

NATIinFVG: NIPT FOR TRACEABLE AND COMPUTERIZED ANEUPLOIDIES IN FRIULI VENEZIA GIULIA

PROJECT OVERVIEW

Final Event, Trieste 17th October 2019

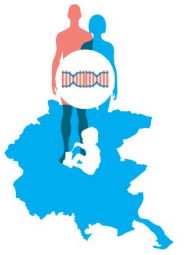
Speaker: Dott. Dino Paladin (AB ANALITICA SRL)



POR FESR
2014 2020
Friuli Venezia Giulia

OPPORTUNITÀ PER UNA CRESCITA SOSTENIBILE





- PROJECT SUMMARY DATA
- NIPT
- PROJECT OBJECTIVES AND RESULTS



PROJECT SUMMARY DATA



PROJECT PARTNERS



MANAGEMENT BODIES OF TECHNOLOGY DISTRICTS AND REGIONAL SCIENCE AND TECHNOLOGY PARKS



END USERS

R&D Infrastructure
Project Coordination)

Dissemination





TIMELINE



FINANCIALS

Eligible costs : €1.393.757,38

Support granted: € 1.085.686,99

NIPT IN A NUTSHELL



- Enabled by new sequencing technologies and discovery of free circulating fetal DNA fragments in maternal plasma
- Ministero della Salute - Consiglio Superiore di Sanità guidelines “Screening prenatale non invasivo basato sul DNA (Non Invasive Prenatal Testing - NIPT)”, May 2015: first or second choice test for the monitoring of the major autosomal aneuploidies
- Many IVD (In-vitro diagnostics) devices and LDTs (Laboratory Developed Tests) already exist
- € 40 millions/year Italian prenatal diagnostic market, 50.000 users/year

NIPT

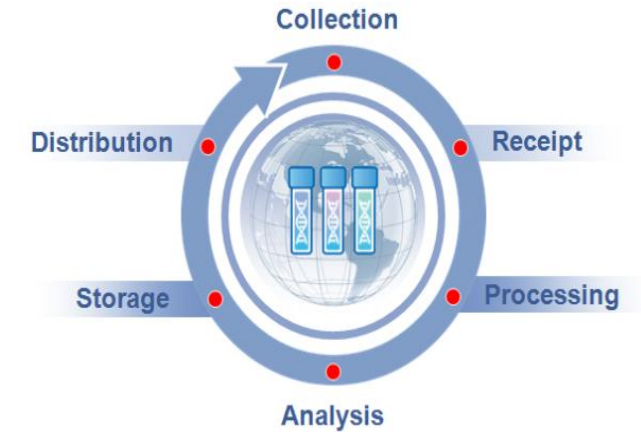
Non invasive
Prenatal Test



NIPT HOTSPOTS



- A screening method for fetal chromosomal abnormalities. Not a diagnostic method
- Critical clinical target due to user sensitivity, and legal and reputation implications



NIPT tests should be:

- integrated into a path that involves pre- and post-test counseling
- carried out in laboratories equipped with specialized staff in NGS techniques
- supported by up-to-date IT tools and equipped with sample monitoring systems optimized for the process.



OBJECTIVE

Full traceability of test execution process, samples, and reference materials for error reduction and correction



RESULT

A laboratory traceability system based on a software for laboratory workflow tracking and sample identification through barcode reading solutions, or alternatively based on full laboratory automation



OBJECTIVE

Improvement of test validation and quality control through robust reference standard materials



RESULT

A **biobank** populated with recognized and accredited **reference standards** suitable for test validation



OBJECTIVE

Reduction of False Negative results due to low Fetal Fraction



RESULT

A rapid and economic test to verify the percentage of the Fetal Fraction in the sample before the NIPT test



OBJECTIVE

Wider coverage of
aneuploidies and chromosomal
micro-rearrangements



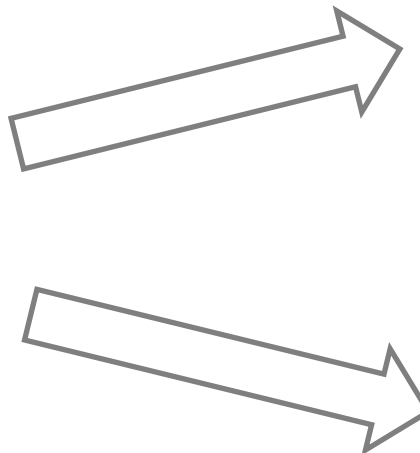
RESULT

Cross validated methods for
detection of aneuploidies and some
chromosomal micro-rearrangements



OBJECTIVE

Integration of the various stages of test management and execution in public healthcare systems, from initial counseling to final medical report discussion through all the stages of test execution



RESULT

Prediction algorithms and their integrated use through a **Decision Support System** (DSS) for operators and end users

RESULT

Data and whole process management in the **hospital electronic medical record** system



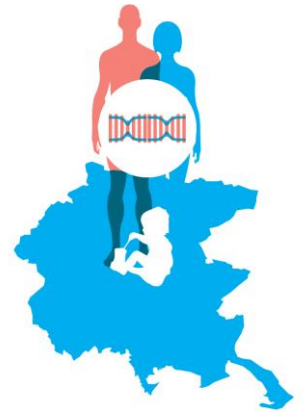
OBJECTIVE

Supporting an **appropriate, effective and informed use** of the NIPT test through learning and information paths targeted to all the involved stakeholders



RESULT

Dedicated **training platform** that can be adapted to different targets



*Thank you
for your attention!*



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