



Monitor 2023

Prenatal screening: NIPT and structural ultrasound scan

*IQ health, commissioned
by RIVM-Centre for Population Screening*

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Conclusions

Figures

- In 93.6% of the 170,721 pregnancies, a counseling session was registered in the national database called Peridos (93.5% in 2022).
- NIPT participation was 67.8% and has increased compared to previous years (from 46.6% in 2019 to 57.8% in 2022).
- The proportion of pregnancies with the choice for secondary findings in NIPT, increased to 74.1% (69.6% in 2022).
- The proportion of abnormal NIPT was 0.5% and comparable to previous years.
- Participation in the 13 week scan has increased to 78.0% (75.5% in 2022).
- Participation in the 20 week scan has slightly increased to 86.6% (85.6% in 2022).
- The proportion of indications for abnormalities on the 20 week scan was 4.0% and has remained stable across years (4.0% in 2022).

Recommendations

- For the monitoring of the screening program (NIPT and structural ultrasound scan), data are required about findings after prenatal screening. This information, together with the outcome of the pregnancy, is necessary to determine the quality of the prenatal screening. However, these data are not available due to mainly legal bottlenecks. It is imperative to stress the importance of this data with all parties involved and to create the right conditions to make this data available for the prenatal screening program.

Prenatal screening: NIPT and structural ultrasound scan

Since 2007, the obstetric care provider offers information about the prenatal screening program to each pregnant woman in the Netherlands who wishes this, in the so-called **counseling session**. The purpose of prenatal screening is to provide pregnant women, who wish so, with timely information about the possible presence of one or more abnormalities in the unborn child, so that they can decide about different treatment options.



The **NIPT (Non-Invasive Prenatal Testing)** is a test to assess whether the baby may have Down syndrome, Edwards syndrome or Patau syndrome and is conducted from 10 weeks into pregnancy. For the NIPT, a blood sample is drawn from the pregnant woman. The pregnant women may opt for being informed about secondary findings. The blood is tested in a laboratory. In case of indications of any disorders, the pregnant women may choose to have follow-up invasive diagnostic testing. As of April 1 2023 the NIPT is a part of the regular prenatal screening program and available for all pregnant women for free, without having to participate in a scientific study.

The **20 week structural ultrasound scan** (20 week scan) is a test performed between week 18 and week 21 of pregnancy, to screen the foetus for physical abnormalities. Since September 1, 2021, the **13 week structural ultrasound scan** (13 week scan) has been offered in a scientific study around the 13th week of pregnancy to screen for physical abnormalities. If there are indications for abnormalities, the pregnant woman may choose for follow-up testing by means of advanced ultrasound examination (GUO) or genotyping. In this follow-up testing the suspicion of an abnormality may or may not be confirmed.

Participation counseling, NIPT, 13 week scan and 20 week scan

Counseling is offered to all pregnant women. Women decide for themselves whether they want to participate in a counseling session. Even after counseling, women can choose not to participate in the screening. In 2023, a counseling session was registered in the Peridos database for 93.6% (n=159,877) of the 170,721 pregnancies. The median age of the pregnant woman was 31 years. Participation rate of the NIPT was 67.8%. A 13 week scan was performed in 78.0% and a 20 week scan was performed in 86.6% of pregnancies. There are women who do not receive a 13 and/or 20 week scan, but are immediately eligible for a diagnostic scan (advanced ultrasound examination type 1, GUO-1), because they have an increased risk of a congenital abnormality in their child based on their history.

