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Receiving the news of Down syndrome in the era of prenatal testing

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Abstract

Objective: To explore the prenatal trajectory and the experiences of mothers of a child with Down syndrome (DS) at the time of receiving information or test results when participating in a nationwide prenatal screening program.

Methods: An online questionnaire study was completed by mothers of children with DS born between January 1, 2010 and February 28, 2016 (n = 212). Data were collected between February 15 and 28, 2016.

Results: Most of the live born children with DS were diagnosed postnatally. The majority of their mothers had explicitly chosen not to have prenatal DS screening. Of the 39 mothers prenatally informed their child might have DS, only 49% were completely or mostly satisfied about the information provided by their clinical providers at that time. About 16% of women (of the 38 that answered this question) recall some perceived emphasis on the option of terminating pregnancy as the first choice. Mothers who had received a postnatal diagnosis rated the experience as more positive than their counterparts who received prenatal diagnoses.

Conclusion: With recent developments in screening, more parents are expected to receive a DS diagnosis before birth. Meeting the parents' individual counseling needs at the time of prenatal diagnosis requires careful exploration of their personal values and preferences.

KEYWORDS

Down syndrome, information-provision, patient-experiences, prenatal testing

1 | INTRODUCTION

Globally, prenatal testing for fetal aneuploidy has increasingly shifted from invasive diagnostic testing for maternal age or for an a priori elevated risk, toward prenatal screening for all expectant parents. This has resulted in a wider range of options for expectant parents.

Until 2007, in the Netherlands, there was no formal prenatal screening program for Down syndrome (DS), but serum screening and first-trimester combined test (ftCT) were widely offered. Beginning in 2007, the ftCT and second-trimester fetal anomaly scan became available for all pregnant women as part of a national screening program. The guidelines stated that all pregnant women should be asked whether or not they wanted information on DS screening. This information was then to be provided by the prenatal care provider. Invasive testing was offered to women with prenatal screening results <1:200 for DS. Until January 2015, ftCT was fully covered by the insurance, for women 36 years and older; younger women paid an additional fee of €165 (Health Council of the Netherlands, 2016a). Since January 2015, an additional fee has been charged to all women pursuing ftCT. As of April 2014, cell-free DNA based non-invasive prenatal screening (called NIPS further in the article) was available as a second-tier test for women with a positive ftCT result (or elevated

risk result). Since April 2017, NIPS has been available as a first-tier test for all women. Under this arrangement, pregnant women in the Netherlands now have a choice to forgo testing, have ftCT at current costs of €170, or NIPS at €175. Regardless, all pregnant women are offered a mid-pregnancy fetal ultrasound at no extra costs. Termination of pregnancy (TOP) is legally allowed under a certain number of conditions up to 24 weeks of pregnancy. Unlike in the USA, offering children with DS up for adoption is very rarely proposed or utilized in the Netherlands.

In 2016, the uptake of ftCT was 34.1%, a relatively low number when compared to other European countries (Harmsen, Liefers, Cruijsberg, & Atsma, 2017). For example, in Denmark, France, Belgium, and Iceland, uptake rates were >80% (Blondel, Lelong, Kermarrec, & Goffinet, 2012; Ekelund et al., 2011; Gottfredsdóttir, Björnsdóttir, & Sandall, 2009; Neyt, Hulstaert, & Gyselaers, 2014), while Finland and England had an uptake of nearly 75% (Ingvoldstad, Öhman, & Lindgren, 2014; Zehrer, Stankeviciene, & Abdel-Aal, 2015). Policy and service delivery are assumed to affect uptake rates, as well as the contextualization of "the offer." In the Netherlands, the additional fee, the strong emphasize on "right not to know," as well as the public debate preceding the introduction of ftCT, may play a role in the relatively low uptake (Crombag, 2016; Crombag et al., 2014). With the introduction of NIPS, uptake in the Netherlands has slightly increased to 42% (NIPT Consortium the Netherlands, 2018).

Even though the number of parents receiving a diagnosis during pregnancy has increased, a significant number of parents still receive a diagnosis of DS after birth (de Groot-van der Mooren, Gemke, Cornel, & Weijerman, 2014). Research from different countries throughout Europe and the USA has reported on dissatisfaction of mothers receiving such a postnatal diagnosis (Skotko, 2005a). To deliver the news, both national and international guidelines are available (Borstlap et al., 2011; Sheets et al., 2011;Skotko, Capone, & Kishnani, 2009; Skotko, Kishnani, & Capone, 2009), but Dutch pediatricians state that not all recommendations are implemented (de Groot-van der Mooren et al., 2014). Research has found that the news should be delivered in a way that meets the parents' preferences (Skotko, Capone, & Kishnani, 2009; Skotko, Kishnani, & Capone, 2009).

Researchers have studied parental decision-making processes (Aune & Möller, 2012; Gitsels-van der Wal et al., 2015; Martin et al., 2013; van Schendel et al., 2016) and their experiences with receiving information on ultrasound findings or other confirmed diagnoses (Asplin, Wessel, Marions, & Öhman, 2013; Hunt, France, Ziebland, Field, & Wyke, 2009; Lotto, Smith, & Armstrong, 2018; Sommerseth & Sundby, 2010), both before and after birth. No research, to date, has explored the experiences of parents of children with DS when participating in a nationwide prenatal screening program (Lou et al., 2018). The purpose of this research, was to survey mothers of children with DS who were informed about and eventually participated in a national prenatal screening program and use their experience to inform improvements in the way care is provided to future (expectant) parents.

2 | METHODS

2.1 | Participants and procedures

Participants were recruited through the e-newsletter, magazine, Facebook page, and website of the Dutch Down Syndrome Foundation (SDS) and invited to complete an online questionnaire which was accessible from February 15, 2016 to February 28, 2016. SDS was founded in 1988 and is a non-profit DS organization with approximately 3,300 members of which 2,100 are families with a child with DS, as of 2016. Of these, 471 families had a child with DS born between 2010 and 2015. In addition, during this same time, there were 245 families who contacted the SDS, but were not official members as of February 28, 2016. Taken together, these 716 families represent about 60% of the total number of families who have a child with DS in this age range in the Netherlands, as estimated by the method of de Graaf et al (de Graaf et al., 2011). Of the 4,000 readers of the SDS e-newsletter, an estimated 80% (3,200) families have a son/daughter with DS. Not all of these 3,200 families are members of the SDS; around 1,100 are non-members, who read the e-newsletter without having registered as a paying member of the SDS.

In the period 2010–2015, a total of 1,267 children with DS were live born in the Netherlands (de Graaf et al., 2017). Using de Graaf et al. to model survival in the first years of life, approximately 1,197 of these children were still alive in 2016 (de Graaf et al., 2011).

The questionnaire took, on average, 10–20 min to complete. Inclusion criteria were limited to biological parents who had a child with DS born between January 1, 2010 and February 28, 2016. In our analysis, we excluded fathers (n = 30) for three reasons: (a) their number was very limited, (b) their responses very much aligned with the responses of the mothers, and (c) we wanted to avoid double counting if more than one parent responded per family.

2.2 | Editorial policies and ethical considerations

According to Dutch law, the Medical Research Involving Human Subjects Act (WMO), this study is exempted from ethical approval from a formal medical ethical committee. Informed consent was obtained from participants to collect data anonymously, and only to be used by the SDS in cooperation with researchers for peer-reviewed publication.

