Pregnancy outcome of fetuses with increased nuchal translucency but normal karyotype and CMA

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Objective
To investigate the pregnancy outcome of fetuses with nuchal translucency (NT) ≥3.5 mm but normal karyotype and CMA.

Methods
A retrospective population-based cohort study was performed. All patients referred to our institution for nuchal translucency ≥3.5 mm were included in the study. We proposed a fetal karyotype by invasive testing to all patients. We followed prenatally all patients until delivery; the pregnancy outcome was also recorded and defined as adverse (termination of pregnancy [TOP], miscarriage [MC], or delivery of a child with structural defects or genetic disorders), or favourable (delivery of a child without any structural defects or genetic disorders diagnosed before being discharged).

Results
From October 2012 to November 2016, we identified 74 fetuses with NT ≥ 3.5 mm with normal karyotype and CMA. Two additional cases with normal karyotype but pathological CNV were excluded from the study. The mean maternal age was 33 years. An adverse perinatal outcome was observed in 27% of cases. In 3 cases (4%) a MC was observed and in one case a premature delivery at 25 WG with post-natal death was observed. The ultrasonographic follow-up showed 6 cases of major congenital heart diseases (CHD) and 4 cases of minor CHD. Further 4 cases of major anomalies were detected at ultrasonographic follow-up including diaphragmatic hernia, Dandy Walker malformation associated with skeletal dysplasia, hydronephrosis and discordance anomaly in monochorionic twins.

Conclusion
Adverse pregnancy outcome increases with NT ≥ 3.5 mm, even with normal karyotype and CMA, as in one third of cases a congenital malformation or a MC was observed.