Table 1: Key figures, participation numbers and rates 2019-2023

| | 2019 | 2020 | 2021 | 2022 | 2023 |
|---|---------|---------|---------------------|---------|---------|
| Total number of pregnancies ¹ | 172,223 | 179,188 | 180,453 | 172,212 | 170,721 |
| Number of pregnancies with counseling session | 155,472 | 166,347 | 170,446 | 160,957 | 159,877 |
| Participation rate counseling (%) | 90.2 | 92.8 | 94.5 | 93.5 | 93.6 |
| Number of pregnancies with NIPT | 80,267 | 91,658 | 99,516 | 99,549 | 115,828 |
| Participation rate NIPT (%) | 46.6 | 51.2 | 55.1 | 57.8 | 67.8 |
| Number of pregnancies with NIPT and opt-in for secondary findings | 55,699 | 62,738 | 70,712 | 69,319 | 85,811 |
| Participation rate NIPT (%) and opt-in for secondary findings | 32.3 | 35.0 | 39.2 | 40.3 | 50.3 |
| Number of pregnancies with 13 week scan | - | - | 40,206 ² | 129,977 | 133,172 |
| Participation rate 13 week scan (%) | - | - | 69.32 | 75.5 | 78.0 |
| Number of pregnancies with 20 week scan | 149,260 | 154,751 | 154,691 | 147,441 | 147,799 |
| Participation rate 20 week scan (%) | 86.6 | 86.4 | 85.7 | 85.6 | 86.6 |

¹ The number of pregnancies is based on the data in the national database (Peridos). The participation rates are based on all pregnancies with a due date 6 months after the reporting year.

² The 13 week scan was introduced from September 1, 2021. The total number of pregnancies from September 1, 2021 to December 31, 2021 is 58,023. This is the denominator for the percentage of participation in the 13 week scan in 2021

Participation: trends over time and across age

Participation NIPT

NIPT was performed in 67.8% of pregnancies. In 50.3% of the pregnancies this included secondary findings, which is 74.1% of the total number of NIPT (*figure 1*). As age increases (up to and including 34 years), NIPT was performed more often (*figure 2*). In addition, there has been an increase in participation NIPT in all age groups and especially in the younger age groups, possibly due to the fact that NIPT became available to all pregnant women for free in 2023.

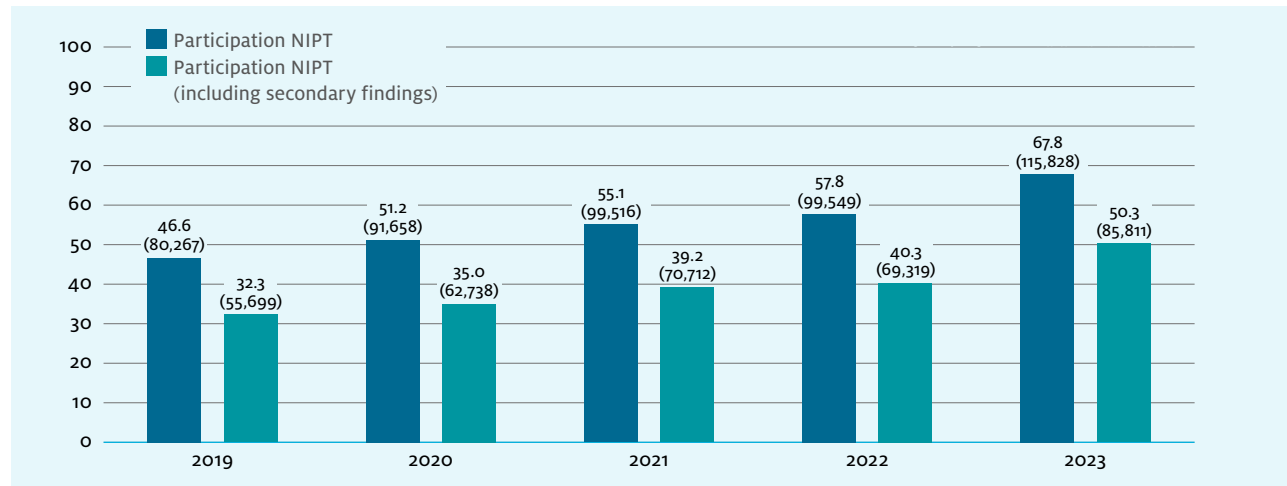


Figure 1 | Participation NIPT in 2019-2023, % and (n)