2.3 | Survey instrument

The questionnaire consisted of 20 questions and was designed by SDS to learn about parental experiences with the prenatal screening program and pre- and postnatal diagnosis for DS (see supporting information). Participants first answered questions on demographics (year of birth of child, and level of education of the mother and of the father) and whether they were members of SDS. The questionnaire was then divided in two parts: screening and diagnosis.

All parents in our survey had obtained information at two important points in time. First, they were informed that there were reasons for assuming a diagnosis of DS—from a prenatal ultrasound finding indicative for DS, a positive prenatal screen result of ftCT or NIPS, or physical features of DS identified after birth. We are calling this time point the "indicative diagnosis." Second, they were informed about the results of the confirmatory genetic tests. We are calling this time point the "confirmed diagnosis." A separate set of questions was asked about these two moments of information: what indicated (the possibility of) DS in the child, who delivered the message, and whether the information provided was sufficient (completely sufficient, mostly sufficient) or insufficient (insufficient to some degree, completely insufficient). In addition, all respondents who received a prenatal test result (indicative) for DS were asked whether, during the prenatal period, they had perceived emphasis by healthcare professionals on the option of terminating the pregnancy. Participants could then answer an open-ended question, which asked them to describe briefly how they had experienced the way the diagnostic information was delivered, both at the time of indicative and confirmed diagnosis.

The last set of questions of the survey was related to screening, starting with the question on whether prenatal testing was offered during pregnancy. Respondents then answered questions regarding the availability and perceived quality of the pre-test information. Finally, respondents were asked whether they decided or not to have a prenatal test. If they had decided to defer, the final (closed-category) question asked why they had decided to decline the offer of prenatal testing.

2.4 | Data analysis

Descriptive analyses were used to describe the characteristics of the respondents. Data were analyzed using SPSS 22 (IBM Statistics). Baseline characteristics were given as numbers and percentages.

The open-ended questions were analyzed by two authors (NC and GdG). Independently, both authors allocated the text fragments into three categories: positive, negative, and not interpretable. "Not interpretable" was used if the text fragments seemed to relate to the mother's feelings or emotions at the moment of receiving the information (instead of relating to how the information provided by the counselor was perceived) or when it was not possible to interpret the text as being negative or positive. Inter-rater reliability was then determined (screening/examination indicative for DS: 84% [prenatal: 88%, postnatal: 83%]; confirmed diagnosis: 85% [prenatal: 88%, postnatal: 85%]) by comparing allocated categories. The answers for which there was disagreement were discussed to reach a level of agreement within both coders (Campbell, Quincy, Osserman, & Pedersen, 2013). In all instances, mutual agreement was achieved. Subsequently, the two categories (positive and negative) were subdivided into three key-themes: atmosphere, information-provision, and setting (thematic analysis) (Ritchie et al., 2013). For example, descriptive words such as "respectful," "warm," "open," "supportive," or "empathic" were categorized as "positive, atmosphere." Wording such as "unemphatic," "hasty," or "business-like" were categorized as "negative, atmosphere." An example of negative setting, as perceived by mothers, would be space in which too many people present.

Finally, as membership status in the SDS and maternal educational level might be of influence on outcome, we explored whether there were any statistically significant differences in answering categories between members of the SDS versus non-members and between parents with a high educational level (high vocational education or university) versus parents with a less high (low or medium) educational level. We tested with a *t*-test for Equality of Means by year of birth of the child and with Pearson Chi-Square for the other items (supplementary material).

3 | RESULTS

3.1 | Sample characteristics

An online questionnaire was completed by parents of children with DS born between 2010 and 2016 (n = 242), of which 212 of these were mothers and 30 were fathers (Table 1). For the reasons explained in the Methodology, these 30 fathers were excluded from the analysis.

From the years of birth 2010–2015, we had 205 mothers of children with DS respond to our survey, which is 17% of the estimated

TABLE 1 Sample characteristics

	n (%)
Number of responding mothers per year of birth of the child (n = 212)	
2010	39 (18)
2011	26 (12)
2012	41 (19)
2013	38 (18)
2014	32 (15)
2015	29 (14)
2016	7 (3)
Level of mother's education (n = 209)[a]	
Low ([pre-]primary school or lower secondary education)	30 (14)
Medium (senior general secondary education or basic to middle vocational education)	51 (24)
High (high vocational education or university)	128 (61)
Level of father's education (as reported by the mothers) (n = 206)[a]	
Low ([pre-]primary school or lower secondary education)	52 (25)
Medium (senior general secondary education or basic to middle vocational education)	39 (19)
High (high vocational education or university)	115 (56)
Membership of the Dutch Down syndrome foundation	
Currently	154 (73)
Never	49 (23)
Earlier, but not currently	9 (4)

[a]Level of education: Three mothers and six fathers had missing data.

total of 1,197 DS mothers in the Netherlands in that period. For this comparison only, we excluded the respondents with a birth in 2016 (*n* = 7), as our survey period closed relatively early in 2016; including them in an estimation of live numbers for the whole year of 2016 would have led to an underestimation of the response rate). Those seven mothers were included in all other analyses in this article. Most of the 212 mothers were highly educated: 61%, as compared to 46% in Dutch women aged 25–45 in the general population as of 2016 (CBS, n.d.). About 56% of the fathers (as reported by the mothers) were highly educated as compared to 40% in the general population. Most of the mothers were members of the SDS (73%); a few were former members (4%), and some had never been members (23%). The mothers who were members much more often had higher educational levels (71%) than that of the former- and non-members (35%).

3.2 | Prenatal screening: Information provision on Down syndrome

Of the mothers to whom ftCT was offered (*n* = 137), 133 answered a question about the information they received at that time. Of these mothers, 54% (*n* = 72) reported that at that time no information about DS was provided. About 11% (*n* = 14) of these mothers recalled receiving information regarding DS but judged this information to be completely insufficient. About 19% received what they felt was sufficient information on medical issues but insufficient information about living with a child with DS (*n* = 25). Only 22 mothers (17%) felt the pre-screening information to be sufficient both in terms of medical information and the information about living with a child with DS. Of the eight mothers to whom NIPS was offered, five (63%) reported that at that time no information about DS was provided; two (25%) received information about DS, but judged this information as completely insufficient; one (13%) got sufficient information on medical issues but no sufficient information about living with a child with DS. None evaluated the pre-screening information to be sufficient in both medical and social terms.

3.3 | Prenatal screening: Choices and test outcomes

In this study, 78% (*n* = 165) of the mothers had been offered prenatal screening or prenatal diagnostic testing; of these, 137 were offered ftCT (65%), 8 NIPS (4%), 47 amniocentesis (22%), and 22 chorionic villus sampling (CVS) (10%). Respondents were allowed to choose more than one answer; therefore, the total percentage is more than 100%, as more than one test may have been offered during their pregnancy.

In total, 50 mothers (24%) pursued some sort of prenatal test, mostly ftCT (*n* = 43; 20%), of which 16 reported a positive screen result (8%), while 27 (13%) had a false-negative ftCT test result. Three mothers opted for NIPS, two after a positive ftCT screen, and 1 because of ultrasound features indicative for DS. Invasive diagnostic procedures were performed in 19 mothers (9%), of which 5 were CVS and 14 were amniocenteses. Of these, seven were undertaken because of a feature on an ultrasound scan (two CVS, and five amniocenteses) (Figure 1).

