# NIPT in contingent screening protocol.

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**Objectives:** To present results of observational study on NIPT contingent screening program in centre of prenatal diagnosis Gennet.

**Methods:** 11378 low risk pregnancies between November 2020 and April 2023 undergoing the combined first trimester test (FTS). Pregnancies with FTS high risk (>1/100) and/or structural defects were offered core diagnostic procedure (CDP) consisting of quantitative fluorescence PCR (QFPCR) and chromosomal microarray (CMA) in QFPCR-negative cases. Prenatal whole exome sequencing (pES) was performed in selected cases. Pregnancies with middle FTS risk 1/100-1/500 were offered NIPT. The third contingent was formed by pregnancies with positive second trimester anomaly scan (STS). The uptake of recommended procedures and the total diagnostic yield (aneuploidies, copy number and gene variants) were evaluated.

**Results:** From 433/11378 (3.8 %) high FTS risk pregnancies 71.6 % accepted CDP with diagnostic yield 28.7 %. All CDP results with NT <3mm were NIPT detectable. From 926/11378 (8.1 %) middle risk pregnancies 70.8 % accepted NIPT with yield 0.3 % (PPV 25 %). From 7328/11378 (64.4 %) pregnancies undergoing STS, 105 (1.43%) had suspect findings. 87 % from them accepted diagnostic procedures with yield 11 %. Altogether 653/11378 (5.75 %) CDPs have been performed with yield 15.9 %. 204/653 (31.2 %) CDPs were indicated by maternal anxiety with diagnostic yield 0.5 %, all NIPT detectable. CNVs represented 5 % and gene variants 7 % incremental yield.

**Conclusion:** Our results support NT=>3 mm as an indication for CDP and the potential of pES as a substitute for CMA. Despite the fact that about 50 % of CDPs could be replaced by NIPT in about 3 % of pregnancies with ultrasound findings CDP should be considered anyway.