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Original Article

Knowledge and Attitudes toward Non-invasive Prenatal Testing among Pregnant Japanese Women

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To assess the knowledge and attitudes of pregnant Japanese women regarding non-invasive prenatal testing (NIPT). Between March and June 2013, 557 pregnant women in the Hyogo and Hiroshima Prefectures were surveyed using an anonymous, self-completed questionnaire. A total of 91.9% respondents (512/557) stated that they "agree" or "conditionally agree" with NIPT implementation in Japan. Approximately 28.5% of respondents stated that they knew that the accuracy of a positive NIPT result can be affected by mother's age and background, while 34.5% of respondents stated that it was necessary for pregnant women with a positive result to undergo fetal chromosome diagnosis using amniocentesis; both percentages were low. Additionally, 92.3% of respondents would "like a detailed explanation of the test," 65.1% of them would "like psychological support if the NIPT test results came back positive," and 5.7% would terminate the pregnancy if the NIPT test results came back positive without undergoing fetal chromosome diagnosis via amniocentesis. Although a high proportion of pregnant Japanese women agreed with the introduction of NIPT into Japanese obstetrical care, there was insufficient knowledge regarding the test. It is necessary for women undergoing NIPT to be provided sufficient information and psychological support.

Key words: amniocentesis, Down syndrome, fetal chromosome diagnosis, non-invasive prenatal testing, psychological support

The technology of screening for diagnosing congenital diseases and abnormalities in fetuses has rapidly developed in the past few decades. In many western countries, prenatal screening for Down syndrome is offered to pregnant women [1].

The possibility of diagnosing aneuploidies with a maternal blood test emerged with the discovery of cell-free fetal DNA in 1997 [2]. Non-invasive prenatal testing (NIPT) for Down syndrome was first implemented clinically in the USA and China/Hong Kong [3]. NIPT involves comprehensive sequencing of cell-free fetal DNA from maternal plasma [4–8], and enables the detection of fetal chromosomal abnormalities (*e.g.*, trisomy 21, 18, 13) as early as the 10th week of pregnancy.

Using NIPT to screen fetuses for trisomy 21 provides 98.6% sensitivity, with a maximum false-positive rate of 0.8% [5, 9–11]. Similar numerical values were reported in Japanese newspapers in 2012. These results, however, were achieved in a study targeting high-risk groups [4, 6]. However, it is

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believed that only a small proportion of pregnant women are aware of the details of NIPT, *e.g.*, the fact that the sensitivity and the rate of false-positives can vary depending on the background of the pregnant women [4, 5].

Furthermore, similar to screening of maternal serum and nuchal translucency (NT) thickness, NIPT does not provide a definitive diagnosis; if the test result is positive, fetal chromosomal diagnosis using amniocentesis must be undertaken to determine the diagnosis. One concern is that pregnant women who are not aware of this two-step procedure of diagnosis may choose to abort the fetus without further genetic amniocentesis if they receive a positive result [4].

In Japan, for prenatal testing of fetal chromosomal abnormalities, maternal serum screening (α-fetoprotein; AFP, human chorionic gonadotropin; hCG, unconjugated estriol; uE3, Inhibin A), and NT measurement have been conventionally performed. Fetal chromosomal abnormalities have been diagnosed by amniocentesis, whereas chorionic villus sampling (CVS) is hardly ever performed in Japan.

In April 2013, NIPT started being implemented in Japan [12]. Professional organizations such as the Japan Society of Obstetrics and Gynecology recommended NIPT for women who meet the following conditions: (i) the fetal ultrasound suggests the possibility of a fetal chromosomal abnormality; (ii) maternal serum screening suggests the possibility of a fetal chromosomal abnormality; (iii) a previous pregnancy had a fetal chromosomal abnormality; (iv) pregnancy occurs at an older age (≥ 35), or (v) trisomies 21 and 13 in the fetus are considered possible due to a balanced Robertsonian translocation in either parent [Guidelines for NIPT in Japan: Japan Society of Obstetrics and Gynecology. URL http://www.jsog.or.jp/news/pdf/guidelineForNIPT_20130309.pdf#search='NIPT+%E6%8C%87%E9%87%9D' (in Japanese) [accessed on 14 April 2014]].

