Objective
The purpose of this study was to evaluate the outcomes of pregnancies with nuchal translucency (NT) greater or equal to 3 mm at the routine first trimester screening. Recently, the association between increased fetal NT at 11–14 weeks' gestation and chromosomal abnormality, congenital cardiac diseases, fetal structural defects, and genetic syndromes has been well established.

Methods
A total of 1490 pregnant women were assessed for first trimester ultrasonography. They had routinely measured crown-rump length (CRL) and nuchal translucency (NT) for screening for Down syndrome between 11 and 14 weeks of gestation. Cases with a NT≥3 mm were counselled further regarding the risk of chromosomal abnormality and prenatal diagnosis by fetal karyotyping. A complete follow-up was obtained in all cases by a review of medical records.

Results
Using a cut-off value of 3 mm, the prevalence of increased fetal NT was 0.74% (n=53). The fetal karyotype was available in all cases from first-trimester chorionic villus sampling or second-trimester amniocentesis. Among the 53 cases, there were fourteen (26.4%) chromosomal abnormalities: six trisomy 21, three trisomy 18 and five Turner’s syndrome. All of these fourteen women chose to terminate the pregnancy. Of the 39 chromosomally normal pregnancies, five resulted in fetal demise (two intrauterine death and three termination of pregnancy due to congenital heart diseases). The remaining 34 pregnancies resulted in live births, including one gestational hypertension and one preterm delivery, respectively. The total incidence of an adverse outcome in the group of increased fetal NT was 39.6%.

Conclusion
The results of this study showed the association between fetal NT and adverse pregnancy outcomes. In a routine population with first-trimester ultrasonography where fetal NT measured greater than or equal to 3 mm, it was associated with a poor pregnancy outcome, mainly chromosomal abnormalities and congenital cardiac diseases. In addition, this study also demonstrated the necessity for fetal assessment and follow-up in cases where fetal NT is increased in the first trimester.