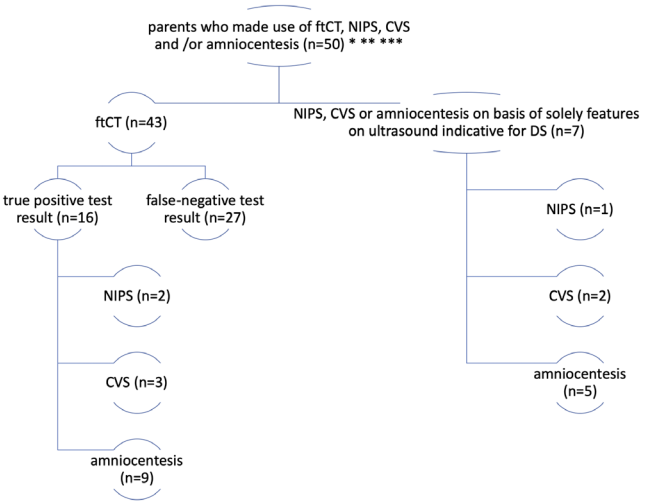

FIGURE 1 Parents who made use of ftCT, NIPS, CVS, and /or amniocentesis.
* Mothers can use more than, one test or not use any test at all. ** Of the ones with a positive ftCT result (16), 4 had a postnatal confirmed diagnosis, of which one after an NIPS, and 3 without a preceding NIPS. The one mother for whom the NIPS was used solely on basis of an ultrasound (right column), did an amniocentesis afterward. *** There were 13 mothers who reported that there was an ultrasound indicative for DS, but they choose not to participate in subsequent ftCT and/or NIPS. These 13 (with a postnatal confirmed diagnosis) are not in this figure. CVS, chorionic villus sampling; DS, Down syndrome; ftCT, first-trimester combined test; NIPS, non-invasive prenatal test
[Color figure can be viewed at wileyonlinelibrary.com]

3.4 | Reasons for declining screening

The majority of mothers in this survey did not have a prenatal test for DS during their pregnancy (*n* = 162, 76%). We asked these parents why they had declined the offer of testing. Respondents were allowed to choose more than one categorical response, and 162 women listed 619 reasons (~ 3.8 per patient). Main reasons for declining the test offer were "a child with DS is welcome in our family" (*n* = 114, 70% of 162) and "I felt already so emotionally attached to my child that abortion was not an option" (*n* = 53, 33%) (Table 2). In Table 2, we divided the answers into four overall categories: information-related, test-related, value-related, and cost-related reasons. Value-related reasons were predominant with 81% of mothers mentioning at least one value-related reason to decline prenatal screening. At least one information-related, respectively, test-related reason was mentioned by 36%, respectively, 35% of mothers. Mothers with higher education significantly more often mentioned test-related reasons than mothers with lower education (42% vs. 25%) and significantly more often answered they perceived the combined test not as a good screening

TABLE 2 Reasons for non-participation in prenatal screening given by mothers (n = 162)

	n (%)
Information-related reasons[a]	58 (36)
I did not know that prenatal tests existed	1 (1)
I assumed that in my case the chance for a child with Down syndrome was small	36 (22)
I just did not want to think about tests	4 (2)
During pregnancy, I never seriously thought about the possibility of Down syndrome	35 (21)
Test-related reasons[a]	56 (35)
The combined test (ftCT) is not a good screening test, as it only estimates s a rough chance	27 (17)
Invasive diagnostics (after a positive ftCT screening result) have a risk of causing a miscarriage	40 (25)
Value-related reasons[a]	132 (81)
A child with DS is welcome in our family	114 (70)
Abortion is against my religious conviction	26 (16)
I am not a proponent of abortion	33 (20)
I felt already so emotionally attached to my child that abortion was not an option	53 (33)
Cost-related reasons[a]	2 (1)
Costs were not covered by the assurance. It was too expensive	2 (1)

[a]We have clustered the different reasons into four overall categories. The numbers and percentages in an overall category refer to the numbers and percentages of the 163 mothers who have mentioned at least one of the reasons in the overall category.

test because of lack of accuracy (25% vs. 6%) (see supplementary material). Cost-related reasons were rarely (only by 1%) mentioned.

3.5 | Communication of a prenatal positive screening result or ultrasound result indicative for DS

Out of all 212 responding mothers, 18% had a screening test result indicative for DS (Table 3). Of these women, 69% reported that an ultrasound scan had given an indication for the presence of DS; 35% mentioned a positive ftCT result; and 5% had a positive NIPS result (categories not exclusive; Table 3). For ultrasound scans, no distinction was made between nuchal fold or a second-trimester anomaly scan in our questionnaire. Obstetricians were the clinicians who informed half of the parents who had a screening result indicative for DS (positive screening result for ftCT or NIPS and/or an indication on an ultrasound scan) (Table 3). Of the women receiving a screening result indicative for DS, 49% reported that the information provided at that moment was completely (23%) or mostly (26%) sufficient, while 26% felt that the information provided was insufficient to some degree, and another 26% rated the information as completely insufficient. Some parents reported an emphasis on the option of terminating pregnancy as the first choice, while none felt pressure toward continuing pregnancy (Table 3).

TABLE 3 Experiences with receiving a prenatal screening result (or ultrasound) indicative for DS, and a diagnostic prenatal genetic test result confirming Down syndrome

	n (%)
Prenatal screening result indicative for DS (n = 39)	
Indication[a]	
Ultrasound	27 (69)
First-trimester combined test (ftCT)[b]	14 (35)
Cell-free DNA based non-invasive prenatal test (NIPS)	2 (5)
Information provided by[a]	
Obstetrician	20 (51)
Midwife	6 (15)
General practitioner	0 (0)
Ultrasound operator	7(18)
Pediatrician	4 (10)
Clinical geneticist	4 (10)
Other (other medical specialist)	4 (10)
Perceived information provision	
Completely sufficient	9 (23)
Mostly sufficient	10 (26)
Insufficient to some degree	10 (26)
Completely insufficient	10 (26)
Perceived emphasis on the option of terminating pregnancy, after a prenatal screening result (or ultrasound) indicative for DS (or later on during the prenatal diagnostic process)[c]	
No perceived emphasis on the option of terminating pregnancy as the first choice	32 (84)
Pressure toward continuing the pregnancy	0 (0)
Perceived emphasis on the option of terminating pregnancy as the first choice	6 (16)
Prenatal confirmed (genetic) diagnosis by amniocentesis or chorionic villus sampling (n = 19)	
Information provided by[a]	
Obstetrician	11 (58)
Midwife	1 (5)
General practitioner	0 (0)
Pediatrician	1 (5)
Clinical geneticist	7 (37)
Other (no consultation)	1 (5)
Perceived information provision	
Completely sufficient	3 (16)
Mostly sufficient	9 (47)
Insufficient to some degree	4 (21)
Completely insufficient	3 (16)

[a]Total percentage is above 100%, as respondents were allowed to choose more than one answering category.
[b]In this question, 14 mothers report a positive ftCT screen result. If we look at the answers on later questions, positive ftCT screen results are mentioned by 16 mothers. Apparently, two of these mothers, in this earlier question, report that an ultrasound was the first indication for DS. So, in these two, probably the ftCT was done afterward on basis of the ultrasound result.
[c]This question has one missing data.

