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Title	Two-decade trends in prenatal genetic testing in Japan
Author(s)	Sasaki, Aiko; Yamada, Takahiro; Sago, Haruhiko et al.
Citation	Journal of Human Genetics, 71, 475-478 https://doi.org/10.1038/s10038-026-01465-y
Issue Date	2026-03-02
Doc URL	https://hdl.handle.net/2115/99116
Rights	This version of the article has been accepted for publication, after peer review (when applicable) and is subject to Springer Nature' s AM terms of use, but is not the Version of Record and does not reflect post-acceptance improvements, or any corrections. The Version of Record is available online at: https://doi.org/10.1038/s10038-026-01465-y
Type	journal article
File Information	manuscript20260205_plain_text.pdf



1 **Two-Decade Trends in Prenatal Genetic Testing in Japan**

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37 **FUNDING**

38 This research was supported by a Health and Labour Sciences Research Grant from the Ministry
39 of Health, Labour and Welfare (project code: 20DA2003) and by a Research Project funded by
40 the Children and Families Agency (project code: 23DA0401).

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42 **CONFLICT OF INTEREST**

43 The authors declare no competing interests.

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ABSTRACT

Prenatal genetic testing in Japan has undergone major changes over the past 2 decades. Maternal serum screening (MSS), amniocentesis (AC), and chorionic villus sampling (CVS) have been the principal tools used to identify fetal chromosomal abnormalities. Recently, noninvasive prenatal testing (NIPT) has reshaped clinical decision-making. However, updated national-level data capturing trends in conventional prenatal testing methods are limited. We aimed to assess the current landscape of prenatal genetic testing in Japan. To update the national data, we compiled test volumes from five major high-volume laboratories that analysed prenatal genetic tests and conducted a supplemental nationwide questionnaire-based survey on ultrasound (US)-only screening between 2020 and 2023. Over this period, MSS declined from 34,887 cases in 2018 to 25,553 in 2023, AC from 16,454 in 2014 to 5,620 in 2023, and CVS from 2,149 in 2015 to 1,167 in 2023, whereas US-only screening increased slightly to >10,000 cases annually. The chromosomal abnormality detection rate among AC cases increased from 8% during 2006–2014 to 20.0% in 2023, with trisomy 21 comprising 43.6% of the cases. During the same period, the proportion of mothers aged ≥ 35 years increased from 14.0% in 2003 to 30.4% in 2023, whereas the proportion of national live births decreased by 35.3%. Thus, declining test volumes cannot be attributed solely to demographic changes but also reflect the influence of NIPT uptake, improved pretest risk stratification, and changing reproductive decision-making supported by expanding genetic counselling. This study provides foundational data for future prenatal genetic

63 testing practices and policies in Japan.

INTRODUCTION

Prenatal genetic testing has undergone significant transformation in Japan over the past 2 decades, reflecting advances in technology and the evolution of clinical practice. Maternal serum screening (MSS), amniocentesis (AC), and chorionic villus sampling (CVS) have been the primary tools used to identify fetal chromosomal abnormalities (CAs). In Japan, non-invasive prenatal testing (NIPT) was first introduced in April 2013 (ref. 1) as a clinical research program; however, this research program was concluded in 2021 (ref. 2). Since then, NIPT has been offered as a self-funded clinical test. It is conducted in accredited facilities following certification by the Japanese Association of Medical Sciences, based on guidance from the Ministry of Health, Labour and Welfare.

In the course of this period, under the Health and Labour Sciences Research Grant project, initiatives were undertaken to establish a comprehensive genetic counselling framework for prenatal genetic testing, including NIPT. These initiatives first involved the development and dissemination of patient-facing informational pamphlets³ for pregnant women and their families, followed by educational activities for obstetricians, role-play-based training programs, and the establishment of a certification system. Beyond the initial clinical introduction of NIPT in 2013, the subsequent development of this counselling framework further reshaped clinical decision-making in prenatal genetic testing by supporting more informed risk assessment and contributing to a sustained decline in the use of invasive diagnostic procedures.