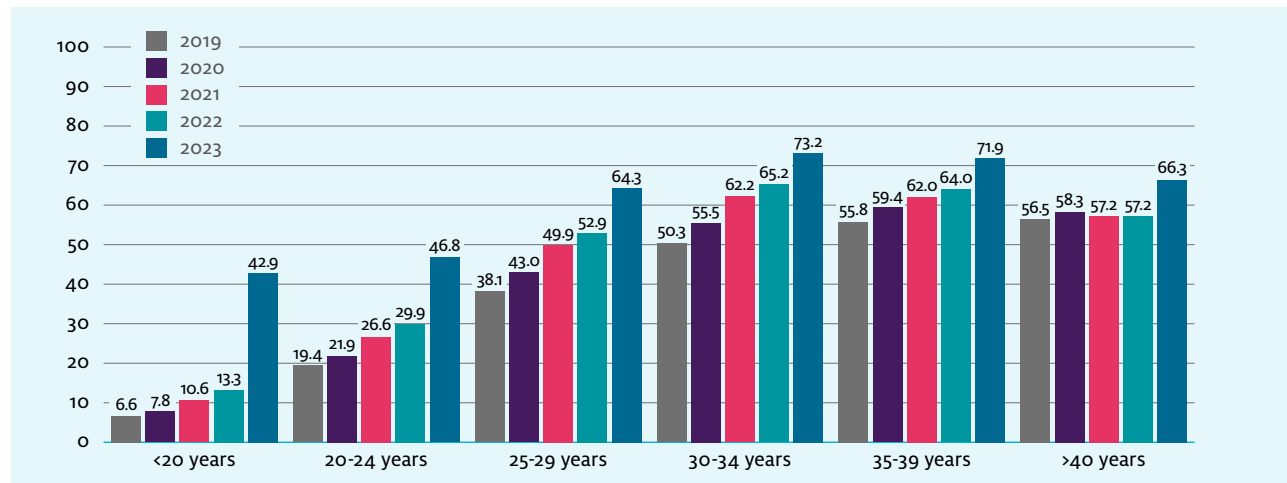


Figure 2 | Participation NIPT across age groups in 2019-2023, %

Participation: trends over time and across age

Participation 13 week scan and 20 week scan

In 2023 a 13 week scan was performed in 78.0% of pregnancies (*figure 3*). A 20 week scan was performed in 86.6% of pregnancies. *Figure 4* shows the trend in participation in the 20 week scan across years. In case no 13 week scan or 20 week scan was performed, a medical indication for a GUO1 may have been given, a miscarriage could have occurred, or the pregnant woman could have decided not to opt for a scan. In the national database (Peridos) there is no information available regarding this. As age increased as of 35 years, participation in a 20 week scan decreased slightly, possibly because older pregnant women more often had an indication for GUO1 and therefore did not receive a 13 week scan or a 20 week scan (*figure 5 and 6*).

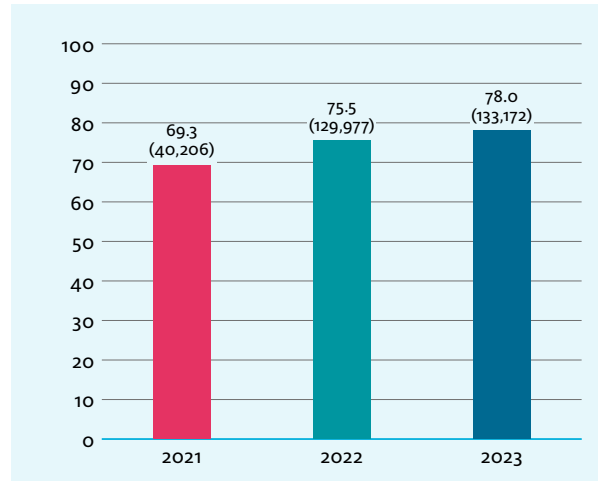


Figure 3 | Participation 13 week scan in 2021-2023, % and (n)

