Prenatal Screening and pregnancy outcome

Update: February 2020

Prenatal screening for congenital anomalies

In the Netherlands a nationwide prenatal screening program was implemented in 2007. The prenatal screening program includes screening for trisomy 21, trisomy 18 and trisomy 13 through the Non Invasive Prenatal Test (NIPT) or combined test (CT) in the first trimester and screening for structural anomalies through an ultrasound scan around 20 weeks of gestation.

Since 2015 all women have to pay for the CT, while the anomaly scan is free of charge. In April 2014 the NIPT was introduced as an extra screening test for women who had an increased risk with the CT or because of a previous child with a trisomy. For these women the NIPT was reimbursed by the health care insurance. When the NIPT indicated an increased risk for trisomy 21, trisomy 18 or trisomy 13, an invasive test was performed for the final diagnosis. Since April 1st 2017, the NIPT is available as first tier screening test for Down, Edwards and Patau syndrome, next to the CT. Women who opt for the NIPT as a first tier screening test have to pay €168, this is about the same as the costs for the CT.

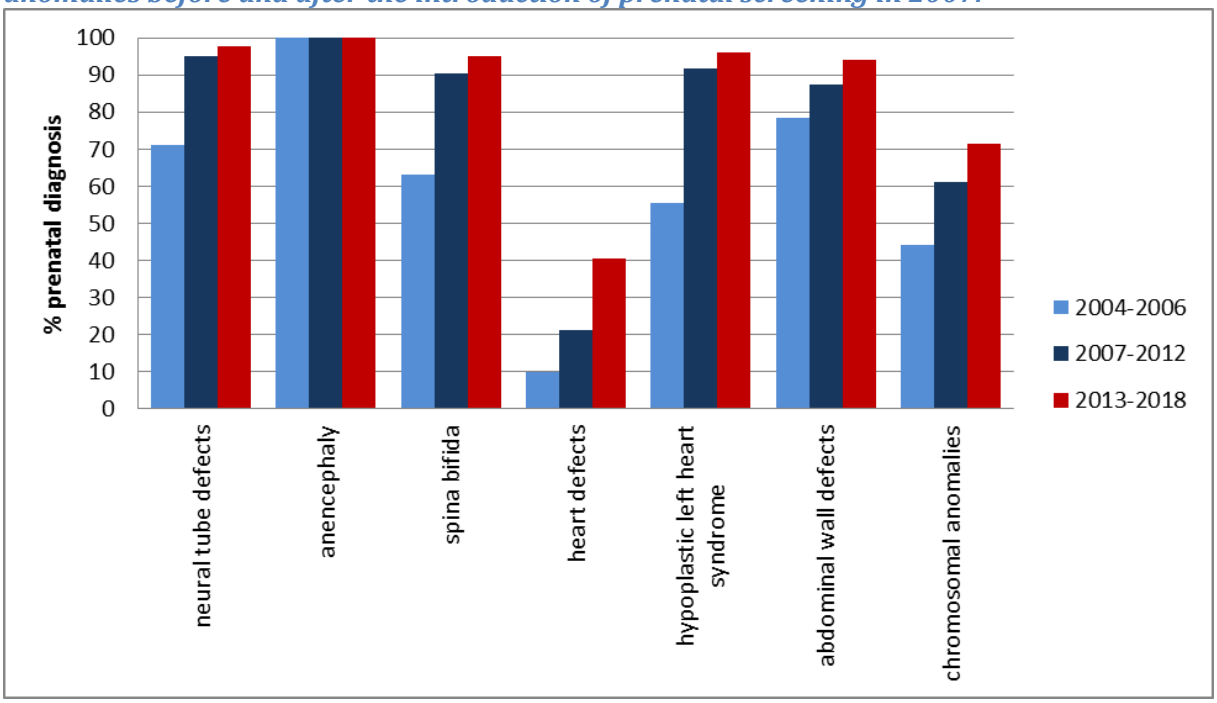
The prenatal screening program is monitored annually by the regional centers for prenatal screening. The monitoring report (in Dutch) can be found at the website of the RIVM. In 2018 in Northeast Netherlands (exceeding the Eurocat NNL region), first trimester screening for Down, Edwards and Patau (DEP) syndrome was performed in 6,618 pregnancies through the NIPT and in 541 pregnancies through the CT. The uptake was 29.3% for the NIPT and 2% for the CT. NIPT resulted in 0.6% in a positive result (i.e. indication that fetus is affected with Down, Edwards or Patau syndrome). In 4.9% of the CT an increased risk for Down, Edwards or Patau syndrome was found.

An anomaly scan was performed in 20,874 pregnancies, corresponding with an uptake after counseling of 82%. An anomaly was suspected in 4.7% of pregnancies.