In an additional open-ended question, we asked the mothers to characterize the way a screening (test or ultrasound) result indicative for DS had been communicated. Thirty-two mothers responded to this question: 9 (28%) described their own feelings but did not reflect on the communication or used neutral terms about the communication, 11 (34%) described the communication in predominantly positive words (for instance, "respectful," "empathic," "taking enough time," "honest" [*atmosphere*]; "clear," "realistic," "forward-looking" [*information-provision*]); 12 (38%) in predominantly negative terms (for instance, "not empathic," "rude" [*atmosphere*]; "vague," "too negative about DS," "only about possible medical complications," "pressing toward TOP" [*information-provision*]).

> "*Our consultation was good, clear, respectful and empathic*" [Mother of a child with DS, screening result indicative for DS]

> "*We got a phone call in which they gave us the result. They literally said: we see a fetus with DS, when do you want an appointment for a termination of pregnancy.*" *[Mother of a child with DS, screening result indicative for DS]*

3.6 | Confirmed prenatal (genetic) diagnosis

Of the total group, 9% (*n* = 19) of the mothers received a confirmed (genetic) diagnosis for DS by invasive diagnostic testing (amniocentesis or chorionic villus sampling [CVS]). Communicating the diagnostic result to the mothers was mainly done by an obstetrician (*n* = 11; 58%). The majority of these mothers (*n* = 12; 63%) thought the information provided at that moment was completely (16%) or mostly sufficient (47%), while 4 (21%) thought this information to be insufficient to some degree, and 3 (16%) considered the information as totally insufficient.

Additionally, we asked the mothers if they could characterize the way the prenatal confirmed diagnosis had been communicated. Of the 16 mothers responding to this question, 4 (25%) only reflected on their own feelings or used neutral terms for the communication; 8 (50%) used predominantly positive terms ("respectful," "supportive," "understanding" (*atmosphere*); "clear," "helpful," "enlightening," "respect for decision" (*information-provision*)); and 4 (25%) predominantly negative wordings (by phone—which was not appreciated by the parent (*setting*); "zero knowledge," "no information," "focused on medical problems," "pressing toward TOP" (*information-provision*)).

> "*There was a positive atmosphere, informative, very respectful and understanding*" *[Mother of a child with DS, confirmed prenatal diagnosis]*

> "*It was entirely negative, only about medical issues that can occur in people with Down syndrome*" *[Mother of a child with DS, confirmed prenatal diagnosis]*

3.7 | Communication of postnatal examination indicative of Down syndrome

Of the responding mothers, the majority (*n* = 173, 72%) received information on indication for DS postnatally—that is, physical features of the neonate gave reason to consider DS (Table 4). In most of these 173 children, this was seen immediately after or within a few hours after birth of the baby (*n* = 139, 80%). Late indications for DS, more than 1 week after birth, were rare (*n* = 10, 6%). Most mothers were informed of an indication for DS by a pediatrician (*n* = 99, 57%) or by a midwife (*n* = 47, 27%) (Table 5). Most respondents (*n* = 107, 62%) thought that the information provided at that moment was completely (32%) or mostly sufficient (30%) while 24% considered the information to be insufficient to some degree, and another 15% (*n* = 25) thought this information was completely insufficient (Table 5).

Of the 155 mothers responding to the open-ended question about the way the postnatal indication for DS had been communicated, 50 (32%) reflected on their own feelings or used neutral terms for the communication; 65 (42%) used predominantly positive terms ("calm," "understanding," "reassuring," "open," "honest," "respectful," "loving," "caring," "careful," "sensitive," "realistic," "with some humor" (*atmosphere*); "competent," "clear," "to-the-point," "honest," "enlightening," "precise," "informative" (*information-provision*)); and 40 (26%) predominantly negative wordings ("when sharing the news there were too many people present," "one parent not present," "while grandparents were present," "while perineal repair was done" (*setting*); "awful," "too quickly," "formal," "painful," "hectic," "not-empathic," "cool," "impersonal," "cold," "distant" (*atmosphere*); "unclear," "vague," "avoiding," "unrealistic," "not informative," "too much about additional medical problems" (*information-provision*)).

TABLE 4 The timing of indication for DS and confirmed (genetic) diagnosis (*n* = 212)

	***n* (%)**
Indication for DS[a]	
Prenatal	39 (18)
Immediately at birth	91 (43)
Not immediately at birth, but within a few hours after birth	48 (23)
Not within a few hours, but within a week after birth	24 (11)
Within 1–4 weeks after birth	6 (3)
Within 4–12 weeks after birth	0 (0)
More than 12 weeks after birth	4 (2)
Confirmed (genetic) diagnosis	
Prenatal	19 (9)
Within a week after birth	150 (71)
Within 1–4 weeks after birth	33 (16)
Within 4–12 weeks after birth	5 (2)
More than 12 weeks after birth	5 (2)

[a]In postnatal situations, an indication for DS, is the anticipation that the child might have DS on basis of physical features of the neonate.

TABLE 5 Experiences with being informed on an indication for DS and/or genetic diagnosis of Down syndrome in newborns

	n (%)
Postnatal indication for DS (*n* = 173)	
Information provided by[a]	
Obstetrician	16 (9)
Midwife	47 (17)
General practitioner	0 (0)
Pediatrician	99 (57)
Clinical geneticist	2 (1)
Nurse practitioner	7 (4)
Doctor at child services	2 (1)
Parents themselves recognized DS and pointed this out to the professionals	18 (10)
Other (medical doctor under training; family friend)	2 (1)
Perceived information provision	
Completely sufficient	56 (32)
Mostly sufficient	51 (30)
Insufficient to some degree	41 (24)
Completely insufficient	25 (15)
Confirmed (genetic) diagnosis (*n* = 193)	
Information provided by[a]	
Obstetrician	3 (2)
Midwife	3 (2)
General practitioner	3 (2)
Pediatrician	174 (90)
Clinical geneticist	10 (5)
Other (no consultation; other medical specialist; nurse practitioner)	8 (4)
Perceived information provision	
Completely sufficient	83 (43)
Mostly sufficient	72 (37)
Insufficient to some degree	33 (17)
Completely insufficient	5 (3)

[a]Total percentage is above 100%, as respondents were allowed to choose more than one answering category.

> "*It was clear, precise, positive, empathic, and at the same time encouraging.*" [Mother of a child with DS, postnatal indication for DS]

> "*It was unrealistic, insensitive, clumsy, and not clear.*" [Mother of a child with DS, postnatal indication for DS]

3.8 | Confirmed postnatal diagnosis

The majority of mothers received a confirmed (genetic) diagnosis for DS (*n* = 193, 91%) postnatally—that is, karyotyping was performed after birth (Table 5). For most of these 193 mothers (*n* = 150; 78%), this diagnosis was delivered within a week after birth. In 33 cases (17%), this was within 1–4 weeks. Genetic diagnoses more than 4 weeks after birth were rare and reported for only 10 children (5%). Most mothers (*n* = 174, 90% of 193) were informed on the postnatal definitive diagnosis by a pediatrician (Table 5). Although the majority (*n* = 155, 80%) thought the information provided at that moment was completely (43%) or mostly sufficient (37%), 33 (17%) considered the information insufficient to some degree, and another 5 (3%) thought this information was completely insufficient (Table 5).

Of the 166 mothers responding to the additional open question about the way the diagnosis was delivered, 40 (24%) reflected on their own feelings or used neutral terms for the communication; 98 (59%) used predominantly positive terms ("open," "nice," "understanding," "calm," "personal," "warm," "sensitive," "empathic," "reassuring," "positive," "safe," "supportive," "concerned" (*atmosphere*); "clear," "extensive," "informative," "competent," "careful," "precise," "factual," "neutral," "to the point," "complete," "open-minded" (*information-provision*)); and 28 (17%) predominantly negative wordings ("business-like," "hasty," "not empathic," "negative," "blaming," "depressing" (*atmosphere*); "outdated," "incompetent," "information through informational leaflet," "vague," "unclear," "too much about potential medical problems," "little information" (*information-provision*)).