Despite this paradigm shift, updated national-level data capturing trends in conventional prenatal testing methods are limited. Moreover, fetal ultrasound (US)-only screening, frequently used in parallel with or as an alternative to other prenatal genetic testing, has not been adequately represented in national surveys. To address this gap, building on our survey results reported in Japan in 2016 (ref. 4) and incorporating an additional seven years of data, we conducted a comprehensive follow-up survey covering the period from 2003 to 2023. We aimed to assess the current landscape of prenatal genetic testing in Japan, including MSS, AC, CVS, and US-only testing, and quantify changes over time relative to birth rates and demographic trends.

MATERIALS AND METHODS

Data Collection

We collected data from five major laboratories across Japan that analyse prenatal genetic testing. In a previous nationwide survey conducted in 2008 (ref. 4), four of these major laboratories accounted for 96.2% of the market share for MSS, 78.9% for AC, and 84.8% for CVS in Japan. The number of major laboratories included in the present analysis varied over time (four laboratories in 2003–2008, five in 2009–2020, and four in 2021–2023). Given the subsequent centralisation of prenatal genetic testing services over time, it is reasonable to assume that these laboratories continued to cover a comparable or greater proportion of the national testing volume during the study period.

The number of tests performed between January 2003 and December 2023 was obtained for the following modalities:

- MSS, including both the first (combined test) and second trimesters
- AC
- CVS

For AC and CVS, analyses included both G-banding and microarray methods. Although microarray testing performed outside of a formal clinical research framework has been available in Japan since 2012, nationwide data with sufficient reliability were only obtained for the four-year period between 2020 and 2023. Additionally, we collected data on the number of abnormal test results and the number of trisomy 21 cases among these abnormalities. For the US-only screening (including markers such as nuchal translucency, nasal bone, ductus venosus flow, and tricuspid flow), a separate questionnaire-based survey was conducted between 2020 and 2023. The survey targeted certified sonographers whose names appeared on the Fetal Medicine Foundation NT certificate list⁵ and whose affiliated institutions could be identified through internet searches based on their English names. Questionnaires were distributed via post and e-mail, and responses identified only by facility name were aggregated at the facility level to estimate the nationwide number of US-only screenings.

The number of NIPT cases was referenced from the fiscal year survey reports published by the NIPT Consortium for the years 2013–2021 and by the Steering Committee on Certification System for

Prenatal Testing of the Japanese Association of Medical Sciences for the latter half of 2022 (1 July 2022 to 31 March 2023) and for 2023 (1 April 2023 to 31 March 2024) (ref. 6). The number of annual births during the corresponding period was referenced from the Vital Statistics of Japan published by the Ministry of Health, Labour and Welfare⁷.

Ethical Approval

This study was approved by the Ethics Committee of the National Center for Child Health and Development (approval number: 2019-024) and was conducted as part of a Health and Labour Sciences Research Grant (grant number: 20DA2003) and a Research Project funded by the Children and Families Agency, Government of Japan (project code: 23DA0401).

RESULTS

Testing Volume and Temporal Trends

Over the two-decade study period, the introduction of NIPT in 2013 attracted substantial public and clinical interest in prenatal genetic testing, leading to a temporary increase in the volumes of both non-invasive (screening) and invasive (diagnostic) tests. Although the overall number of non-invasive prenatal tests has continued to increase, a marked shift in test composition has occurred, with maternal serum screening being progressively replaced by NIPT. Correspondingly, the number of invasive

136 diagnostic procedures has declined substantially.

137 The total number of non-invasive prenatal tests increased following the clinical introduction of NIPT
138 in 2013, rising from 23,280 in 2012 to 77,174 in 2023 (a 232% increase). In contrast, the total number
139 of invasive diagnostic tests showed a transient increase from 17,500 in 2012 to 18,500 in 2014, likely
140 reflecting heightened public awareness after the introduction of NIPT, but subsequently declined
141 steadily to 6,787 in 2023.

