ILLUMI SCREEN

CATGGOCCTNCC
TGTIAACPTTRAAFGG
ACCMAAGGTTTGG
AAACCCCTTT

Illumiscreen Non-Invasive Prenatal Screening – Important Information

LIMITATIONS OF THE TEST

This test is designed to screen for chromosome aneuploidies and is validated for chromosomes 21, 18, 13 and for fetal gender. The test is validated for singleton and twin pregnancies with gestational age of at least 10 weeks 0 days, as estimated by last menstrual period, crown rump length, or other appropriate method (equivalent to 8 weeks fetal age as determined by date of conception).

This is a screening test with a risk of false positive or false negative results. A negative test result does not eliminate the possibility of chromosomal abnormalities such as trisomy 21, trisomy 18, trisomy 13, Monosomy X, XXX, XXY and XYY. For confirmatory diagnosis, invasive testing such as chorionic venous sampling and amniocentesis is required.

This test does not cover other genetic conditions including but not limited to chromosomal abnormalities other than the chromosomes stated above, subchromosomal abnormalities, triploidy, single gene disorders, birth defects such as open neural tube defects or other congenital conditions.

When an aneuploidy detected result is reported in a twin pregnancy, the status of each individual fetus cannot be determined. Although the presence or absence of Y chromosome material can be reported in a twin pregnancy, the occurrence of sex chromosome aneuploidies such as MX, XXX, XXY and XYY cannot be evaluated in twin pregnancies.

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 of the mother (chromosomal mosaicism).

TEST PERFORMANCE

	Sensitivity	Specificity
T21 Down Syndrome	99.14%	99.94%
T18 Edward Syndrome	98.31%	99.90%
T13 Patau Syndrome	98.15%	99.95%
XX Female	97.6%	99.2%
XY Male	99.1%	98.9%

TEST METHOD

IllumiScreen is performed using massively parallel sequencing at Labtests Auckland. The whole genome sequencing is performed using circulating cell-free DNA extracted from maternal plasma to determine the copy number of chromosome 13, 18, 21 and sex chromosomes. DNA libraries are prepared using Illumina TruSeq Nano DNA Library Prep kits and next generation sequencing is performed in NextSeq 500. The data is analysed by the proprietary algorithm SAFeR™.