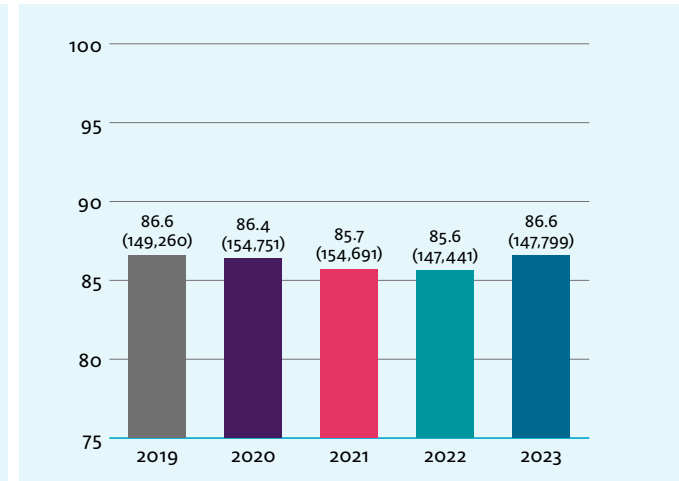


Figure 4 | Participation 20 week scan in 2019-2023, % and (n)

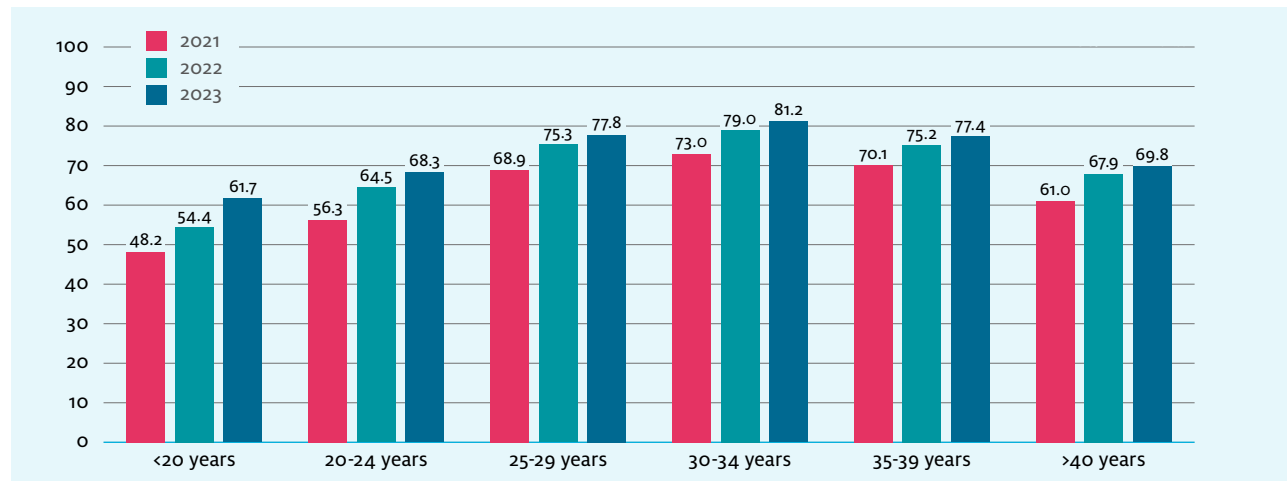


Figure 5 | Participation 13 week scan across age groups in 2021-2023, %

Participation: trends over time and across age

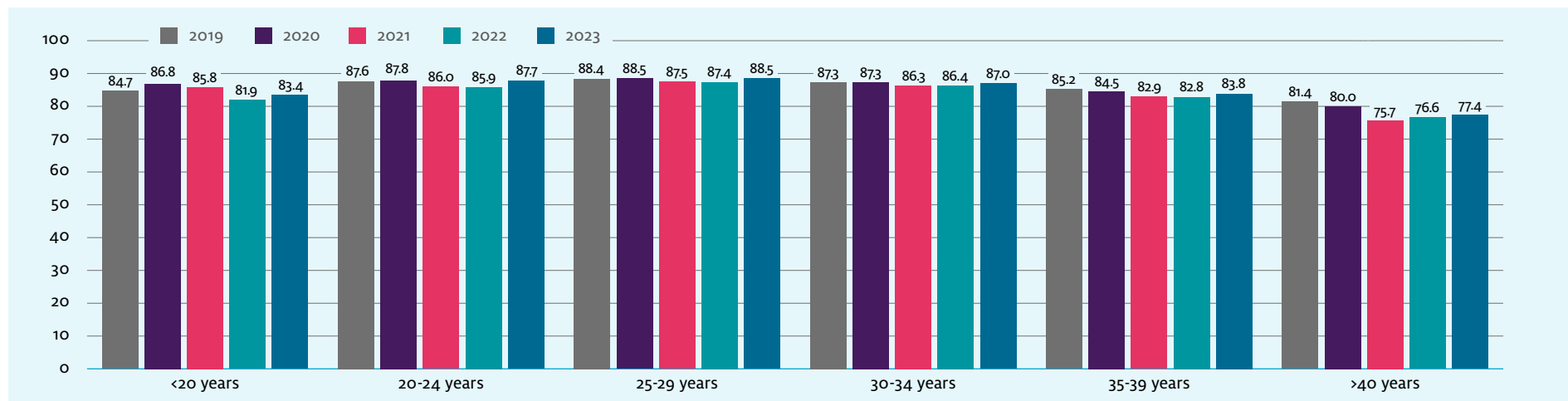


Figure 6 | Participation 20 week scan across age groups in 2019-2023, %

Participation: trends over time and across age

Participation repeat test NIPT and 20 week scan

Sometimes the NIPT does not produce a test result, for example because there is too little DNA from the foetus (placenta) in the pregnant woman's blood sample. In that case, a repeat test is needed. A repeat test for the NIPT was performed in 2.2% of pregnancies. In 0.2% of pregnancies two repeat tests were performed (figure 7). The participation in a repeat test after NIPT without a test result was 93.6%.

For a 20 week scan, a repeat test may be necessary if the child's position is for example unfavourable, which makes observation by the sonographer difficult. The proportion of pregnancies with a repeat test for a 20 week scan was 5.4% (figure 8). Repeat tests performed on the same day as the primary test are not recorded as such and therefore not included in this calculation.