> "*It was an open communication, without prejudices, and informative. It was matched to our experience as parents.*" *[Mother of a child with DS, confirmed postnatal diagnosis]*

> "*It was very medical. It would have been better if the message had been that it is 'only' Down syndrome, and that good support and care, if needed, is available. We got a booklet with all possible medical complications. Necessary information, but it should be communicated with more nuance.*" *[Mother of child with DS, confirmed postnatal diagnosis]*

3.9 | Differences between subgroups

In the supplementary material, we present an analysis of the differences (a) between members of the SDS and non-members and (b) between mothers with a different educational level. Mothers who were members of the SDS more often had a high educational level, which also holds true for the fathers connected to these mothers. Highly educated mothers significantly more often mentioned one or more test-related reasons for not choosing prenatal screening (42% vs. 25% in less highly educated mothers) and that ftCT test properties (only estimating a rough chance) were a reason to decline prenatal screening (25% vs. 6% in less highly educated mothers).

4 | DISCUSSION

In this study, we have explored the experiences and opinions around prenatal and postnatal testing in mothers with (live born) children with

DS. During the study period of this research, ftCT was the initial screening test offered, followed eventually by NIPS in case of a positive screen result. The majority of the mothers in our study received the indication for/diagnosis of DS *after* their child was born. This not only reflects the relatively low uptake of prenatal screening for DS in the Netherlands during the study period (2010–2016) but also that in the majority of cases a prenatal diagnosis of DS at the time was followed by a TOP.

Value-related, as well as information- and test-related reasons, affected uptake (Crombag et al., 2016). Our study suggests that Dutch pregnant women who have declined screening, by and large, did not consider DS a condition severe enough to justify TOP, while some women also seemed to incorporate screening test characteristics into their decision (Crombag, van Schendel, Schielen, Bensing, & Henneman, 2016). In the period under study, until January 2015, ftCT was fully covered for women 36 years and older, whereas younger women paid an additional fee. Charging a fee only for younger women might have conveyed the reassuring message that screening is not needed for younger women, because having a child with DS was less probable. This might in part explain the relatively low Dutch uptake rates, in general, during this period. However, in our study, value-related reasons were predominant in declining screening. A minority mentioned information-related reasons (36%), among whom 22% believed screening to be not necessary assuming that in their case the chance for having a child with DS was slim.

Since the study was completed, pregnant women more often choose NIPS (42%) over ftCT (3%), leading to an increase of the overall uptake (NIPT Consortium the Netherlands, 2018), which is consistent with earlier predictions (Lewis, Hill, Silcock, Daley, & Chitty, 2014; van Schendel et al., 2015; Verweij, Oepkes, & Boer, 2013). However, the uptake in the Netherlands still is relatively low in comparison to other European countries (Crombag, 2016; Crombag et al., 2014). This is what one might expect on the basis of our findings, which suggest that most women decline screening for value-related reasons. Parents who declined screening for test quality and to avoid invasive testing probably will be more inclined to pursue NIPS. As a consequence, a somewhat larger group of parents will receive a prenatal test-result. Some of these parents might decide to terminate the pregnancy, while others will use the test results as a means to prepare themselves for the birth of their baby (Crombag, 2016; Verweij et al., 2013). In prenatal counseling, the needs of both these groups are to be accommodated.

Receiving news of a prenatal or postnatal indication or diagnosis for DS is often unexpected and can be overwhelming. In these situations, parents seem to be particularly sensitive to verbal and non-verbal communication (Asplin et al., 2013; Black, 2011; Lou et al., 2017). In our study, if mothers were positive about the communication, the communication was described in very positive words; and vice versa, when negative, the description used was very negative, in line with the strong emotional impact of this specific life event. Prenatally, "positive"-scored wordings were related to supportive information that was focused on the future (forward-looking). In other words, the positive information was optimistic toward having a child with DS while negative wording was related to negative aspects of DS and perceived as over-emphasizing of options of TOP. A similar association was found by Skotko (Skotko, 2005a; Skotko, 2005b). Mothers receiving a prenatal diagnosis for DS, and intending to continue their pregnancy, would be more likely to have a positive experience when the positive aspects of DS are discussed alongside other information (Skotko, 2005a). The information communicated and parents' perception of this information is crucial, as it affects the ability of parents to cope with the diagnosis (Kratovil & Julion, 2017), both in prenatal and in postnatal situations. Besides the impact of the information, the perceived atmosphere is crucial on how the conversation is judged. Atmosphere is determined by level of empathy. Terms as "respect, warmth and support" were used for positive rated conversations, while words as "hasty, negative, cool, and distant" were used for negative rated conversations.

In this study, few mothers (16% of 38), recall some perceived emphasis on the option of TOP as the first choice, while none perceived pressure toward continuation of their pregnancy. Dutch health council reports, professional guidance, and patient information are unanimous about the goal of prenatal screening: "to inform prospective parents of the risk of DS in the ongoing pregnancy, providing them with timely options, including invasive diagnostic procedures in the case of an increased risk for DS (screen positive), and if diagnosed for DS, to prepare for caring for a disabled child or a TOP" (National Institute for Public Health and the Environment, n.d.; Health Council of the Netherlands, 2007; Health Council of the Netherlands, 2016b). The finding that some mothers perceived an emphasis on the option TOP as the first choice is discordant with this aim. Whether counselors explicitly pressed parents toward a TOP or whether parents interpreted mentioning TOP as pressure is difficult to conclude from a retrospective analysis, but it is undesirable that parents are left with this feeling (Tijmstra, Bosboom, & Bouman, 2000). Some parents deciding to continue their pregnancy appear to prefer to distance themselves from experiencing negative, medical information and risk scenarios (Black, 2011; Lalor & Begley, 2006). Contrary, for prospective parents considering a TOP, this is different (Asplin et al., 2013; Hunt et al., 2009; Lou et al., 2017).

From the results of this study, satisfaction was highest among those mothers who had received a confirmed postnatal diagnosis and lowest among women receiving a prenatal screening result indicative for DS, potentially related to the available options and difficult choices being discussed. As presented in this study, parents continuing their pregnancy prefer to receive positive information on DS, focused on the future. Apparently, this is more likely to be discussed in postnatal situations.

Although the findings of this study regarding the delivery of postnatal diagnoses are mainly positive, there is certainly room for improvement. Recommendations based on parental preferences and guidelines are available (Borstlap et al., 2011; de Groot-van der Mooren et al., 2014; Sheets et al., 2011; Skotko, Capone, & Kishnani, 2009). Setting, atmosphere, and information-provision in postnatal communication are crucial and remembered for a long time (Committee on Genetics, 2001; Dent & Carey, 2006).

Guidelines also exist for the delivery of prenatal screening results indicative for DS or confirmed prenatal diagnosis. Expectant parents often use this information to make pregnancy decisions, so the circumstances are essential for both the provider and the couple. Discussing these options in a balanced way can be challenging, particularly when parents already have an unspoken preference toward one pregnancy option (Lou et al., 2018) as reflected in the results of this study. Careful exploration of preferences seems to be crucial, and adequate use and compliance to existing guidelines would be helpful (Sheets et al., 2011; Skotko, Capone, & Kishnani, 2009).

