Increased nuchal translucency with normal karyotype
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Objective
Prior research has demonstrated that an enlarged nuchal translucency (NT) on fetal ultrasound can be associated with aneuploidy and many different genetic syndromes. Yet there is no clinical guideline for further genetic testing after a normal karyotype. Our objective was to determine the series of investigations that can be compiled by 14 weeks of gestation if we have an enlarged nuchal translucency with normal karyotype.

Methods
We give details about five cases of patients who had a sonographic assessment of nuchal translucency (NT) thickness with karyotypically normal fetuses.

Results
The average age of patients was 31 years. The average size of the neck was 3.76 mm. Fetal karyotype made by trophoblastic biopsy for five patients were normal. One patient had cystic hygroma (CH) on fetal ultrasound. Morphological examination 14SA: Persistence of increased nuchal translucency for one patient, wave was negative on Arantius for two patients. No identifiable heart abnormality, diaphragmatic hernia, exomphalos, or Megacystis. A medical abortion has been accepted for one patient. Pathologic studies were without anomalies.

Conclusion
In euploid fetuses, the prevalence of fetal abnormalities and adverse pregnancy outcome increases exponentially with NT thickness. However, the parents can be reassured that once the presence of aneuploidy is ruled out and the fetus is shown to be normal. The vast majority of fetal abnormalities associated with increased NT can be diagnosed by a series of investigations such as comparative genomic hybridization array (CGH-array).