Previous studies on the attitudes of pregnant women to maternal serum screening, NT measurement and fetal chromosome diagnosis by amniocentesis have clarified that they have insufficient knowledge and information about prenatal diagnoses to make informed decisions [13–16], and that providing this knowledge reduces the anxiety of pregnant women [17]. However, there are no studies in pregnant Japanese women on their knowledge of NIPT, or their attitudes toward undergoing NIPT.

In the present study, we investigated knowledge and attitudes toward NIPT among pregnant Japanese women in Japan.

Methods

Between March and June 2013, 760 pregnant women undergoing pregnancy health checks at one of 5 general maternity clinics or hospitals in Hyogo and Hiroshima Prefectures in Japan were asked to complete an anonymous questionnaire after informed consent. Participants filled out their age, educational background, previous pregnancies, and history of infertility treatment, as well as responding to other questions about their background. The survey addressed their knowledge of NIPT, along with their attitudes as whyone would undertake the test. Women with known health problems or known fetal complications were excluded from the subjects. Questionnaire responses were obtained from 591 pregnant women. Data from 557 respondents were eventually analyzed.

Data were analyzed using SPSS for Windows [ver.20] (IBM Inc., Chicago, IL, USA), using descriptive statistics, the chi-squared test and the *t* test. Statistical significance was set at $p < 0.05$. Results are presented as mean ± standard deviation.

This study was conducted with the approval of the Research Ethics Committee of Graduate School of Health Sciences, Okayama University of Japan.

Results

Background of respondents. Questionnaire responses were obtained from 591 pregnant women (response rate 77.8%); responses of 34 were considered invalid, such that data from 557 respondents were eventually analyzed (valid response rate 73.3%). The mean gestational period of the respondents was 26.1 ± 8.4 [5–40] weeks, and the mean age was 32.1 ± 4.6 [18–44] years (Table 1). In all, 49.2% respondents were primigravida, 16.9% had undergone infertility treatment, 23.0% had experienced a miscarriage, and 13.1% had experienced medical termination of a pregnancy. The percentages of women aged ≥ 35 years who were primigravida, had received infertility treatment, or had experienced a miscarriage were significantly higher than those of women

aged ≤ 34 years. These rates were almost the same as those of the general Japanese population.

Knowledge of NIPT. In total, 76.5% respondents answered that they either "knew well" or "had heard of" NIPT (Table 2). Women aged ≥ 35 years had a higher level of knowledge regarding all types of prenatal testing than those aged ≤ 34 years. Nevertheless, a high overall level of knowledge was observed regarding the fact that "the test can screen whether or not the fetus has Down syndrome" (74.5%). However, not many women seemed to be aware of the facts that "At present, the anticipated conditions include only three chromosomal abnormalities (trisomy 21, 18, and 13)" (16.7%). Furthermore, there were low rates of women who knew that "if a positive test result is returned, it is necessary to undergo fetal chromosome diagnosis using amniocentesis, to diagnose whether or not there is actually an abnormality with the fetus" (34.5%), and "the accuracy of a positive test result can be affected by the age and background of the

Table 1 Background of respondents

	Total (n = 557)	≤ 34 years of age (n = 383)	≥ 35 years of age (n = 174)	*p* value
Gestational age in weeks	26.1 ± 8.4[5–40]	26.0 ± 8.3[8–40]	26.4 ± 8.8[5–40]	0.674
Age (years)	32.1 ± 4.6[18–44]	29.8 ± 3.3[18–34]	37.3 ± 2.1[35–44]	
Parity				
Primiparous	274 (49.2%)	200 (52.2%)	74 (42.5%)	<0.05
Multiparous	283 (50.8%)	183 (47.8%)	100 (57.5%)	
Experience with infertility treatment	94 (16.9%)	38 (9.9%)	56 (32.2%)	<0.001
Experience with miscarriage	128 (23.0%)	69 (18.0%)	59 (33.9%)	<0.001
Experience with artificial termination	73 (13.1%)	54 (14.1%)	19 (10.9%)	0.303
Experience with stillbirth	10 (1.8%)	8 (2.1%)	2 (1.1%)	0.349

