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European non-invasive trisomy evaluation (EU-NITE) study: a multicenter prospective cohort study for non-invasive fetal trisomy 21 testing.

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Abstract

OBJECTIVE: To evaluate the performance of a directed non-invasive prenatal testing method of cell-free DNA analysis for fetal trisomy 21 (T21) by shipping the whole blood samples from Europe to a laboratory in the USA.

METHODS: A European multicenter prospective, consecutive cohort study was performed enrolling pregnant women from Sweden and the Netherlands. Blood samples were drawn just prior to a planned of invasive diagnostic procedure in a population at increased risk for fetal T21 and then shipped to the USA without any blood processing. Chromosome-selective sequencing was carried out on chromosome 21 with reporting high risk or low risk of T21. Karyotyping or rapid aneuploidy detection was used as the clinical reference standard.

RESULTS: Of the 520 eligible study subjects, a T21 test result was obtained in 504/520 (96.9%). Risk assessment was accurate in 503/504 subjects (99.8%). There was one false negative result for T21 (sensitivity 17/18, 94.4%, and specificity 100%).

CONCLUSION: This is the first prospective European multicenter study showing that non-invasive prenatal testing using directed sequencing of cell-free DNA applied to blood samples shipped across the Atlantic Ocean, is highly accurate for assessing risk of fetal T21.