142 Consistent with these national trends, a notable decrease was observed in the number of prenatal tests
143 performed at the five major laboratories in Japan. Based on our survey data and compared with the
144 peak testing volumes during the study period, the number of maternal serum screening (first- and
145 second-trimester) tests declined from 34,887 in 2018 to 25,553 in 2023 (a 26.8% decline). Similarly,
146 the number of amniocentesis (AC) procedures decreased from 16,454 in 2014 to 5,620 in 2023 (a
147 65.8% decline), while the number of chorionic villus sampling (CVS) procedures decreased from
148 2,149 in 2015 to 1,167 in 2023 (a 45.7% decline).

149 In addition, the number of US-only screenings increased moderately from 9,041 cases in 2020 to
150 10,808 in 2023 (19.5% increase). In the 2020–2021 and 2022–2023 surveys, questionnaires were
151 mailed to 86.6% (220/254) and 85.7% (222/259) of certified sonographers listed on the Fetal Medicine
152 Foundation website, respectively, with valid responses obtained from 75.9% (167/220) and 69.8%
153 (155/222). Throughout this period, the number of live births in Japan showed a steady downward trend

(decreasing by 35.3%) from 1,123,610 in 2003 to 727,288 in 2023, notably exceeding the rate of decrease in live births. Among the invasive tests performed in 2023, chromosomal microarray analysis was performed in 14 of 1,167 CVS cases (1.2%) and 103 of 5,620 AC cases (1.8%), indicating the limited but present adoption of advanced genomic testing (Table 1).

CA Detection

The CA detection rate in AC cases remained stable at approximately 8% between 2006 and 2014. However, from 2015 onwards, the CA rate showed a consistent upward trend, reaching 20.0% in 2023. Concomitantly, the proportion of trisomy 21 among the detected CAs also increased from approximately 31–34% during 2006–2014 to 43.6% in 2023 (Figure 1). During a steady decline in the number of live births, Japan has been among the countries with the highest proportion of advanced maternal age pregnancies⁷, with the percentage of mothers aged ≥ 35 years rising markedly from 14.0% in 2003 to 30.4% in 2023, reflecting the ongoing demographic shift towards late childbearing (Table 1).

NIPT in Japan

The number of NIPT cases was derived from fiscal year survey data collected by the NIPT Consortium between 2013 and 2021 and by the Steering Committee on the Certification System for Prenatal

Testing from July 2022 to March 2024 (ref. 6). These data are summarised in Table 1.

Rate of Prenatal Genetic Testing in Japan

Currently, non-diagnostic tests such as NIPT are often performed first, followed by invasive testing based on the results. Although some degree of double counting was expected when summing the numbers across test types, we calculated the overall rate of prenatal genetic testing by aggregating the number of analyses obtained through the above methods and dividing this total by the number of live births for each year. In our 2011 report, the rate of prenatal genetic testing was approximately 3%; however, as of 2023, this rate was estimated to have increased to 11.5% (Table 1).

DISCUSSION

This study provides an updated overview of prenatal genetic testing trends in Japan and documents the marked decline in invasive testing methods over the past 2 decades. These findings build upon our prior nationwide survey from 1998 to 2008, which first highlighted the very low uptake of prenatal genetic testing (approximately 3% of all pregnancies) compared with the high rates in other developed countries and revealed that amniocentesis was the predominant invasive diagnostic procedure at that time⁸. By capturing data from the post-NIPT era, this study offers critical insights into how testing behaviours have evolved with technological advances and demographic changes.

The observed reductions in MSS, AC, and CVS volumes were multifactorial. First, the increasing use of NIPT, which was not the focus of this study but was captured through other national monitoring efforts, has likely led to a decrease in the demand for conventional methods of prenatal genetic testing. This pattern is consistent with reports from Europe⁹ and North America¹⁰, where the introduction of NIPT similarly displaced conventional invasive testing; a meta-analysis indicated that the proportion of women undergoing invasive procedures after a high-chance screening result decreased globally from 75% to 43%, representing an absolute reduction of 38% (ref. 11). Second, demographic shifts, particularly declining birth rates, have broadly influenced test volumes. However, the reductions in AC and CVS cases far exceed the national decline in live births, suggesting additional factors beyond demographics. In Japan, these broad demographic trends are closely linked to societal changes, including the increasing participation of women in the workforce, which has contributed to a steady increase in the maternal age at pregnancy. Advanced maternal age is strongly associated with a high risk of CAs and may influence both the demand for and outcomes of prenatal genetic testing. Simultaneously, Japan's total fertility rate reached a historic low of 1.20 in 2023, reflecting the country's progression towards further population decline and providing an important context for interpreting long-term trends in prenatal genetic testing volumes. In addition to these demographic and societal shifts, increasing awareness of sexual and reproductive health and rights among women and their partners, together with the expansion of genetic counselling