Table 2 Knowledge of NIPT in pregnant women

	Total (n = 557)	≤34 years of age (n = 383)	≥35 years of age (n = 174)	*p* value
Have you heard of NIPT?				
Know it well	35 (6.3%)	21 (5.5%)	14 (8.0%)	
Have heard of it	391 (70.2%)	257 (67.1%)	134 (77.0%)	0.676
Do not know it	131 (23.5%)	105 (27.4%)	26 (14.9%)	
Test is carried out by taking blood from the mother	417 (74.9%)	271 (70.8%)	146 (83.9%)	<0.01
Used to screen for Down syndrome in fetus	415 (74.5%)	279 (72.8%)	136 (78.2%)	0.182
If a positive test result is returned from the NIPT, it is necessary to undergo fetal chromosome diagnosis using amniocentesis, to diagnose whether or not there is actually an abnormality in the fetus	192 (34.5%)	118 (30.8%)	74 (42.5%)	<0.01
The accuracy of a positive test result can be affected by the age and background of the mother	159 (28.5%)	105 (27.4%)	54 (31.0%)	0.381
The test can be carried out from the 10th week of pregnancy	113 (20.3%)	63 (16.4%)	50 (28.7%)	<0.01
If the test result is negative, the probability that the fetus has no chromosomal abnormality is almost 100%, regardless of age or background	112 (20.1%)	62 (16.2%)	50 (28.7%)	<0.01
At present, the anticipated condition include only three chromosomal abnormalities (trisomy 21, 18, and 13)	93 (16.7%)	45 (11.7%)	48 (27.6%)	<0.001
Test result accuracy is 95% in pregnancies among older women, and those indicated in ultrasound testing as having the possibility of a fetal abnormality those had children	70 (12.6%)	47 (12.3%)	23 (13.2%)	0.755
Test result accuracy is 75% if all pregnancies in women over the age of 35 are tested, and 30% if all pregnancies are tested	36 (6.4%)	28 (7.3%)	8 (4.6%)	0.227

mother" (28.5%).

In 5 of the 9 categories related to knowledge of NIPT, a greater proportion of pregnant women aged ≥ 35 years responded that they knewthe facts than women aged ≤ 34 years.

Attitude to NIPT

1. Indication and facilities to provide NIPT

When asked whether NIPT should be implemented in Japan, 41.8% women responded that they "agreed" with the implementation and 50.1% "agreed subject to conditions." Only 4.1% were "opposed" to its implementation (Table 3).

When asked who should be the subject of NIPT, a high proportion of women (69.1%) felt that "NIPT should be offered to all pregnant women who want it." Some (6.3%) responded that they believed it "should be used as a mass screening program for all pregnant women." Only 19.2% of women stated that NIPT should be "limited to pregnant women who have a high risk of carrying a fetus with a chromosomal abnormality" and 5.4% believed that it should be given to "all pregnant women at or above a certain age"; both of those are currently the recommended subjects according to the Japanese NIPT guidelines. A significantly high number of women who responded that NIPT should be implemented "as a mass screening program for all pregnant women" were ≤ 34 years of age (8.1%).

When asked where they would like to undergo NIPT, 41.5% (231/557) responded "at a University hospital," 21.5% (120/557) responded "at a perinatal maternal and child medical center," and 67.0% (373/557) responded "at a general obstetric hospital/clinic." A significantly high proportion of women aged ≥ 35 years (28.2% vs. 18.5%) gave the response "at a perinatal maternal and child medical center."

