Is there an association between increased nuchal translucency and autism?

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Objective
To investigate the association between nuchal translucency thickness (NT) and autism spectrum disorders (ASDs) in chromosomally normal children.

Methods
The study included all singleton infants born in Denmark, who had routine first trimester screening between January 1st 2008 and March 31st 2012. Children were divided into three groups by NT thickness: NT > 99th percentile (group 1), NT 95th-99th percentile (group 2), and NT < 95th percentile (group 3). Follow-up was undertaken for all children at 2 to 6 years of age, depending on date of inclusion. We obtained information on diagnoses of ASDs (childhood autism (CA), Asperger’s syndrome, and atypical autism) from the Danish Psychiatric Central Registry, a nationwide register of inpatient and outpatient contacts to psychiatric departments. Logistic regression was used to estimate the odds ratio (OR) of ASDs for children in groups 1 and 2 compared to those in group 3.

Results
Of the 222,966 liveborn children meeting the inclusion criteria of NT scan at fetal CRL 45-84 mm, 222,507 had no known chromosomal anomaly. They were distributed with 642 (0.3%) in group 1, 4,760 (2.1%) in group 2, and 217,105 (97.6%) in group 3, respectively. 706 cases of ASDs were identified, 5 (0.7%), 15 (2.1%), and 686 (97.2%) in groups 1, 2 and 3, respectively. We found a small but statistically significant correlation between NT > 99th percentile and all ASDs (OR 2.48; 95% CI, 1.02 – 5.99). There was no correlation between NT 95th -99th percentile and ASDs (OR 1.00; 95% CI, 0.60 - 1.66). The estimated risk of childhood autism (CA) alone was statistically non-significant for both groups 1 and 2 (OR 1.82; 95% CI, 0.45 – 7.32; OR 1.10; 95% CI, 0.57 – 2.14) based on 2 and 9 cases, respectively.

Conclusion
First trimester NT > 99th percentile is associated with a moderately increased risk of ASDs, when compared to NT < 95th percentile. We found no association between increased NT and CA alone. Estimates for both ASDs and CA were, however, based on a small number of cases and warrants further follow-up.