Even with some training on breaking difficult news and existing guidelines, sharing the news on a prenatal or postnatal indication or diagnosis for DS can be challenging (Horwitz & Ellis, 2007). Pediatricians are typically the communicators of the postnatal findings. While guidelines were used, de Groot et al. showed that among a group of Dutch pediatricians, still 10% recalled a dissatisfaction with first communication of sharing the news of a DS diagnosis (de Groot-van der Mooren et al., 2014). However, because of the multiple-step procedure toward diagnosis and the different moments in time, various other healthcare professionals are often involved. Those healthcare professionals have different levels of experience and knowledge with this type of conversation (Farrell, Agatisa, Mercer, Mitchum, & Coleridge, 2016), not always in line with the patient's preferences (J. G. Lalor, Devane, & Begley, 2007; Martin, Hutton, Spelten, Gitsels-Van der Wal, & Van Dulmen, 2014).

4.1 | Strengths and limitations

With this study, we have presented the reasons of parents with a DS child to refrain from prenatal testing and their experiences in receiving the diagnosis. This is the first study interrogating mothers of children with DS on the background of their prenatal choices and their experiences with the screening and diagnostic process. On basis of their feedback, we make recommendations for delivering prenatal (indicative and confirmed) diagnosis.

Our study has the following limitations. First, the study might be subject to recall bias, as respondents had to reflect on life events which could be up to 6 years ago. Their strong positive or negative evaluations of the delivery of the diagnosis might have been morphed as a result of the parents' growing knowledge of DS and intimate subsequent experiences with their own child. On the other hand, there are studies that show that such experiences are “flashbulb memories,” accurate and vivid memories of the events, even years afterward (Carr, 1988; Skotko, 2005a; Skotko, 2005b).

Second, our study could be subject to selection bias. As participants were invited through the e-newsletter, magazine, Facebook page, and website of the SDS, a majority of participants were members of SDS. If these parents have different opinions than those of parents who were not involved in the SDS, the results of our survey might not be generalizable to all parents of children with DS. Third, there is an overrepresentation of highly educated mothers, which might also limit generalizability. However, when comparing mothers by SDS membership status, we found no statistically significant differences in answering categories, apart from differences in maternal and paternal educational level by membership (supplementary material). In addition, an analysis by maternal educational level (supplementary material) reveals that the content of the answers of highly educated mothers and less highly educated mothers (apart from differences in membership status of the SDS, and differences in the educational level of their partners) are highly similar; the only statistically significant differences were that highly educated mothers more often mention a test-related reason for non-participation in screening (42% vs. 25%), which is the result of these mothers more often reporting that the impreciseness of the ftCT test was a reason to abstain from screening (25% vs. 6%). So, at least inside our sample, there is no evidence that there are large differences in experiences and opinions between SDS members and non-members, or between highly educated mothers and less highly educated mothers. Fourth, our results are limited by a lack of information on ethnicity of our respondents. This item was not included in our survey (to maximize participation, we chose for a short questionnaire, without too many possibly intrusive questions). Another recent study which recruited participants though the media channels of the SDS reveals that only very few parents with an original non-Dutch cultural background participated (only 3% of the sample) (de Graaf, Levine, Goldstein, & Skotko, 2019), so we expect this also to be the case in the current study. There have been a few Dutch studies that targeted the screening experiences and opinions of pregnant women of different ethnic backgrounds (Fransen et al., 2009; Fransen et al., 2010), and there studies reveal some differences in informational background and needs. More research into these cultural differences is recommended.

Although we have given a voice to mothers of children with DS, the important voices of women who chose to terminate a pregnancy are absent, as a substantial group of Dutch parents will decide to have TOP after a diagnostic prenatal genetic test result confirming DS. There is some research into this group (Korenromp, Page-Christiaens, van den Bout, Mulder, & Visser, 2007; Tijmstra et al., 2000), however, more research is needed on their experiences and needs to complete practical implications.

In addition, as we only had a very limited number of responding fathers, we have excluded fathers from our analyses. However, this does not mean that their experiences and opinions do not matter. More research into their informational needs is recommended.

5 | CONCLUSIONS

In conclusion, with recent developments in screening, more parents are expected to receive the diagnosis of fetal DS before birth, including parents who will continue their pregnancy. Women/ prospective parents do not just have one reason for declining the offer of prenatal screening. In 81% of mothers of a live born child with DS, value-related motives played a role. One third of the mothers also stated that the low predictive values of the available screening test (ftCT) and the potential risks associated with follow-up tests prevented

them from opting for prenatal screening. The percentage of women perceiving the information received at different stages of the pre- or postnatal trajectory, as completely insufficient ranged between 3 and 26% for the different situations, providing room for improvement in both pre-and postnatal delivery of the news. Meeting the individual needs of expectant parents requires careful exploration of pregnancy preferences (e.g., termination or continuation), with special attention to setting and atmosphere and adequate implementation of existing guidelines.

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CONFLICT OF INTEREST

G. C. M. L. P.-C. reports that he has been a full-time employee of Illumina since January 18, 2016.

DATA AVAILABILITY STATEMENT

Data Availability Statement The data that support the findings of this study are available on request from the corresponding author. The data are not publicly available due to privacy or ethical restrictions.

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REFERENCES

Asplin, N., Wessel, H., Marions, L., & Öhman, S. G. (2013). Pregnant women's perspectives on decision-making when a fetal malformation is detected by ultrasound examination. *Sexual and Reproductive Healthcare*, *4*, 79–84. https://doi.org/10.1016/j.srhc.2013.02.001

Aune, I., & Möller, A. (2012). 'I want a choice, but I don't want to decide'—A qualitative study of pregnant women's experiences regarding early ultrasound risk assessment for chromosomal anomalies. *Midwifery*, *28*, 14–23. https://doi.org/10.1016/j.midw.2010.10.015

Black, B. P. (2011). Truth telling and severe fetal diagnosis: A virtue ethics perspective. *Journal of Perinatal and Neonatal Nursing*, *25*, 13–20. https://doi.org/10.1097/JPN.0b013e318201edff

Blondel, B., Lelong, N., Kermarrec, M., & Goffinet, F. (2012). Trends in perinatal health in France from 1995 to 2010. Results from the French National Perinatal Surveys. *Journal de Gynecologie Obstetrique et Biologie de La Reproduction*, *41*, e1–e15. https://doi.org/10.1016/j.jgyn.2012.04.014

Borstlap, R., van Gameren-Oosterom, H., Lincke, C., Weijerman, M., van Wieringen, H., van Wouwe, J., & S, W. D. (2011). *An update of the multidisciplinary guideline for the medical guidance of children with Down syndrome*, Leiden. Retrieved from https://www.tno.nl/media/1934/richtlijn-downsyndroom-dec-2011-definitief.pdf

Campbell, J. L., Quincy, C., Osserman, J., & Pedersen, O. K. (2013). Coding in-depth semistructured interviews: Problems of unitization and intercoder reliability and agreement. *Sociological Methods and Research*, *42*, 294–320. https://doi.org/10.1177/0049124113500475

Carr, J. (1988). Six weeks to twenty-one years old: A longitudinal study of children with Down's syndrome and their families. *Journal of Child Psychology and Psychiatry*. https://doi.org/10.1111/j.1469-7610.1988.tb00734.x

CBS. (n.d.). *Tabel Bevolking: hoogstbehaald onderwijsniveau en onderwijsrichting*.