2. Support prior to NIPT

Most women (92.3%) responded that they would "want a detailed explanation related to the test" when deciding whether or not to undergo it (Table 4). Of them, 63.7% would want an explanation from an "obstetrician specializing in genetic medicine." In contrast to this, the proportion that responded from an "accredited genetic counselor" and a "genetic specialist nurse" was low (17.4% and 10.2%, respectively). Among women aged ≥ 35 years, 28.7% responded that they would want an explanation from an "accredited genetic counselor", which was significantly higher than those aged ≤ 34 years (12.3%).

Furthermore, when undergoing NIPT, 69.8% of respondents would "like psychological counseling" and 63.8% would prefer receiving it from a psychological counselor.

3. Support for women with a positive NIPT result

If NIPT results are positive, 84.9% women responded that they would "want support" (Table 5). Of these, 65.1% responded that they would "want psychological support." When asked from whom they would like to hear an explanation, a high proportion (82.8%) responded they would prefer "a pediatrician knowledgeable about Down syndrome and other conditions." In addition, a relatively high proportion (41.4%) of these women stated that they would like to "hear from the mothers of children with Down syndrome."

Table 3 Conditions for implementing NIPT

	Total (n = 557)	≤34 years of age (n = 383)	≥35 years of age (n = 174)	*p* value
Implementation of NIPT in Japan				
Agree	233 (41.8%)	162 (42.3%)	71 (40.8%)	0.676
Agree subject to certain conditions	279 (50.1%)	190 (49.6%)	89 (51.1%)	
Oppose	23 (4.1%)	14 (3.7%)	9 (5.2%)	
No answer	22 (3.9%)	17 (4.4%)	5 (2.9%)	
Conditions for implementing NIPT in Japan				
Focus of NIPT implementation				
All pregnancies	35 (6.3%)	31 (8.1%)	4 (2.3%)	<0.05
All pregnant women wishing to undergo NIPT	385 (69.1%)	265 (69.2%)	120 (69.0%)	
Limited to pregnant women with a high possibility of chromosomal abnormality	107 (19.2%)	69 (18.0%)	38 (21.8%)	
Limited only to pregnant women over a certain age	30 (5.4%)	18 (4.7%)	12 (6.9%)	

Table 4 Support prior to NIPT implementation

	Total (n = 557)	≤34 years of age (n = 383)	≥35 years of age (n = 174)	*p* value
Would like detailed explanation when deciding whether or not to undergo NIPT	514 (92.3%)	350 (91.4%)	164 (94.3%)	0.240
Preferred source of explanation				
Obstetrician specializing in genetic medicine	355 (63.7%)	237 (61.9%)	118 (67.8%)	0.333
Obstetrician currently caring for pregnancy	343 (61.6%)	235 (61.4%)	108 (62.1%)	0.772
Pediatric genetic specialist	121 (21.7%)	84 (21.9%)	37 (21.3%)	0.720
Accredited genetic counselor	97 (17.4%)	47 (12.3%)	50 (28.7%)	<0.001
Genetics specialist nurse	57 (10.2%)	38 (9.9%)	19 (10.9%)	0.806
Obstetric nurse	38 (6.8%)	26 (6.8%)	12 (6.9%)	0.964
Nurse	16 (2.9%)	12 (3.1%)	4 (2.3%)	0.547
Clinical psychologist	14 (2.5%)	8 (2.1%)	6 (3.4%)	0.268
Would like psychological counseling when undergoing NIPT	389 (69.8%)	263 (68.7%)	126 (72.4%)	0.372
Preferred source of psychological counseling				
Psychological counselor	248 (63.8%)	167 (63.5%)	81 (64.3%)	0.880
Obstetrician	167 (42.9%)	109 (41.4%)	58 (46.0%)	0.051
Clinical psychologist	146 (37.5%)	90 (34.2%)	56 (44.4%)	0.392
Obstetric nurse	98 (25.2%)	70 (26.6%)	28 (22.2%)	0.882
Nurse	48 (12.3%)	32 (12.2%)	16 (12.7%)	0.356
Other	8 (2.1%)	6 (2.3%)	2 (1.6%)	

