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Non Invasive Prenatal Test

NACE®

NON-INVASIVE
ANALYSIS FOR
CHROMOSOMAL
EXAMINATION

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PIONEERS IN REPRODUCTIVE GENETICS

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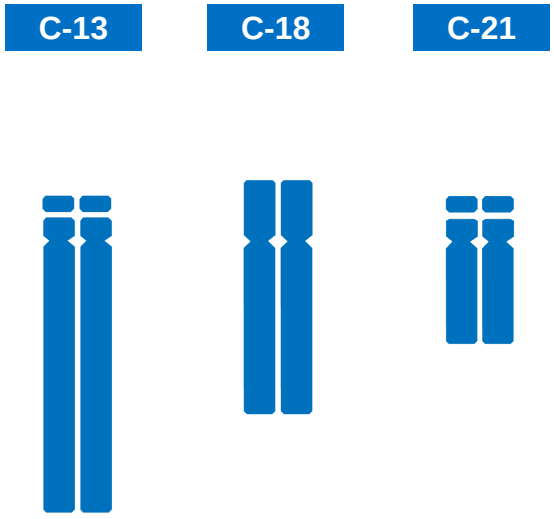
TEST RESULT

Patient Information		Sample Information		Clinic Information
MRN/external ref.	XYZ001	**Sample type**	Blood	**Centre**
Patient name		**Date of draw**		
Date of birth		**Report date**		
Gender	Female	**Clinical indication**	Advanced maternal age	**Referral physician**
		Gestational age	14 weeks	
		Gestation type	Singleton	

ANEUPLOIDY RESULTS

Chromosome	Clinical Interpretation
13	Normal
18	Normal
21	Normal



Normal Test Result

The result obtained in this non-invasive genetic screening suggest, with a precision higher than 99% (see tables below), a chromosomally NORMAL pregnancy for the analyzed chromosomes.

Fetal fraction is estimated to be 6%. Fetal fraction is one component of the analysis algorithm and is combined with other quality metrics to determine the aneuploidy screening result. The fetal fraction estimate is not used in isolation to exclude samples.

CHROMOSOME MAP INTERPRETATION

(The chromosome map is not a test result. Only intended to help test result interpretation)

A normal result consists of two copies of each chromosome, and two copies of the sex chromosomes. All those situations in which more than two copies or less than two copies of each chromosome are present, represent an altered result. Monosomy of any chromosome is defined as the presence of a single copy of the affected chromosome (in the graph, if a monosomy is present, a chromosome appears colorless and with a dashed border). Trisomy of any chromosome means that there is an extra copy of the affected chromosome, i.e., three copies of that chromosome (in the graph, an extra red chromosome appears).

METHODS

For the realization of this screening test has been performed: extraction of blood plasma, isolation of free circulating DNA in blood, DNA sequencing using massively parallel sequencing of new generation, and bioinformatic analysis of the data using the technology platform of Illumina, Inc.

COMMENTS

These results correspond to a high precision genetic screening test, with high specificity and sensitivity. Nevertheless, a normal result should be in line with clinical correlation based on ultrasound findings and other analytical screening.

LIMITATIONS

This test has been designed and validated to detect aneuploidies for chromosomes 21, 18, 13. The test has been validated for singleton and twin pregnancies with gestational age of at least 10 weeks as estimated by last menstrual period. When an aneuploidy is detected in a twin pregnancy, the genetic status of each individual fetus cannot be determined. A negative test result does not preclude the absence of chromosomal abnormalities such as trisomy 21, trisomy 18 and trisomy 13. There is a small possibility that the test results might not reflect the chromosomes of the fetus, but may reflect chromosomal changes of the placenta (confined placental mosaicism) or the mother (chromosomal mosaicism). These results do not eliminate the possibility that this pregnancy may be associated with other chromosomal or subchromosomal abnormalities, birth defects, and other clinical conditions. This test is not intended to identify pregnancies at risk for open neural tube defects. The accuracy of these tests may be compromised if there is (i) maternal chromosomal aneuploidy; (ii) mosaicism (fetal or confined to the placenta); (iv) analysis performed in week of pregnancy under week 10. There is a slight chance that the test results may not reflect the chromosome status of the fetus (false positives), reflecting instead subchromosomal changes in the placenta or the mother; (iii) allogeneic blood transfusion, transplant or stem cell therapy; (iv) vanishing twin syndrome; (v) multiple gestation.

	Specifity	False Positive	Sensitivity	False Negative
Trisomy 21	**99,8%**	0,2%	>99,9%	<0,1%
Trisomy 18	**99,6%**	0,4%	97,4%	2,6%
Trisomy 13	**>99,9%**	<0,1%	87,5%	12,5%

EXEMPTION CLAUSE OF DIAGNOSTIC LIABILITY

The genetic diagnosis services carried out by %1 are exclusively intended to qualified health professionals. The result obtained by this test and the information that could be derived from it, cannot be considered in any case as substitute of genetic counselling or medical treatment by a trained professional neither represent itself a medical enquiry.

Any result should be interpreted in the context of all available clinical findings, within the general context of a medical enquiry, which must be conducted by genetic diagnosis and / or clinical trained professionals. IGENOMIX, S,L is not responsible for the use made by the contracting party of their services, neither the obtained results by means of their study analysis, nor the harmful temporary consequences diverted by its use, making specific discretion of taking appropriate legal measures assuming an improper use of those mentioned studies and analysis.

Sample name, PhD

Biologist

Sample name, PhD

Laboratory Manager