

## عنوان مقاله:

Specificity and Effectiveness of NIPT Screening Test in Pregnant Mothers in Fars Province

## محل انتشار:

اولین کنگره بین المللی پریناتالوژی، هفتمین کنگره سراسری پریناتالوژی و نئوناتالوژی (سال: 1398)

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## نویسنده:

Sara Senemar

## خلاصه مقاله:

**Introduction:** Genetic anomalies are major causes of abortions, stillbirths and neonatal mortalities. The prevalence of congenital anomalies of these disorders is 2-3% in Iran. Prenatal diagnosis means identifying the structural or functional abnormalities in the fetus. Trisomies are responsible for about 25% of abortions and 4% of stillbirths. Trisomy 21, 18 and 13 are among the most common karyotypic abnormalities in Iran. The current study, the most recent non-interventional embryo screening method for NIPT testing is reviewed. **Materials and methods:** This is a diagnostic study that was performed in a retrospective form. Participants in this study are 389 pregnant mothers with gestational age of 11 to 18 weeks who were referred to the Academic Center for Education, Culture and Research of Shiraz during the period from May 2017 to December 2018 for NIPT test. 10 ml of venous blood was taken for test. Samples were sent to Singapore to perform the NIPT test. We've received the result of NIPT test after 2 weeks. The aim of this study was to evaluate the efficiency of NIPT test in diagnosis of trisomy 21, 18 and 13. The results were reviewed and analyzed. **Results:** One case (0.26%) due to high maternal weight (105 kg) failed to provide a reliable result and was excluded from the study. Among the studied pregnancies, the mean of the mother (range of 17- 46 years old) and the mean of gestational age were 15.91 (range 12-28 weeks). Most of women (53.2%) were more than 34 years old and most samples (67.8%) were collected up to 16 weeks. Out of 388 blood samples of NIPT pregnant mothers, 11 sample (2.84%) were positive, including 8 sample for trisomy 21, 1 sample for trisomy 18, and 2 sample for trisomy 13. Two positive cases of NIPT (both trisomy 21) were aborted before amniocentesis (self-induced abortion), but the rest of the embryos with a disorder were subjected to an amniocentesis test and the karyotype of the disease was proven. The follow-up of 377 remained women with negative NIPT test results revealed no health problem in neonates after delivery. **Discussion:** The results of this research showed a high accuracy of NIPT testing. Due to the high diagnostic value, it can be replaced with more invasive procedures such as amniocentesis and CVS for having complications such as miscarriage, leakage of amniotic fluid, etc and can be used for high-risk pregnant women.

## کلمات کلیدی:

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