Table 5 Support during fetal chromosome diagnosis using amniocentesis subsequent to undergoing NIPT

	Total (n = 557)	≤34 years of age (n = 383)	≥35 years of age (n = 174)	*p* value
Would want support if NIPT test result is positive	473 (84.9%)	322 (84.1%)	151 (86.8%)	0.408
Preferred means of support				
Psychological support	308 (65.1%)	202 (62.7%)	106 (70.2%)	0.112
Information provision				
Explanation from pediatrician knowledgeable about Down syndrome and other conditions	392 (82.8%)	270 (83.9%)	122 (80.8%)	0.411
Want to meet and talk with mothers of children with Down syndrome	196 (41.4%)	149 (43.5%)	56 (37.1%)	0.188
Meet children with Down syndrome	78 (16.5%)	52 (16.1%)	26 (17.2%)	0.770
Interview with a group or organization of people with experience regarding Down syndrome	46 (9.7%)	26 (8.1%)	20 (13.2%)	0.077
Interview with a group or organization of people with experience on such matters, in cases where prenatal testing establishes that the fetus has Down syndrome				
Strongly want	25 (4.5%)	19 (5.0%)	6 (3.4%)	0.848
Want	326 (58.5%)	222 (58.0%)	104 (59.8%)	
Do not want	177 (31.8%)	121 (31.6%)	56 (32.2%)	
Absolutely do not want	21 (3.8%)	16 (4.2%)	5 (2.9%)	
No response	8 (1.4%)	5 (1.3%)	3 (1.4%)	

Furthermore, 58.5% of respondents stated that if it was prenatally determined that they were carrying a child with Down syndrome, they would "want to meet and talk with a group or organization with experience

dealing with such matters."

4. Attitude to fetal chromosome diagnosis using amniocentesis in the case of a positive NIPT result

If a positive NIPT result was returned, 74.0% of women responded that they would "undergo fetal chromosome diagnosis using amniocentesis" (Table 6). A further 20.3% responded that they would "continue the pregnancy without undergoing fetal chromosome diagnosis using amniocentesis," while 5.7% responded that they would "terminate the pregnancy without undergoing fetal chromosome diagnosis using amniocentesis."

When asked for the reasons why they would continue the pregnancy without undergoing fetal chromosome diagnosis using amniocentesis, 66.4% (75/113) responded that they would "want to raise the child even if it had an abnormality," while 49.6% (56/113) responded that "fetal chromosome diagnosis using amniocentesis carries a risk of miscarriage."

When the subset who would terminate the pregnancy without undergoing fetal chromosome diagnosis using amniocentesis were asked the reasons for this choice 59.4% (19/32) responded that they would "terminate the pregnancy if there was even a small chance of abnormality," while 40.6% (13/32) felt that it would be a "pity for the fetus to have to terminate it at a later date" when further tests conclusively show abnormality. And 25.0% of women (8/32) responded that "the anxiety is too great to wait several more weeks" for further testing to be completed.

Looking at the background of the 5.7% of pregnant women who would "terminate the pregnancy without undergoing fetal chromosome diagnosis using amniocentesis," 62.5% (20/32) "agree" that NIPT should be performed in Japan, while 56.3% (18/32) responded that they would "not want any support" prior to NIPT implementation if they received a positive result from NIPT, and 53.1% (17/32) responded that they would "not want psychological counseling" when undergoing NIPT.

If the results of NIPT were positive, a lower proportion of women aged ≥ 35 years responded that they would "undergo fetal chromosome diagnosis using amniocentesis" (65.5%) (Table 6). In addition, a high proportion of women aged ≥ 35 years responded that they would "terminate the pregnancy without undergoing fetal chromosome diagnosis using amniocentesis" (8.6%, 15/174).

