Impact of ultrasound screening in timing of detection and prevalence of anencephaly in the Netherlands

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Objective
To assess the prevalence and timing of prenatal detection of anencephaly in the Netherlands during a five year period.

Methods
A retrospective cohort study was undertaken in the Northwest and the Northeast of the Netherlands. A case-list of all pre-and postnatally diagnosed cases of anencephaly with or without additional anomalies was compiled. Cases were included if the estimated due date was between 1st of August 2008 and 31st of July 31 2013.

Results
During the study period 110 cases of anencephaly were diagnosed. Overall prevalence of anencephaly was 5.4 per 10,000 pregnancies. The majority of the cases (60%) was detected before 14 weeks gestation. The detection rate before 14 weeks of pregnancy was significantly higher in the Northwest compared to the Northeast. (p=.003). During the study period, gestational age at the time of detection did not change significantly (p=.579). The percentage of cases with additional (chromosomal) anomalies was 24.5%.

Conclusion
The majority of the anencephaly cases in this study was diagnosed prior to the mid-trimester scan. Early detection requires that scans are performed by sonographers trained and experienced in recognizing fetal anomalies in the first trimester. Pregnant women should be informed that an ultrasound scan in early pregnancy can reveal serious anomalies like anencephaly.