services, has likely contributed to the evolving decision-making processes around prenatal genetic testing in recent years. This trend is consistent with international efforts led by organisations such as the World Health Organization and United Nations Population Fund, which have highlighted the importance of sexual and reproductive health and rights in improving reproductive autonomy and informed choice in maternity care.

One such factor may be improved pretest risk stratification. The CA detection rate among AC cases remained steady at approximately 8% from 2006 to 2014 but began to increase thereafter, reaching 20.0% in 2023. Over the same period, the proportion of trisomy 21 among CA cases also increased from approximately 31–34% in prior years to 43.6% in 2023. These findings suggest a shift in the population undergoing AC, likely towards high-risk pregnancies, and a potentially increased efficiency of prenatal screening strategies before invasive testing. In other words, patients selected for AC in recent years may depict increased CAs, reflecting the impact of NIPT, improved US screening, and refined counselling.

Despite these shifts, the volume of US-only screenings has remained relatively stable at approximately 10,000 cases annually. This suggests that sonographic evaluation continues to play a vital role in prenatal risk assessment, particularly in settings where NIPT is not covered by public insurance or is otherwise inaccessible. Collectively, these findings indicate that trends in prenatal genetic testing in Japan are shaped by technological innovations and broad societal

contexts, offering insights relevant to international discussions on the future of prenatal care.

Limitations

This study was based on data from five major laboratories, which, although representative of national trends, may not capture the full institutional variability across Japan. Furthermore, the rate of prenatal genetic testing was calculated by simply summing the number of nondiagnostic and diagnostic tests, which likely included duplicate cases and may have overestimated the true rate. Additionally, while referring to external data on NIPT volumes, we did not directly assess NIPT-related trends, and the number of NIPT procedures performed at non-accredited facilities remains difficult to ascertain¹², although a considerable number of such tests are widely presumed to be conducted outside the accredited system. This situation represents a significant concern, not only because non-accredited facilities may fail to provide appropriate genetic counselling, but also because it precludes an accurate understanding of the true volume of NIPT testing at the national level.

Japan also has a unique prenatal care system in which approximately 14 fetal US examinations are routinely performed during pregnancy, with the cost generally subsidised by local governments as part of maternity checkup fees rather than as fees for prenatal genetic testing. Consequently, US anomalies may occasionally be incidentally detected¹³ during scans performed

for fetal well-being rather than for detecting chromosomal disorders, and such cases may subsequently lead to prenatal genetic testing. These instances were included in this study if they led to prenatal genetic testing but were excluded from the US-only screening.

CONCLUSION

Ongoing nationwide surveys are warranted to comprehensively assess the prenatal genetic testing landscape. In Japan, prenatal genetic testing has been implemented in the context of a distinctive bioethical paradigm. This study provides foundational data to deepen our understanding of the current situation and guide future directions for practice and policy in this field.

ACKNOWLEDGEMENTS

The authors would like to express their sincere gratitude to the NT-certified sonographers who participated in the questionnaire survey and provided valuable data for this study.