Prenatal diagnosis of congenital anomalies

Since the introduction of prenatal screening an increase in proportion prenatal diagnosis for several congenital anomalies is observed (see figure 1). For certain anomalies the increase in prenatal diagnosis continued in the most recent years (2013-2018). The proportion of prenatally diagnosed spina bifida cases increased from 63% before 2007 to 90% in period 2007-2012 and further to 95% in 2013-2018. For heart defects the prenatal diagnosis proportion increased to 41%, whereas for hypoplastic left heart the increase was observed in the period immediately after the introduction and remained more or less stable in the most recent time period.

Figure 1. Proportion of prenatally detected congenital anomalies for selected congenital anomalies before and after the introduction of prenatal screening in 2007.



Prenatal diagnosis of common trisomies

Prenatal diagnosis of common trisomies increased after the introduction of the prenatal screening program in 2007. Since the NIPT became available as a second screening test for trisomy 21, trisomy 18 and trisomy 13 in 2014 and as first screening test in 2017, the prenatal diagnosis rate showed an increase for trisomy 21 and trisomy 18, but not for trisomy 13, see table 1. However, numbers are low in particular for Patau syndrome.

Table 1. Number of cases and proportion prenatally detected for common trisomies before and after introduction of prenatal screening in 2007

	Before screening 2004-2006		After screening 2007-2013		After screening (NIPT available) 2014-2018	
Common trisomy	Total (n)	prenatally diagnosed	Total (n)	prenatally diagnosed	Total (n)	prenatally diagnosed
Trisomy 21, Down	91	40.7%	228	54.8%	161	65.8%
Trisomy 18, Edwards	38	78.9%	98	89.8%	40	98.2%
Trisomy 13, Patau	7	71.4%	31	96.8%	14	87.0%
All common trisomies	136	52.9%	357	68.1%	185	75.3%

In 2018, 85% of the common trisomies were prenatally diagnosed. For Down syndrome, 23 cases (77% of the total number of Down syndrome cases in 2018) were prenatally diagnosed of which about half (n=12, 52%) after a positive NIPT, 6 after abnormal findings at an US in the first trimester and 5 after abnormal findings at an US in the second trimester or later in pregnancy.
For Edwards syndrome all 14 cases were prenatally diagnosed: 4 (29%) after a positive NIPT, 8 (57% after abnormal findings at US in the first trimester or an increased risk at CT and 2 after abnormal findings at US in second trimester. For Patau syndrome, 7 of the 8 cases of Patau syndrome were prenatally diagnosed, either after abnormal US findings in the first trimester (n=2) or in the second

trimester (n=5). In total, for 36% of the common trisomies that were diagnosed prenatally, NIPT was the first positive prenatal test NIPT and 64% had abnormal findings at US as a first indication.

Pregnancy outcome after prenatal diagnosis

The number of terminated pregnancies because of a congenital anomaly is presented in table 2. The data are presented for a selected number of anomalies, separately for the three-year period before prenatal screening (2004-2006) and for two periods (2007-2012 and 2013-2018) after the introduction of prenatal screening on January 1st, 2007.

In table 2 the groups of neural tube defects, heart defects and abdominal wall defects are presented excluding chromosomal and monogenic disorders and deletions. This results in a group of so-called 'non genetic' anomalies. Table 2 shows that terminations of pregnancy increased significantly for some congenital anomies, e.g. neural tube defects, trisomy 18 and heart defects. Among heart defects, for which terminations are relatively rare, specifically the hypoplastic left heart syndrome resulted in more terminations after introduction of prenatal screening. This increase seems to be relatively stable in the second period after the introduction of prenatal screening.

Table 2. Number and proportion of terminated pregnancies for a selected number of congenital anomalies (non genetic) and chromosomal anomalies before and after introduction of prenatal screening in 2007

Anomaly groups	Before screening (2004-2006)			After screening (2007-2012)			After screening (2013-2018)		
	total	n	%	total	n	%	total	n	%
Neural tube defect	38	17	44,7%	84	61	72,6%	87	71	81,6%
- Spina bifida	30	10	33,3%	42	25	59,5%	39	31	79,5%
- Anencephaly	7	6	85,7%	39	34	87,2%	41	36	87,8%
Heart defect	365	8	2,2%	676	49	7,2%	563	49	8,7%
- Hypoplastic left heart syndrome	18	3	16,7%	36	19	52,8%	26	15	57,7%
Abdominal wall defect	14	3	21,4%	40	14	35,0%	52	20	38,5%
Chromosomal abnormality	212	62	29,2%	522	230	44,1%	428	217	50,7%
- Trisomy 21	91	33	36,3%	208	81	38,9%	181	86	47,5%
- Trisomy 18	39	17	43,6%	87	68	78,2%	67	56	83,6%
- Trisomy 13	7	4	57,1%	28	18	64,3%	26	15	57,7%