PrenaTest®

lifecodexx

non.invazif
moleküler.genetik
prenatal.tanı testi

PrenaTest®

- Anne ve bebek için herhangi bir risk taşımayan, maternal kandan Serbest Fetal DNA analizi ile trizomi 21, 18 ve 13'ün non-invazif olarak prenatal teşhisi artık mümkün olabilmektedir.

- Amerikan Kadın Doğum Derneği (**ACOG**), Maternal Fetal Tıp Derneği (**SMFM**) ve Dünya Prenatal Tanı Derneğinin (**ISPD**), Serbest Fetal DNA hakkındaki ortak komite görüş raporlarına göre; aşağıda listelenen durumlarda bu testlerin önerilebileceği bildirilmiştir:

  - **35 yaş ve üzerindeki anne adayları**
  - **Ultrasonda kromozomal anomali riskini işaret eden bulgular**
  - **Önceki gebelikte trizomi hikayesi**
  - **İkili, üçlü, dörtlü veya entegre tarama testlerinden birinde kromozomal anomali riskinin yüksek saptanması**
  - **Ebeveynlerde dengeli translokasyonların varlığı**

- LifeCodexx laboratuvarında yapılan klinik validasyon çalışmaları ile, **PrenaTest® CE – IVD onayı alan tek non-invazif prenatal tanı testidir;**

  - Yeni nesil DNA dizileme (NGS) cihazı İllumina HiSeq 2000 ile massif paralel sekanslama (MPS) yapılmakta ve **PrenaTest® DAP.21 plus** biyoinformatik yazılımı ile değerlendirilmektedir.

  - Serbest fetal DNA'yı koruyan özel kan tüpleri ile (Cell-Free DNA BCT™) ile **≥%99 sensitivite ve ≥%99,8 spesifiteye ulaşılmıştır.**

centro laboratuvarları

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Actual aspects of non-invasive prenatal detection of fetal aneuploidies using massively parallel sequencing

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Objectives:
Non-invasive prenatal detection of fetal aneuploidies using cell-free DNA from maternal plasma has been shown to be feasible by massively parallel sequencing (MPS) in high risk pregnancies as well as in general pregnancy populations. We report our results of a large-scale European clinical study in a high risk group and compare the results with studies performed with general risk groups. The aim of these studies is to validate the diagnostic accuracy of the non-invasive prenatal diagnostics based on MPS for detecting fetal aneuploidies.

Methods:
In our own study maternal blood samples were collected from a total of 517 pregnant women with high risk for aneuploidy prior to invasive prenatal procedures at different clinics located in Germany and Switzerland. The extracted maternal plasma DNA was analyzed using Illumina sequencing platform in a multiplexed fashion. For data analysis a z-score equation was used to distinguish samples with fetal aneuploidies from samples with a set of normal fetal chromosomes. The results of MPS analysis were compared with the fetal karyotype obtained from conventional cytogenetic analysis of chorionic villi, amniocytes or fetal lymphocytes. Furthermore, a new bioinformatics algorithm based on GC normalization (PraenaTest® DAP.plus) was tested for detection of trisomy 13 and 18.

Results:
Overall, 39 out of 40 trisomy 21 samples were correctly classified (sensitivity 97,5%; one-sided lower confidence limit [CL]: 88.7%) and 427/427 samples were correctly classified as trisomy 21-negative (specificity 100%; one-sided lower CL: 99.3%). Bioinformatics analysis using a new algorithm detected additionally all cases of trisomy 13 (n=5) and trisomy 18 (n=8). The overal detection rate for the most common aneuploidies 13, 18 and 21 was 98,11% **Sensitivity and specificity rates of >99% were recently reported also in studies with patients from the general pregnancy population.**

Conclusions:
MPS of maternal plasma DNA is an accurate diagnostic test for non-invasive detection of fetal aneuploidies 13, 18 and 21. **The implementation of the new non-invasive prenatal aneuploidy diagnostics in prenatal care will further decrease the use of invasive procedures and therefore reduce the number of diagnostic associated pregnancy loss.**

Keywords:
non-invasive prenatal diagnosis, (NIPD)
massively parallel sequencing, (MPS)
aneuploidy, cell-free fetal DNA