Parity and attitude to NIPT among women aged ≥ 35 years

Among women aged ≥ 35 years, there were no significant differences between primiparous and multiparous women in the results for "Knowledge of NIPT", "Conditions for implementing NIPT", "Support prior to NIPT implementation", "Support during fetal chromosome diagnosis using amniocentesis subsequent to undergoing NIPT", and "Attitude to fetal chromosome diagnosis using amniocentesis subsequent to a positive NIPT result."

Discussion

Knowledge of NIPT. Japanese newspapers reported extensively on NIPT in 2012. It was reported that NIPT can be implemented simply by collecting a maternal blood sample, and that at least 99% of all pregnancies with Down syndrome can be detected. When NIPT was implemented in Japan in 2013, aspects that were relatively well known by pregnant Japanese women were the facts that "blood is collected from the pregnant woman and tested" and that "the test can screen whether or not the fetus has Down

Table 6 Awareness of fetal chromosome diagnosis using amniocentesis subsequent to a positive NIPT result

	Total (n = 557)	≤34 years of age (n = 383)	≥35 years of age (n = 174)	*p* value
Would you undergo fetal chromosome diagnosis using amniocentesis subsequent to a positive NIPT result?				
Would undergo fetal chromosome diagnosis using amniocentesis	412 (74.0%)	298 (77.8%)	114 (65.5%)	<0.01
Would continue pregnancy without undergoing fetal chromosome diagnosis using amniocentesis	113 (20.3%)	68 (17.8%)	45 (25.9%)	
Would terminate the pregnancy without undergoing fetal chromosome diagnosis	32 (5.7%)	17 (4.4%)	15 (8.6%)	

syndrome." Only a few pregnant women were aware that "NIPT positive predictive value can vary depending on the subject tested," and that "if a positive test result is returned in NIPT, it is necessary to undergo fetal chromosome diagnosis using amniocentesis to establish a definitive diagnosis." For these reasons, some women may have opted for medical termination of a pregnancy merely because they have received a positive NIPT result. Thus, this information must be provided to a pregnant woman before she undergoes NIPT or even before she becomes pregnant.

Attitude of the implementation of NIPT. Our study results demonstrate that many pregnant women approve of NIPT implementation in Japan, and that they would like to see a relaxation in the current restrictions [18]. However, a previous study revealed that people were more positive about NIPT's clinical applications the less they knew about it [19]. In our study, $<10\%$ of pregnant women responded that they "knew a lot" about NIPT, and $<30\%$ knew anything about its predictive values. This disparity in knowledge and the fact that 90% of women approved of NIPT implementation indicate that women are giving their approval without having sufficient knowledge about it. Although only a few facilities offer the test in Japan, our study results reveal that women prefer the general obstetric hospital or clinic where they are receiving pregnancy health care rather than a doctor they meet for the first time at a more distant hospital. When the facilities providing NIPT in Japan increase in number, a thoughtful discussion on the institutional conditions for implementing NIPT will be necessary.

Support when implementing NIPT. In this study, 90% of respondents hoped for a detailed explanation when deciding whether to undergo NIPT. At present, the Japanese guidelines recommend undergoing NIPT only after receiving genetic counseling from an obstetrician and pediatric genetic specialist or an approved genetic counselor. Consistent with this, we found that a high proportion of women (around 60%) hoped for an explanation from an obstetrician, particularly at the hospital they were attending regularly. This may be because women feel more comfortable taking advice from familiar medical staff. When implementing prenatal diagnoses in order to provide a pregnant woman with sufficient information and reduce anxiety, doctors and midwives who have been trained in the area must counsel the women [13, 20–23]. However, the numbers of obstetric genetic specialists, pediatric genetic specialists, and approved genetic counselors in Japan is insufficient [24]. Training of not only obstetricians but also midwives and nurses should be required to give informed advice in order to implement NIPT testing at local obstetric clinics.