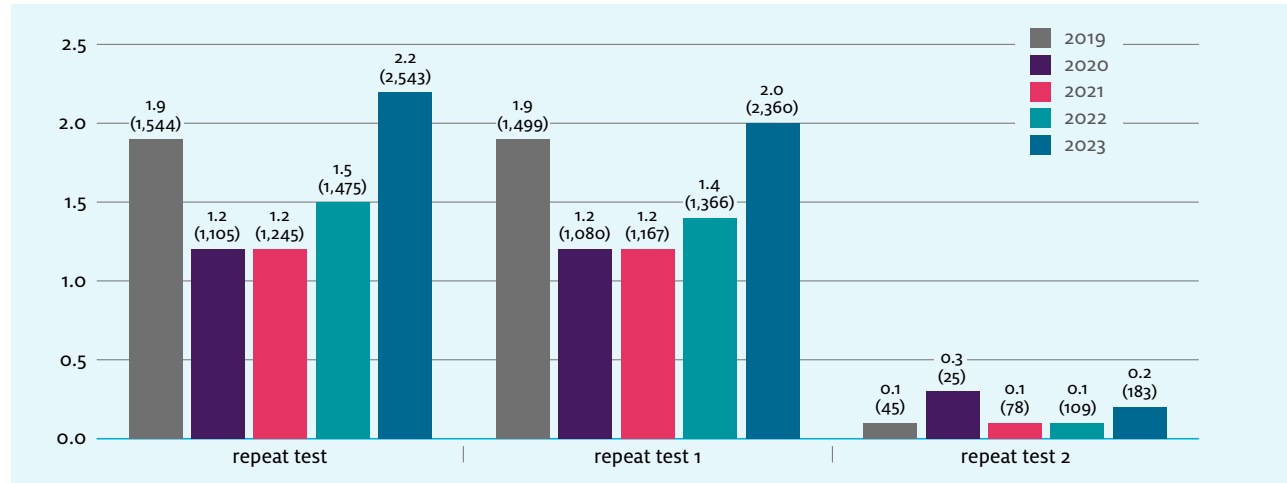


Figure 7 | Participation repeat test NIPT 2019-2023, % and (n)

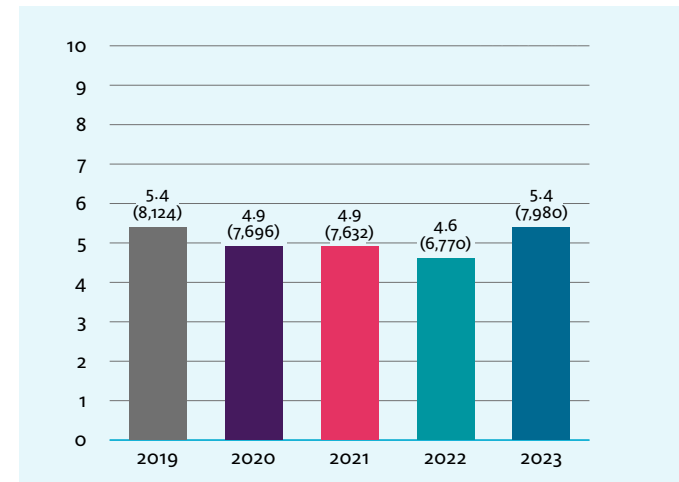


Figure 8 | Participation repeat test 20 week scan 2019 t/m 2023, % and (n)

Test results

Test results NIPT

Figure 9 presents the number and proportion of abnormal results of the NIPT. The proportion with an abnormal result for Down, Edwards and/or Patau syndrome was 0.51%. The proportion of pregnancies with an increased risk of Down syndrome (T21) was 0.32%. The percentage of pregnancies with an increased risk for Patau syndrome (T13) was 0.10% and for Edwards syndrome (T18) also 0.10%. The secondary finding rate is 0.46%.

