Exome sequencing improves the genetic diagnosis in fetuses with increased nuchal translucency.

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Objectives
The purpose of this study was to investigate the value of exome sequencing in fetuses with increased nuchal translucency (NT).

Methods
The study comprised 356 singleton pregnancies with increased fetal NT and chromosomal microarray analysis (CMA) results. Six of them have performed fetus–mother–father trio exome sequencing.

Results
The number of normal CMA results was 249. There were fourteen cases with pathogenic copy number variants (CNV). Total number of cases with variants of unknown significance (VOUS) CNV was nine. In fifty-two fetuses with NT≥5.5mm, three cases (3/52, 5.8%) with pathogenic CNV could be detected by CMA while five cases (5/52, 9.8%) with pathogenic mutations could be detected by exome sequencing.

Conclusion
In the investigation of fetuses with increased NT, exome sequencing should be considered in fetuses with negative CMA findings. The limitation of this study is the small sample size. The larger prospective study is needed.

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