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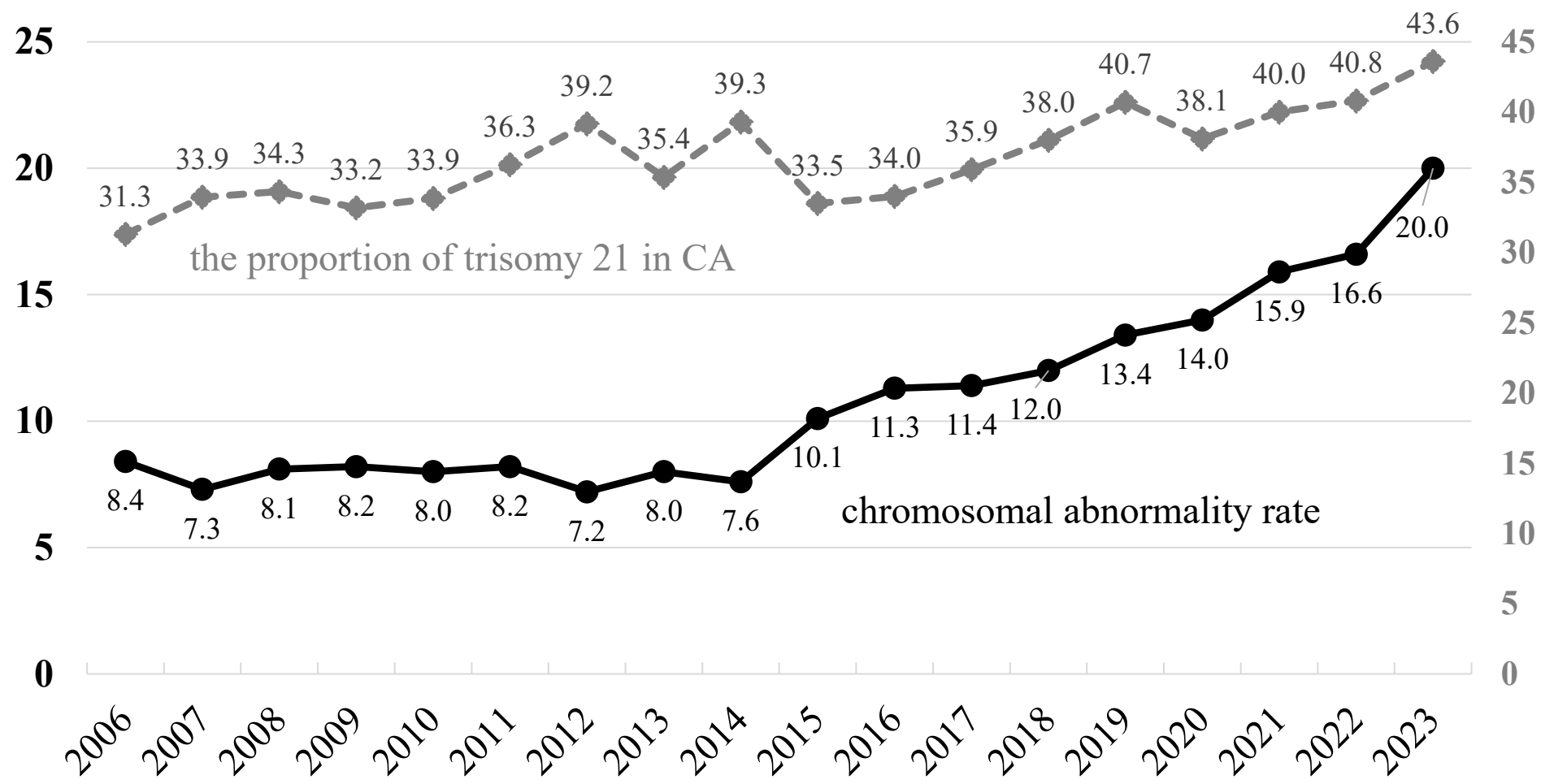
303 **TITLES AND LEGENDS TO FIGURES**

304 Figure 1

305 **Chromosomal abnormality rate and the proportion of trisomy 21 among chromosomal**
306 **abnormalities in amniocentesis cases, 2006–2023.**

307 The solid line represents the proportion of chromosomal abnormalities among amniocentesis
308 cases, whereas the dashed line represents the proportion of trisomy 21 among these abnormalities.

