or Sub-Saharan Africa, and women with two or more children. Women who declined screening or were recorded as not having been offered screening were also less likely to be in private obstetric care within a public hospital. However, the extent to which these differences reflect patient preference, informed choice, barriers to access, or limitations of the screening status variable cannot be determined from our data.

The marked increase in NIPT use among women who underwent prenatal screening, from 20% of all screening tests in 2015[15] to more than 80% in 2024, shows that NIPT has become the dominant screening modality in this population. This rapid NIPT uptake, despite the substantial out-of-pocket costs, suggests that clinicians and pregnant women place a high value on the improved accuracy and earlier reassurance that NIPT offers. This is consistent with Australian survey data reporting that most clinicians offer women a choice between NIPT and CFTS, and that many women are willing to pay for what they perceive as the most accurate screening test available.[9, 27]

However, lower NIPT use among groups more likely to experience barriers to care suggests potentially unwarranted healthcare variation.[12, 13, 28] Women who entered antenatal care after 12 weeks' gestation, lived in socioeconomically disadvantaged areas, were unvaccinated for pertussis in pregnancy, or continued to smoke during pregnancy were less likely to use NIPT. This interpretation aligns with previous Australian studies showing that younger women, lower-income women, and rural residents report cost, information gaps, and lack of recommendation from health professionals as barriers to NIPT.[9, 10, 29]

These differences in NIPT uptake may expose women to avoidable harms. Compared with NIPT, the lower sensitivity of CFTS and STSS increases the likelihood of missed or delayed diagnosis of a fetal chromosome condition. Their lower specificity also increases the likelihood of false positive screening results, and in turn, confirmatory invasive prenatal diagnostic testing.[30] In Victoria in 2023, among women undergoing prenatal diagnostic testing after a high chance screening result, the positive predictive value was 60% following NIPT, compared with 22% following CFTS.[31]

Addressing these inequities will likely require structural change. The absence of public funding for NIPT in Australia has been consistently identified by clinicians and women as a major barrier to access, particularly those on lower incomes and in rural areas.[17, 29, 32] Previous Medical Services Advisory Committee (MSAC) applications to support a Medicare Benefits Schedule (MBS) listing for NIPT found that, although NIPT is clinically superior to CFTS with higher detection rates of trisomy 21 and fewer procedure related pregnancy losses, it was not cost-effective as a universal first-line or contingent test under the assumptions made at the time.[14, 33–35] Our contemporary data show that NIPT has already become the first-line screening test for > 80% of women accessing public hospital care with recorded prenatal screening data, and that self-funding appears to contribute to socioeconomic inequities in access. A future MSAC application could therefore consider a more targeted funding model, prioritising women on lower incomes or those living in rural and remote areas, where access to CFTS or STSS may also be limited. Future planned work will link statewide prenatal screening data with diagnostic and pregnancy outcome datasets to quantify the benefits and harms of current practice, to model the impact of public funding, and to identify implementation strategies that reduce unwarranted variation in prenatal screening uptake and test choice (RCH HREC ref. no 116666).

Strengths and limitations

Key strengths of this study include the large multicentre design and broad geographic coverage across metropolitan Melbourne public maternity services. Unlike prior single-centre studies[36, 37] or analyses limited to prenatal diagnostic procedures[11, 38], this study captured screening uptake and test choice, including women who declined or were not offered screening. This enabled a more comprehensive assessment of utilisation patterns and variation in real-world practice. However, prenatal screening data were only available for pregnancies at ≥ 20 weeks' gestation, so early miscarriages and termination of pregnancy were not captured. As a result, the number of pregnancies having prenatal screening and screening type will be underestimated, as our findings may not reflect screening pathways and decision-making earlier in pregnancy. Screening status was missing for around one in five births (22,374/102,729, 21.8%) because these fields were not mandatory during the entire study period, and missingness was more common among younger women, those from more socioeconomically disadvantaged areas, and some migrant groups. Importantly, even under a conservative assumption that women with missing aneuploidy screening data had similar overall screening uptake (82.6%) but none underwent NIPT, estimated NIPT uptake would be 63.0% among women who accepted screening. Finally, this study included women booked for maternity care at metropolitan public hospitals only and may not reflect practices in private and regional settings, where patterns of screening uptake, test choice, and ability to self-fund NIPT may differ.

CONCLUSION

Among women birthing in public maternity hospitals in Melbourne, self-funded NIPT has become the predominant prenatal screening test, but utilisation varies substantially across population groups.