Committee on Genetics. (2001). American Academy of Pediatrics: Health supervision for children with Down syndrome. *Pediatrics*, *107*, 442–449. https://doi.org/10.1542/peds.107.2.442

Crombag, N. M. T. H., Vellinga, Y. E., Kluijfhout, S. A., Bryant, L. D., Ward, P. A., Iedema-Kuiper, R., … Hirst, J. (2014). Explaining variation in Down's syndrome screening uptake: Comparing The Netherlands with England and Denmark using documentary analysis and expert stakeholder interviews. *BMC Health Services Research*, *14*(1), 437. https://doi.org/10.1186/1472-6963-14-437

Crombag, N. M.T.H. (2016). *Explaining low uptake for down syndrome screening in the Netherlands (and predicting utilisation of other programmes)*. Utrecht University.

Crombag, N. M. T. H., Boeije, H., Iedema-Kuiper, R., Schielen, P. C. J. I., Visser, G. H. A., & Bensing, J. M. (2016). Reasons for accepting or declining down syndrome screening in Dutch prospective mothers within the context of national policy and healthcare system characteristics: A qualitative study. *BMC Pregnancy and Childbirth*, *16*, 121. https://doi.org/10.1186/s12884-016-0910-3

Crombag, N. M. T. H., van Schendel, R. V., Schielen, P. C. J. I., Bensing, J. M., & Henneman, L. (2016). Present to future: What the reasons for declining first-trimester combined testing tell us about accepting or declining cell-free DNA testing. *Prenatal Diagnosis.*, *36*, 587–590. https://doi.org/10.1002/pd.4824

de Graaf, G., Engelen, J. J. M., Gijsbers, A. C. J., Hochstenbach, R., Hoffer, M. J. V., Kooper, A. J. A., … Voorhoeve, E. (2017). Estimates of live birth prevalence of children with down syndrome in the period 1991–2015 in The Netherlands. *Journal of Intellectual Disability Research*, *61*, 461–470. https://doi.org/10.1111/jir.12371

de Graaf, G., Haveman, M., Hochstenbach, R., Engelen, J., Gerssen-Schoorl, K., Poddighe, P., … van Hove, G. (2011). Changes in yearly birth prevalence rates of children with down syndrome in the period 1986-2007 in The Netherlands. *Journal of Intellectual Disability Research*, *55*, 462–473. https://doi.org/10.1111/j.1365-2788.2011.01398.x

de Graaf, G., Levine, S., Goldstein, R., & Skotko, B. (2019). Parents' perceptions of functional abilities in people with down syndrome. *American Journal of Medical Genetics, Part A*, *179*(2), 161–176. https://doi.org/10.1002/ajmg.a.61004

de Groot-van der Mooren, M. D., Gemke, R. J. B. J., Cornel, M. C., & Weijerman, M. E. (2014). Neonatal diagnosis of down syndrome in The Netherlands: Suspicion and communication with parents. *Journal of Intellectual Disability Research*, *58*, 953–961. https://doi.org/10.1111/jir.12125

Dent, K. M., & Carey, J. C. (2006). Breaking difficult news in a newborn setting: Down syndrome as a paradigm. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, *142C*, 173–179. https://doi.org/10.1002/ajmg.c.30100

Ekelund, C. K., Petersen, O. B., Skibsted, L., Kjaergaard, S., Vogel, I., & Tabor, A. (2011). First-trimester screening for trisomy 21 in Denmark: Implications for detection and birth rates of trisomy 18 and trisomy 13. *Ultrasound in Obstetrics and Gynecology*, *38*, 140–144. https://doi.org/10.1002/uog.8929

Farrell, R. M., Agatisa, P. K., Mercer, M. B., Mitchum, A. G., & Coleridge, M. B. (2016). The use of noninvasive prenatal testing in obstetric care: Educational resources, practice patterns, and barriers reported by a national sample of clinicians. *Prenatal Diagnosis*, *36*, 499–506. https://doi.org/10.1002/pd.4812

Fransen, M. P., Essink-Bot, M. L., Vogel, I., Mackenbach, J. P., Steegers, E. A. P., & Wildschut, H. I. J. (2010). Ethnic differences in informed decision-making about prenatal screening for Down's syndrome. *Journal of Epidemiology and Community Health*, *64*, 262–268. https://doi.org/10.1136/jech.2009.088237

Fransen, M. P., Wildschut, H. I. J., Vogel, I., Mackenbach, J. P., Steegers, E. A. P., & Essink-Bot, M. L. (2009). Ethnic differences in considerations whether or not to participate in prenatal screening for

down syndrome. *Prenatal Diagnosis*, *29*, 1262–1269. https://doi.org/10.1002/pd.2391

Gitsels-van der Wal, J. T., Martin, L., Manniën, J., Verhoeven, P., Hutton, E. K., & Reinders, H. S. (2015). A qualitative study on how Muslim women of Moroccan descent approach antenatal anomaly screening. *Midwifery*, *31*, e43–e49. https://doi.org/10.1016/j.midw.2014.12.007

Gottfredsdóttir, H., Björnsdóttir, K., & Sandall, J. (2009). How do prospective parents who decline prenatal screening account for their decision? A qualitative study. *Social Science and Medicine*, *69*, 274–277. https://doi.org/10.1016/j.socscimed.2009.05.004

Harmsen, M., Liefers, J., Cruijsberg, J., & Atsma, F. (2017). *De prenatale screening op downsyndroom en het structureel echoscopisch onderzoek*. Publieksmonitor 2016. Nijmegen. Retrieved from https://www.rivm.nl/sites/default/files/2018-11/Monitor2016PubliekscreeningsprogrammadownsyndroomSEO-1.pdf

Health Council of the Netherlands. (2016a). *Health Council of The Netherlands*. *Prenatal screening*. The Hague. https://doi.org/978-94-6281-088-4

Health Council of the Netherlands. (2016b). *Population Screening Act: Non-Invasive Prenatal Test (NIPT) as initial test for Down's, Patau's and Edwards' syndrome*. The Hague. https://doi.org/publication.

Health Council of the Netherlands. (2007). *Population Screening Act: Prenatal Screening: Down's syndrome and neural tube defects*. The Hague. https://doi.org/2007/05WBO

Horwitz, N., & Ellis, J. (2007). Paediatric SpRs' experiences of breaking bad news. *Child: Care, Health and Development*, *33*, 625–630. https://doi.org/10.1111/j.1365-2214.2007.00742.x

Hunt, K., France, E., Ziebland, S., Field, K., & Wyke, S. (2009). "My brain couldn't move from planning a birth to planning a funeral": A qualitative study of parents' experiences of decisions after ending a pregnancy for fetal abnormality. *International Journal of Nursing Studies*, *46*, 1111–1121. https://doi.org/10.1016/j.ijnurstu.2008.12.004

Ingvoldstad, C., Öhman, S. G., & Lindgren, P. (2014). Implementation of combined ultrasound and biochemistry for risk evaluation of chromosomal abnormalities during the first trimester in Sweden. *Acta Obstetricia et Gynecologica Scandinavica*, *93*, 868–873. https://doi.org/10.1111/aogs.12445

Korenromp, M. J., Page-Christiaens, G. C. M. L., van den Bout, J., Mulder, E. J. H., & Visser, G. H. A. (2007). Maternal decision to terminate pregnancy in case of down syndrome. *American Journal of Obstetrics and Gynecology*, *196*, 149.e1–149.e11. https://doi.org/10.1016/j.ajog.2006.09.013