309 CA, chromosomal abnormality



	2003	2004	2005	2006	2007	2008	2009	2010	2011	2012	2013	2014	2015	2016	2017	2018	2019	2020	2021	2022	2023
Birth number	1,123,610	1,110,721	1,062,530	1,092,674	1,089,818	1,091,156	1,070,035	1,071,305	1,050,806	1,037,231	1,029,816	1,003,539	1,005,721	977,242	946,146	918,400	865,239	840,835	811,622	770,759	727,288
Pregnant women aged ≥35 years (rate, %)	14.0	15.3	16.4	17.7	19.4	20.9	22.5	23.8	24.7	25.9	26.9	27.6	28.1	28.5	28.6	28.7	29.1	29.2	30.0	30.0	30.4
AC (microarray)	7,811 (–)	7,877 (–)	8,074 (–)	8,749 (–)	9,723 (–)	10,545 (–)	10,965 (–)	12,023 (–)	12,725 (–)	15,836 (–)	16,336 (–)	16,454 (–)	15,985 (–)	14,808 (–)	12,458 (–)	11,992 (–)	10,767 (–)	9,361 (135)	8,424 (193)	7,273 (157)	5,620 (103)
CVS (microarray)	31 (–)	40 (–)	19 (–)	25 (–)	30 (–)	35 (–)	363 (–)	995 (–)	1,194 (–)	1,664 (–)	1,826 (–)	2,046 (–)	2,149 (–)	1,901 (–)	1,782 (–)	1,744 (–)	1,734 (–)	1,688 (85)	1,540 (74)	1,443 (45)	1,167 (14)
Diagnostic prenatal genetic testing	7,842	7,917	8,093	8,774	9,753	10,580	11,328	13,018	13,919	17,500	18,162	18,500	18,134	16,709	14,240	13,736	12,501	11,049	9,964	8,716	6,787
MSS (1st trimester-)	15,271 (–)	15,332 (–)	15,214 (–)	16,680 (–)	16,525 (–)	17,523 (–)	18,752 (446)	19,896 (1,302)	20,006 (862)	23,280 (1,685)	25,478 (2,575)	28,824 (3,516)	32,424 (4,719)	34,738 (5,315)	33,364 (4,997)	34,877 (5,435)	32,875 (6,242)	36,998 (7,995)	34,010 (8,201)	31,835 (8,128)	25,553 (6,805)
US-only	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	9,041	9,490	10,650	10,808
NIPT	–	–	–	–	–	–	–	–	–	–	7,775	10,531	12,487	13,887	13,497	14,347	14,734	14,406	15,577	20,639	40,813
Non-diagnostic prenatal genetic testing * (with US-only)	15,271 (–)	15,332 (–)	15,214 (–)	16,680 (–)	16,525 (–)	17,523 (–)	18,752 (–)	19,896 (–)	20,006 (–)	23,280 (–)	33,253 (–)	39,355 (–)	44,911 (–)	48,625 (–)	46,861 (–)	49,224 (–)	47,609 (–)	51,404 (60,445)	49,587 (59,077)	52,474 (63,124)	66,366 (77,174)
Total prenatal genetic testing	23,113	23,249	23,307	25,454	26,278	28,103	30,080	32,914	33,925	40,780	51,415	57,855	63,045	65,334	61,101	62,960	60,110	71,494	69,041	71,840	83,961
Prenatal genetic testing rate (%)	2.06	2.09	2.19	2.33	2.41	2.58	2.81	3.07	3.23	3.93	4.99	5.77	6.27	6.69	6.46	6.86	6.95	8.50	8.51	9.33	11.54

Annual trends in prenatal genetic testing in Japan, 2003–2023.

The table summarizes annual numbers of births, the proportion of births to mothers aged 35 years or older, and the nationwide volumes and uptake rates of prenatal testing modalities, including MSS, AC, CVS, ultrasound-only screening, and NIPT.

AC: Amniocentesis, CVS: Chorionic villus sampling, MSS: Maternal serum screening, US-only: Ultrasound-only screening, NIPT: Noninvasive prenatal testing, – : No data,

*Non-diagnostic genetic testing: MSS+NIPT