Effect of a national screening program based on the 20 weeks scan on prenatal detection and outcome of selected severe congenital anomalies

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Objective
The aim of the study was to determine time of diagnosis and pregnancy outcome in open spina bifida, anencephaly, abdominal wall defects (gastroschisis and exomphalos), severe congenital heart defects and cleft lip (with or without cleft palate) in the period after the introduction of a national prenatal screening program in 2007, and compare these results to the period prior to this introduction.

Methods
Data from 2 of the 8 Dutch regional centers for prenatal screening located in the North-East (Groningen) and the North-West (Amsterdam) was collected. Findings at screening scan as well as confirmation at referral centers and pregnancy outcome were collected prospectively. Data from birth defect registries (Eurocat Northern Netherlands and the Dutch Association for Cleft Palate and Craniofacial Anomalies) and the Netherlands Perinatal Registry and surgery lists were used to complement and validate the case lists.

Results
After introduction of the screening program in 2007 the detection rate for open spina bifida increased by more than 40%, from 50 to almost 90%. Pregnancy terminations increased by %, but this did not lead to significantly fewer children surviving the neonatal period, due to a decrease in perinatal deaths. Evaluation of the timing of detection of anencephaly showed that about 70%of cases were diagnosed at first trimester scans and the remaining 30% at the 20-week scan. Pregnancy termination in this lethal anomaly occurred in over 90% of cases. Detection rate for cleft lip was less than 60% before the introduction of the 20-week scan and increased to over 90% of the cleft lips cases (with or without additional anomalies) diagnosed before 24 weeks’ gestation, thereafter. No change in overall live birth rate, nor increase in terminations of pregnancy was observed. Prenatal detection rate for abdominal wall defects increased from about 70% to 95%, with 2/3 of the cases diagnosed before the 20-weeks. Associated anomalies are far more common in exomphalos with a 4-fold lower survival rate than gastroschisis. With increased prenatal detection the pregnancy termination rate has doubled in exomphalos and in gastroschisis. Prenatal detection rate for severe congenital heart defects (CHD) with an abnormal 4 chamber view increased from 35% to 89%, and for cases with a normal 4 chamber view from 14% to 54%. Pregnancy termination has increased by almost 50% in the former group, while no significant increase has occurred in the latter (from 1% to 7%).

Conclusion
After the introduction in 2007 of a national prenatal screening program the prenatal detection rates for all congenital anomalies has increased considerably. Especially anencephaly, gastroschisis and exomphalos are amenable for early detection (in the first trimester). Increased prenatal detection did not influence the number of live born children with open spina bifida, cleft lip and severe CHD with a normal four-chamber view. It did reduce the number of live born children with gastroschisis, exomphalos and severe CHD with an abnormal 4-chamber view.