Kratovil, A. L., & Julion, W. A. (2017). Health-care provider communication with expectant parents during a prenatal diagnosis: An integrative review. *Journal of Perinatology*, *37*, 2–12. https://doi.org/10.1038/jp.2016.123

Lalor, J., & Begley, C. (2006). Fetal anomaly screening: What do women want to know? *Journal of Advanced Nursing*, *55*, 11–19. https://doi.org/10.1111/j.1365-2648.2006.03884.x

Lalor, J. G., Devane, D., & Begley, C. M. (2007). Unexpected diagnosis of fetal abnormality: Women's encounters with caregivers. *Birth*, *34*, 80–88. https://doi.org/10.1111/j.1523-536X.2006.00148.x

Lewis, C., Hill, M., Silcock, C., Daley, R., & Chitty, L. S. (2014). Non-invasive prenatal testing for trisomy 21: A cross-sectional survey of service users' views and likely uptake. *BJOG: An International Journal of Obstetrics and Gynaecology*, *121*, 582–594. https://doi.org/10.1111/1471-0528.12579

Lotto, R., Smith, L. K., & Armstrong, N. (2018). Diagnosis of a severe congenital anomaly: A qualitative analysis of parental decision making and the implications for healthcare encounters. *Health Expectations*, *21*, 678–684. https://doi.org/10.1111/hex.12664

Lou, S., Carstensen, K., Petersen, O. B., Nielsen, C. P., Hvidman, L., Lanther, M. R., & Vogel, I. (2018). Termination of pregnancy following a prenatal diagnosis of down syndrome: A qualitative study of the decision-making process of pregnant couples. *Acta Obstetricia et Gynecologica Scandinavica*, *97*, 1228–1236. https://doi.org/10.1111/aogs.13386

Lou, S., Jensen, L. G., Petersen, O. B., Vogel, I., Hvidman, L., Møller, A., & Nielsen, C. P. (2017). Parental response to severe or lethal prenatal diagnosis: A systematic review of qualitative studies. *Prenatal Diagnosis*, *37*, 731–743. https://doi.org/10.1002/pd.5093

Lou, S., Petersen, O. B., Jørgensen, F. S., Lund, I. C. B., Kjærgaard, S., Vogel, I., … Hansen, J. F. (2018). National screening guidelines and developments in prenatal diagnoses and live births of down syndrome in 1973–2016 in Denmark. *Acta Obstetricia et Gynecologica Scandinavica*., *97*, 195–203. https://doi.org/10.1111/aogs.13273

Martin, L., Van Dulmen, S., Spelten, E., De Jonge, A., De Cock, P., & Hutton, E. (2013). Prenatal counseling for congenital anomaly tests: Parental preferences and perceptions of midwife performance. *Prenatal Diagnosis*, *33*, 341–353. https://doi.org/10.1002/pd.4074

Martin, L., Hutton, E. K., Spelten, E. R., Gitsels-Van der Wal, J. T., & Van Dulmen, S. (2014). Midwives' views on appropriate antenatal counselling for congenital anomaly tests: Do they match clients' preferences? *Midwifery*, *30*, 600–609. https://doi.org/10.1016/j.midw.2013.08.012

National Institute for Public Health and the Environment. (n.d.). *Information about the screening for Down syndrome, Edwards' syndrome and Patau's syndrome*. Retrieved from https://www.rivm.nl/dsresource?objectid=4803473c-eb05-4f68-b1cf-99b235df710f&type=pdf&disposition=inline

Neyt, M., Hulstaert, F., & Gyselaers, W. (2014). Introducing the non-invasive prenatal test for trisomy 21 in Belgium: A cost-consequences analysis. *BMJ Open*, *4*, e005922. https://doi.org/10.1136/bmjopen-2014-005922

Nipt Consortium the Netherlands. (2018). Retrieved from https://niptconsortium.nl/nieuws/nipt-beschikbaar-voor-alle-zwangeren-resultaten-eerste-jaar/

Ritchie, J., Lewis, J., Nicholls, C. M. N., & Ormston, R. (2013). *Qualitative research practice a guide for social science students and researchers*. London, UK: Sage.

Sheets, K. B., Crissman, B. G., Feist, C. D., Sell, S. L., Johnson, L. R., Donahue, K. C., … Brasington, C. K. (2011). Practice guidelines for communicating a prenatal or postnatal diagnosis of down syndrome: Recommendations of the National Society of genetic Counselors. *Journal of Genetic Counseling*, *20*, 432–441. https://doi.org/10.1007/s10897-011-9375-8

Skotko, B. (2005a). Mothers of children with down syndrome reflect on their postnatal support. *Pediatrics*, *115*, 64–77. https://doi.org/10.1542/peds.2004-0928

Skotko, B. G., Capone, G. T., & Kishnani, P. S. (2009). Postnatal diagnosis of Down syndrome: Synthesis of the evidence on how best to deliver the news. *Pediatrics*. *124*(4), e751–e758. https://doi.org/10.1542/peds.2009-0480

Skotko, B. G. (2005b). Prenatally diagnosed down syndrome: Mothers who continued their pregnancies evaluate their health care providers. *American Journal of Obstetrics and Gynecology*, *192*, 670–677. https://doi.org/10.1016/j.ajog.2004.11.001

Skotko, B. G., Kishnani, P. S., & Capone, G. T. (2009). Prenatal diagnosis of down syndrome: How best to deliver the news. *American Journal of Medical Genetics*, *Part A*, *149A*, 2361–2367. https://doi.org/10.1002/ajmg.a.33082

Sommerseth, E., & Sundby, J. (2010). Women's experiences when ultrasound examinations give unexpected findings in the second trimester. *Women and Birth*, *23*, 111–116. https://doi.org/10.1016/j.wombi.2010.01.001

Tijmstra, T. J., Bosboom, G. J., & Bouman, K. (2000). Ervaringen van vrouwen die besloten de zwangerschap uit te dragen na de diagnose "Down-syndroom." [Experiences of women who decided to continue the pregnancy after diagnosis of Down's syndrome]. *Ned Tijdschr Geneeskd*, *144*, 2104–2107.

van Schendel, R. V., Dondorp, W. J., Timmermans, D. R. M., van Hugte, E. J. H., de Boer, A., Pajkrt, E., … Henneman, L. (2015). NIPT-based screening for down syndrome and beyond: What do pregnant women think? *Prenatal Diagnosis*, *35*, 598–604. https://doi.org/10.1002/pd.4579

van Schendel, R. V., Page-Christiaens, G. C. L., Beulen, L., Bilardo, C. M., de Boer, M. A., Coumans, A. B. C., … Henneman, L. (2016). Trial by Dutch laboratories for evaluation of non-invasive prenatal testing. Part II—Women's perspectives. *Prenatal Diagnosis*, *36*, 1091–1098. https://doi.org/10.1002/pd.4941

Verweij, E. J., Oepkes, D., & de Boer, M. A. (2013). Changing attitudes towards termination of pregnancy for trisomy 21 with non-invasive prenatal trisomy testing: A population-based study in Dutch pregnant women. *Prenatal Diagnosis*. (33), 397–399. https://doi.org/10.1002/pd.4063

Zehrer, T., Stankeviciene, I., & Abdel-Aal, M. (2015). *Comparison of National Downs Screening Uptake, Results, and Outcomes to a District General Hospital*.

SUPPORTING INFORMATION

Additional supporting information may be found online in the Supporting Information section at the end of this article.

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