Although overall prenatal screening uptake was high, access to the most accurate screening test appeared uneven and may reflect both personal and structural barriers. These findings provide contemporary evidence to support policy responses, including funding models, aimed at reducing unwarranted variation and supporting equitable access to best practice in prenatal screening.

Declarations

Human Research Ethics Committee (HREC) which approved our ethics application was Mercy Health (HREC ref no. 2023-027, 21 November 2023) and they waived the need for consent.

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Tables

Tables are available in the Supplementary Files section.

Figures

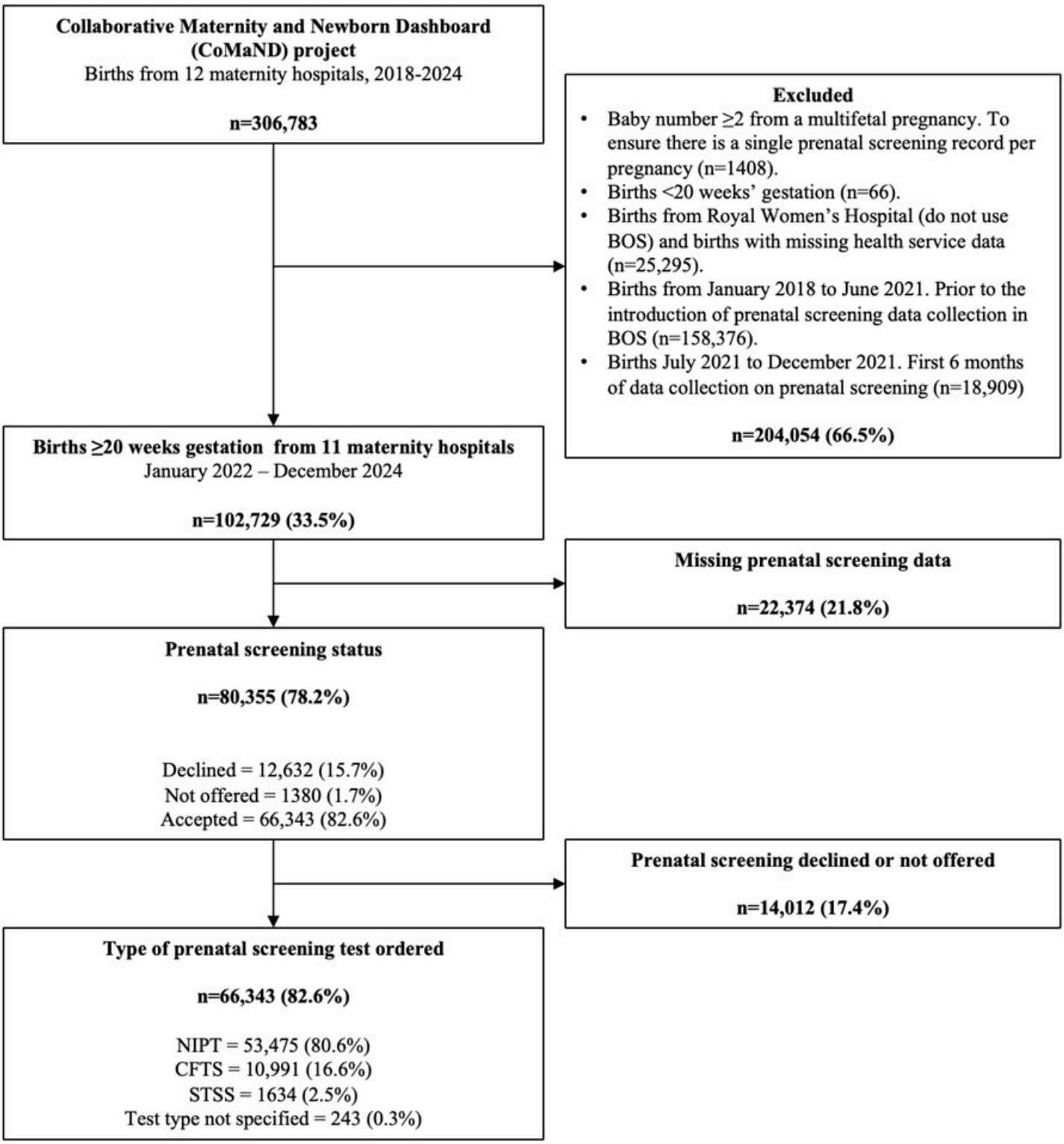


Figure 1

Study Flowchart

CFTS, combined first trimester screening; BOS, birthing outcome system™; STSS, second trimester serum screening; NIPT, non-invasive prenatal testing.

Supplementary Files

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