Topic: Non-invasive Prenatal Testing

Title: Implementation of Non-Invasive Prenatal Testing (NIPT) for fetal aneuploidy in a general screening population in a Spanish center

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Objectives:

To describe the clinical experience of one European Center following implementation of a single-nucleotide polymorphism-based (SNP) Non-Invasive Prenatal Test (NIPT) for fetal aneuploidy screening.

Methods:

A general pregnant population presented for NIPT to Echevarne Laboratory (Barcelona), 14.353 maternal blood samples were sent to the CLIA-certified Laboratory at Natera, Inc. (San Carlos, US). Samples were analyzed for risk of Trisomy 21, Trisomy 18, Trisomy 13 and Monosomy X, and also for Sexual Aneuploidy, Fetal Sex, and Triploidy.

Clinical outcome information (karyotype) was collected on samples receiving a high-risk result for any of the tested indications.

The average maternal age was 36.55 ± 3.8 years and the average gestational age was 13.51 ± 2.7 weeks. The average fetal fraction was 9.87% ± 3.9.

The average turn-around-time after sample reception at the testing laboratory was 4 business days.

NIPT results showing a high-risk for fetal aneuploidy were 252 cases (1.76 %). Specifically: 192 cases of trisomy 21, 25 of trisomy 18, 19 of trisomy 13 and 16 of monosomy X.

The sexual aneuploidy cases were 18 (0.13%), in particular: 7 cases of XXX, 8 of XXY and 3 of XYY. Fetal Sex showed 100% accuracy and 13308 cases of low-risk were reported.

Follow-up information on high-risk results was received from 100% (252/252) of high-risk patients. Follow up supports that this is an accurate screening test.

The results show a PPV Combined of 98.01% (247/252) for T21, T18, T13 and MX and an NPV combined of 99.97% (13305/13308).

Conclusion:

These results support that clinical implementation of NIPT offers an early, safe, and accurate option for the general pregnant population.