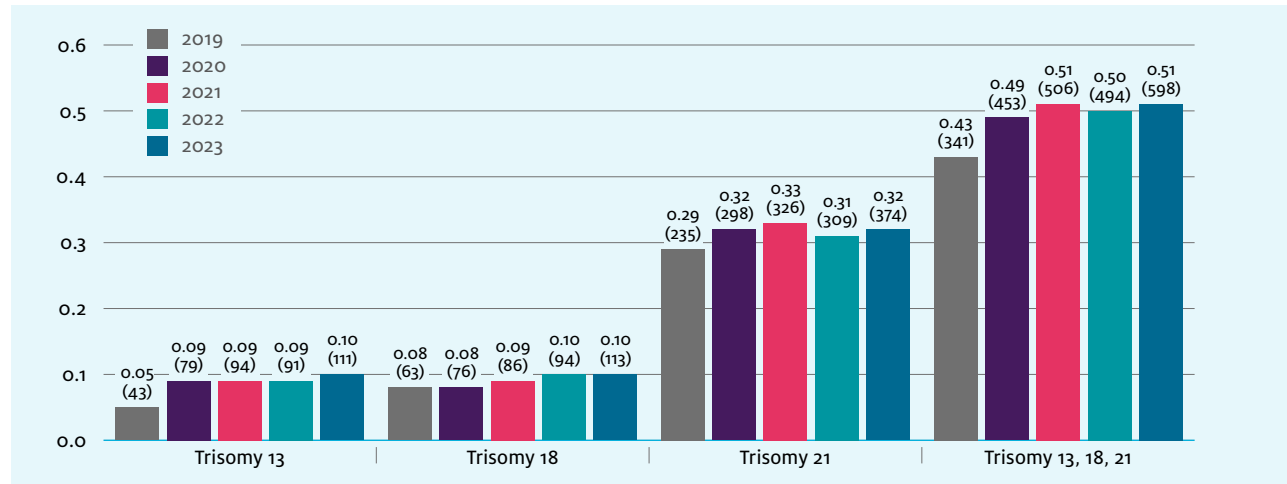


Figure 9 | Test results NIPT: pregnancies with an abnormal result in 2019-2023, % and (n)



Test results

Test results 13 week scan and 20 week scan

In 2023, 1.3% (n=1,713) of pregnancies had an abnormal 13 week scan. This is the first year of calculation and therefore no trends are available.

Figure 10 presents the abnormal results after a 20 week scan. An abnormal 20 week scan was present in 4.0% of pregnancies.

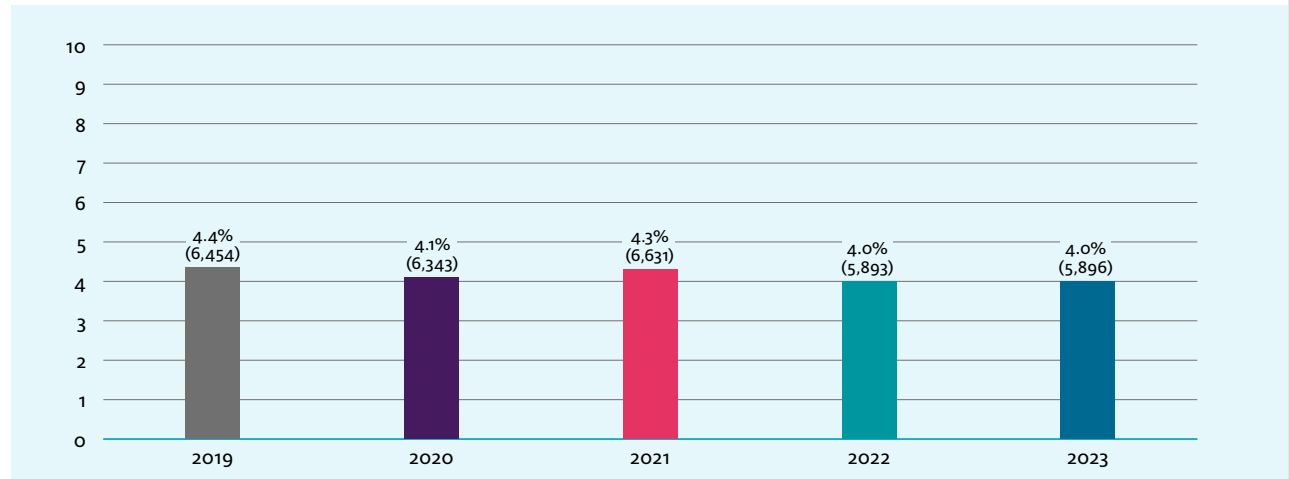


Figure 10 | Test results 20 week scan: pregnancies with an indication for physical abnormalities in 2019-2023, % and (n)



Time of counseling and screening

Counseling

The purpose of prenatal screening is to provide pregnant women with options. Offering timely counseling and testing is therefore crucial. Since 2017, there is no separate counseling session for different screening components, but one overall counseling session in which distinct options are discussed. In 2023, the counseling session took place before the 14th week in 98.2% of pregnancies, which is comparable with previous years (*figure 11*).

NIPT, 13 week scan and 20 week scan

NIPT was performed before the 19th week in 99.3% of pregnancies (*figure 11*). In 99.0% of pregnancies, a 13 week scan is performed before 14+4 weeks. The 20 week scan was performed before the 21st week in 97.8% of pregnancies.

