Yield of early vs 20 weeks US investigation for the diagnosis of congenital structural anomalies
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Objective
The uptake of the combined test in the Netherlands is notoriously low, not exceeding 30%. The test is presented mainly as a trisomy screening and women are not aware of the possibility of early diagnosis of severe congenital anomalies. In contrast, over 90% of women choose for a 20 weeks scan. Irrespective of the low CT uptake increasingly more structural anomalies are diagnosed early in pregnancy, but outside a systematic screening offer. Recently the ministry of health has suggested that all Dutch women should be informed about the cell free fetal DNA, owing to its superiority as screening for aneuploidies. The possible disappearance of an early trimester ultrasound could seriously affect the percentage of early diagnosed anomalies, with detrimental effect with respect to the advantage of early termination in case of severe anomalies. Aim of this study was to assess the performance of a early anatomical survey of the fetus at 12-13 weeks gestation with respect to the traditional 20 weeks scan.

Methods
Within the Dutch population screening act a licence was granted by the Health Council for performing this study implying a new "early screening scan". The study took place between november 2012 and december 2015. Women undergoing a CT or a early scan in view of an increased risk of anomalies in the North-East of the Netherlands (Groningen area) were asked to participate into the study prospectively. Only scans performed by ultrasonographer with a NT diploma and where the early fetal anatomy had been investigated were used for the analysis. Data on number of increased NT (in those opting for the CT), suspected anomalies, including early markers for abnormal development, supplemented by the results at karyotyping or at advanced scans in referral centers, were collected in early pregnancy and at the traditional 20 weeks scan. Data on termination of pregnancy, IUD, NND and pregnancy outcome were collected for the total population.

Results
Information were available for 5532 pregnancies: 5390 seen at the time of the CT or at an early anomaly scan in view of an increased a-priori risk of anomalies and 142 were early referrals for suspicion of anomalies in the first trimester. Early scans were performed at 9-17 weeks (mean 12, 6 weeks). Of the 218 (SPR 0, 4%) abnormal findings (including isolated 1st trimester markers and increased NT) at early trimester scans, anomalies were confirmed in 143 (66%), giving a false positive rate of 0, 1%. The NT was increased in 30% of the cases with structural anomalies. In 11 of the referrals for suspicion of anomalies (5%) the anomaly was not confirmed or disappeared within 2 weeks, leading to change of the final diagnosis (omphalocele, megacystis, very large NT, gastroschisis). In the cohort there were in total 8 IUD, 57 Top and 4 NND. Parents choose for TOP in about 60% of the early diagnosed anomalies. At the 20 weeks scan 36 additional structural anomalies were diagnosed. Follow-up is still being completed.

Conclusion
Even in a country without a policy of early anatomical fetal assessment, scans performed by certified ultrasonographers lead to a high detection of the majority of severe anomalies < 18 weeks. Of all the anomalies detected at 1st and 2nd trimester US, the majority, over 75%, are detected already in the late first trimester. This allows for early and less traumatic TOP in the majority of severe and lethal anomalies.