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Artificial intelligence to detect increased nuchal translucency in the first trimester of pregnancy

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Objective

Many fetal anomalies can already be diagnosed by ultrasound in the first trimester of pregnancy. Unfortunately however, in clinical practice, detection rates for anomalies in early pregnancy are low. Our aim was to use an automated image segmentation algorithm to detect one of the most common fetal anomalies: a thickened nuchal translucency (NT), which is a marker for genetic and structural anomalies.

Methods

Standardized mid-sagittal ultrasound images of the fetal head and chest were collected for 560 fetuses between 11 and 14 weeks of pregnancy, 88 (15.7%) of whom had an NT thicker than 3.5mm. Image quality was graded as high or low by two experts. Images were divided in a training- (n=451, 55 thick NT) and a test-set (n=109, 33 thick NT). We then trained a U-Net convolutional neural network to segment the fetus and the NT region, and computed the ratio of these regions. The ability of this ratio to separate thick (anomalous) NT regions from healthy, typical NT regions was first evaluated in ground-truth segmentation to validate the metric, then with predicted segmentation to validate our algorithm, both using the area under the receiver operator curve (AUROC).

Results

The ground-truth NT: fetus ratio detected thick NTs with 0.97 AUROC in both the training and test sets. The fetus and NT regions were detected with a Dice score of 0.94. The NT: fetus ratio based on model segmentation detected thick NTs with AUROC of 0.96. At a 91% specificity, 94% of thick NT cases were detected (sensitivity) in the test set. The detection rate was statistically higher in high vs. low quality images (AUROC 0.98 vs 0.90, respectively).

Conclusion

Our model provides an explainable deep-learning method for detecting an increased NT. This technique can be used to screen for other fetal anomalies in the first trimester of pregnancy.