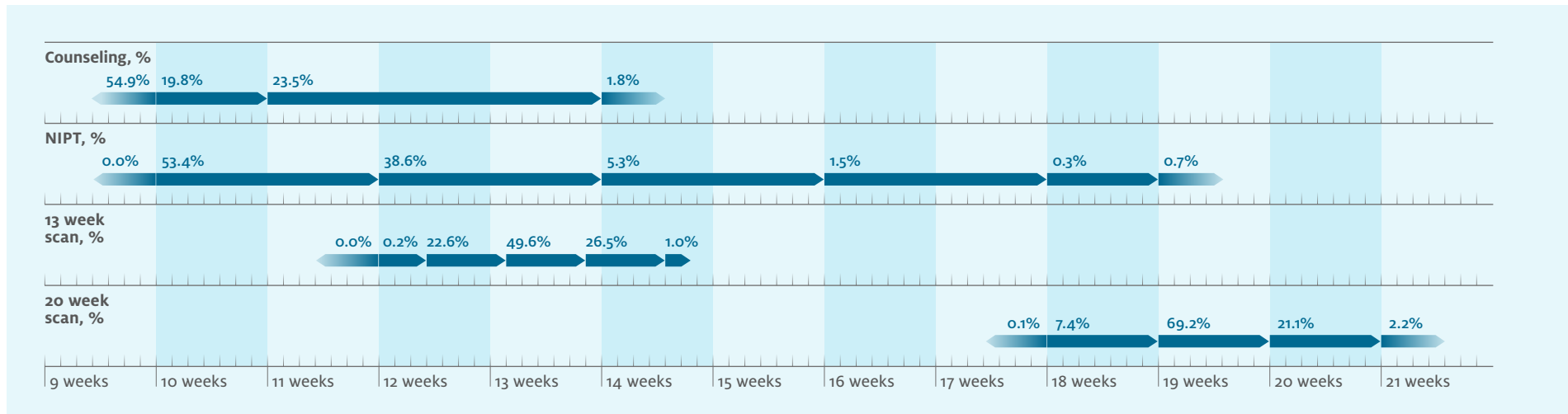


Figure 11 | Time of counseling, NIPT, 13 week scan and 20 week scan

About this monitor

This monitor describes the screening and test results of the NIPT, the 13 week scan and 20 week scan in the year 2023. On behalf of the Ministry of Health, Welfare and Sport, the Center for Population Research (CvB) of the RIVM is accountable for prenatal screening.

The monitor is a shortened version of the professionals' monitor, which is also publicly available (in Dutch). No data regarding follow-up diagnostics are available in this monitor. As a result, no genotyping and GUO indicators could be calculated. The monitor presents trends in indicators over the past five years (2019 to 2023). Some indicators have become (reliably) available later in time and are therefore presented over fewer years.

All indicators were calculated based on 2023 data recorded in the national database for prenatal screening (Peridos) as of September 16, 2024. This national database records data on prenatal screening in the Netherlands since 2010. All indicators were calculated at the level of individual pregnancies. Regarding indicators about outreach and participation, the total number of pregnancies,

based on the figures in the Peridos database, was used as the denominator. In previous years, figures from Statistics Netherlands were used to assess the number of pregnancies. The precise calculation of the indicators and details at regional level are reported in the professionals' monitor. Further details of indicators about time of counseling and 20 week scan can also be found in the professionals' monitor.

The Trident-2 study started on April 1, 2017, where pregnant women could choose for the NIPT as the first test, i.e., as an alternative to the combined test. As of April 1, 2023 the NIPT is a part of the regular prenatal screening program and available for all pregnant women for free without having to participate in a scientific study. In the professionals' monitor, the indicators are divided into two periods;

before and after April 1, 2023. The 13 week scan became available from September 1, 2021.

Through the efforts of all those involved, the data registry has improved over the years, both quantitatively and qualitatively. However, it is not (always) evident whether certain results of the monitor can be explained by registry issues and/or reflect actual changes in the screening program.