Support for positive results in NIPT. The results of our study indicate that many women will require support (*e.g.*, medical and psychological) if they receive a positive test result from the NIPT. Approximately 70% women in our survey wanted to receive a "detailed explanation from a pediatrician knowledgeable about Down syndrome and other associated complications." Pediatricians not only provide medical support but also offer the necessary psychological and social support for both the affected infant and his/her family and implement long-term follow-up for children with congenital abnormalities [25]. Thus the access of women receiving a positive NIPT result to a pediatrician, who can take a neutral position and provide medical advice and psychological support, may assist them in reaching an appropriate decision. This, in turn, will significantly impact the subsequent decisions. Furthermore, around 30% of pregnant women also indicated a desire to hear from other mothers of children with Down syndrome. At present in Japan, few medical staff are proactive in this area and there is no system in place to promote this kind of informational exchange. Prior to undergoing NIPT, it would be preferable for pregnant women or for people who may have children in the future to be given an opportunity to learn specifically about the lifestyles of children and adults with chromosomal abnormalities such as Down syndrome.

Definitive diagnosis if the NIPT result is positive. Undergoing prenatal diagnosis can cause a lot of conflict and anxiety in women [26, 27]. Mental preparation is thus necessary before undergoing NIPT. NIPT can be implemented simply by drawing blood from the mother, and in certain subjects provides a high degree of accuracy [4–8]. The results of NIPT indicate the probability that varies depending on the age of the mother and individual risk of chromosomal abnormality [4, 6]. In other words, NIPT is a screening test, and when the results are positive, amniocentesis must be undertaken to establish a definitive diagnosis. However, only 70% of

women in our study responded that they would undergo amniocentesis if their NIPT result was positive, and 5.7% responded that they would opt for medical termination of pregnancy without undergoing amniocentesis. In previous research into the respondents who would have NIPT, 30.7% said that they were 'likely' to terminate an affected pregnancy [28]. In the present study, more than half of these 5.7% women responded that if they received a positive NIPT result, they would "not want any support," and that when undergoing NIPT they would "not want psychological counseling"; this further indicates the high possibility that these women would opt for medical termination of the pregnancy based solely on their own interpretation of the NIPT result.

Some women may decide not to undergo NIPT or to continue the pregnancy even if they receive a positive NIPT result without undergoing amniocentesis. In such women, NIPT may be suggested for deciding the delivery method or preparing for the requisite post-natal care; alternatively, it may be appropriate to select fetal chromosome diagnosis using amniocentesis right from the start. It is necessary that people have knowledge regarding fetal chromosomal abnormalities prior to pregnancy, for example, during school education, and that opportunities be created for them to think about these medical, social and ethical issues. A system is required whereby pregnant women should not be allowed to undergo the test without sufficient knowledge of the birth implications of NIPT test results, or of the conflicts and anxieties that can occur when undergoing NIPT.

Comparison by age. Compared with women aged ≤ 34 years, a greater proportion of pregnant women aged ≥ 35 years were knowledgeable about NIPT. This is probably because women on reaching the age of ≥ 35 years themselves become aware of the fact that they are more prone to giving birth to children with chromosomal abnormalities and therefore look for information on the subject. There was no significant difference by age group in attitude to NIPT implementation, support prior to or subsequent to undergoing the test, or the implementation of fetal chromosome diagnosis using amniocentesis. Thus, it will be necessary to ensure that sufficient explanation and support prior to or subsequent to the test is given to all women seeking NIPT regardless of their age.

In conclusion, the present study results revealed that pregnant Japanese women have a low level of knowledge regarding NIPT, and that in some cases, their response to the test results may contradict their real intentions. Approximately 90% of the pregnant women surveyed agreed with its implementation in Japan and wished that the test was more easily available than under the current system. This may, however, result from their lack of knowledge and inappropriate information. Furthermore, some women may undergo NIPT without adequate support or information and then be subjected to distress and conflict by the results, and may opt for medical termination of pregnancy without definitive diagnosis testing. A system is required that encompasses the need for psychological support for pregnant women both when deciding on whether to undergo NIPT and when the NIPT result is positive; such a program would allow women to consider what the life of a child with Down syndrome or other chromosomal abnormality may be like. Public awareness of people with Down syndrome or other chromosomal abnormalities could also lead to their improved welfare in society.

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